INNOVCare

Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions

www.innovcare.eu

Training Curricula

Case Manager for Rare Diseases

65%

of people living with a rare disease have to visit different health, social and local support services in a short space of time*

67%

of people living with a rare disease and carers say that different services communicate badly between them*

30%

of carers for people living with a rare disease spend more than 6h per day on health-related tasks*

38%

of patients and carers were absent from work due to health-related issues for over 30 days in the last 12 months*

*This survey was conducted via EURORDIS survey initiative, RareBarometer Voices, and in the scope of the INNOVCare project.

www.creenfermedadesraras.es | www.apwromania.ro | www.cjsj.ro | www.eurordis.org | www.finovatis.com | www.ier.si | www.ki.se | www.zsi.at

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1. Introduction

1.1. Project presentation

INNOVCare - Innovative Patient-Centered Approach for Social Care Provision to Complex Conditions is a project co-funded by the European Commission in the Call for Projects "Innovation in social policy to support social reforms" of the Program "Employment and Social Innovation ″EASI" - 2014-2020, PROGRESS component″.

The project addresses the social challenges faced by people with rare diseases and the gaps in the coordination of medical, social and support services in the European Union (EU) Member States (MS). In most MS, care pathways are not structured and patients have problems accessing the services they need.

The recommendation of the European Commission's Rare Disease (RD) Expert Group (2016) on supporting the inclusion of rare diseases in social services and policies underlines that Member States "will promote measures to facilitate multidisciplinary, holistic, continuous, participatory and person-centered care for people with rare diseases, supporting them to fully accomplish their fundamental rights."

The project develops and tests a holistic, personalized care path that aims to strengthen the medical, social and educational service partnerships between public, private service providers and the civil society.

The objectives of the project:

- Assessing the unsatisfied social needs of the persons with RD and of their families;
- Analysis of care patterns in the Member States;
- Exchange of experience/good practices between resource centers;
- Proposal of an optimized care path model:
  - Implementation and assessment within a pilot project in Romania - NoRo Center;
  - Assessment of the social, economic and cost-efficiency impact of the care pattern;
  - Analysis of the possibility of transposing the pattern for other diseases in other Member States;

- Strengthening the partnership between service providers from different sectors – public sector, private sector, civil society;

The pattern is being implemented in a pilot study in Romania at the **NoRo Pilot Reference Center for Rare Diseases (NoRo Center) of the Romanian Prader Willi Association (RPWA/APWR)**, and the socio-economic impact and the cost-benefit ratio will be **evaluated by the expert teams** from other Member States. The implementation of this pattern will optimize the care, will increase the efficiency of the authorities and will ensure the increase of the quality of the services provided by the NoRo Center.

The NoRo Center already has the following social and medical services:

1. **Day care center for recovery of children with rare diseases and from the autistic spectrum** - individualized intervention for children with rare diseases and autistic spectrum disorders;
2. **Recovery and rehabilitation center –** Groups of patients with rare diseases in 5-day residential system;
3. **"Trial for Flight" Independent Life Training Center -** a program for young people with special needs to develop independent life skills;
4. **NoRo Help Line -** Information and Counseling Service;
5. **Special ambulatory -** pediatric psychiatry, medical genetics, pediatrics.

Since the beginning of its work, the NoRo Center has highlighted the interdisciplinary nature of addressing people with rare diseases, this being the only possibility for effective intervention to increase the quality of life of the people with rare diseases. The interdisciplinarity and collaboration with other community services is of major importance in terms of service continuity.

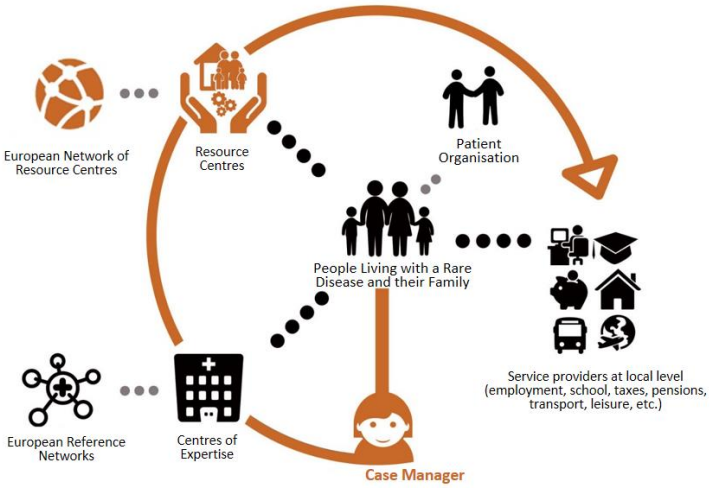
The importance of the INNOVCare project from the point of view of the NoRo Center lies precisely in the fact that through the activities carried out within the project, we will have scientific proof of the necessity of this approach and of the effectiveness of coordinated interdisciplinary intervention.

- Among the **essential needs** of the patient and of his/her family there is also the need of **coordinating the care services**: access to medical, social and educational services.

In Romania there is the legislation on the inclusion of the case management in social and child protection services according to O.G. no. 288/2006 from 06/07/2006, published in the Official Gazette, Part I no. 637 from 24/07/2006 for the approval of the Minimum Mandatory Standards for case management in the field of the protection of children's rights. There has been a case management project for patients with Duchene Dystrophy implemented by the Parent Project - ANBRaRo member organization.

Brief information about the Pilot:

- *Time frame*: 18 months total, starting from 03/2017 to 08/2018; 9 months for each patient/family
- *Target population*:
  - Patients with rare/complex conditions (children and adults) and their families;
  - Current beneficiaries of NoRo and new ones;
  - 120 cases*;* each case includes patient + family;
  - Each case has access to the service for 9 months;



- Geographical scope: region;
- Number of case managers employed: 2 FTE, 4 case managers;
- Profile of case managers: social workers, legal advisor, special education teacher;
- Nr. of simultaneous "cases" per FTE case manager: 30;
- Service provision should be focused on a few key areas in order to have more meaningful results.

Case management **roles and definition** might be slightly different for each country based on local realities and needs.

Help lines can be a tool of case management, but it is not necessary. In help line, without personal contact, you provide **advices** but, in case management you **connect the patient and the family with the services they need**. In case management, you have a better evaluation of needs, not only the person's need, but also the needs of the whole family.

Another tool for case management is **networking**, using networks of services, professionals. Besides helping the beneficiaries, it can also be a support system for the case managers – for information from all the fields involved.

Can be trained as case manager a person with any kind of education in a profession that is involved in the management of the rare diseases (at least bachelor's degree – although some said it should be higher education. We kept "at least bachelor's degree" as in some countries, the nurses have no higher education, but represent good candidates for case managers). In a team of case managers, it is important to have different professions, to have more complementary information and experiences. It is an added value if the case managers have a personal experience – themselves or through a family member with a rare disease.

As **case management** involves empowerment too, it should be a case by case decision if something can be done by the patient himself (or by the family) or it has to be a direct intervention of the case manager. Case managers can have fewer interventions for one case in the periods they've been empowered enough, but they have to be able to intensify the collaboration with the case manager when they have changes, transition periods.

For **long term sustainability** is proposed to have in every region a small team of case managers, to create the regional networks. County councils should be convinced to promote our experience and create 2-3 people teams in each county. These case managers should be trained to be able to create their own networks in the counties.

Training courses for professionals are important for the network members. Accredited courses should be used. Different professional societies should agree with the curricula for rare disease management. A good starting point is the curricula used by ProRaris. Online trainings should be considered.

In local professionals' job description should be included responsibilities regarding collaboration with the case managers and other responsibilities regarding rare diseases.

Training, empowering brothers and other **family members** is important in order to improve the resilience capacity of the families.

Case manager's **experience can be used** in asking to create new/more services if they are not available in the region.

Virtual case management should be considered, especially for those who use regularly technology and/or have a busy daily schedule. It should be a case by case decision if this kind of communication fits the family or not.

There should be a **system of protecting the case managers**: intervision, supervision, raising resilience.

Case manager – the **wording** was discussed. "Case" is not human enough. Proposals like "care coordinator for rare diseases" or "care facilitator" might be discussed in future development of this profession.

1.2. Case study - case management services for rare diseases

1.2.1. "Navigators" project, Rare Diseases Denmark

Rare Diseases Denmark *implemented a project granted by the Danish Health Authority to educate and employ Volunteer Rare Navigators. The goal was to equalize disparities and promote health and health literacy in the most vulnerable PLWRDs in the Danish healthcare system.*

Navigators draw on personal and professional skills

A Navigator is a person that based on experience-based knowledge provides personal guidance to vulnerable PLWRD navigating the Danish health care system and accessing the social system.

Navigators are either suffering from a rare disease themselves or a relative of a PLWRD.

The task

Providing coordination between and within the systems is a task for social- and health care professionals. However, Rare Diseases Denmark is prepared to contribute to improve the conditions for PLWRD through voluntary action and participate in the important effort to support PLWRDs in the process of:

- *Acknowledging personal situation*
- *Creating an overview of options*
- *Encouraging health literacy and support compliance*

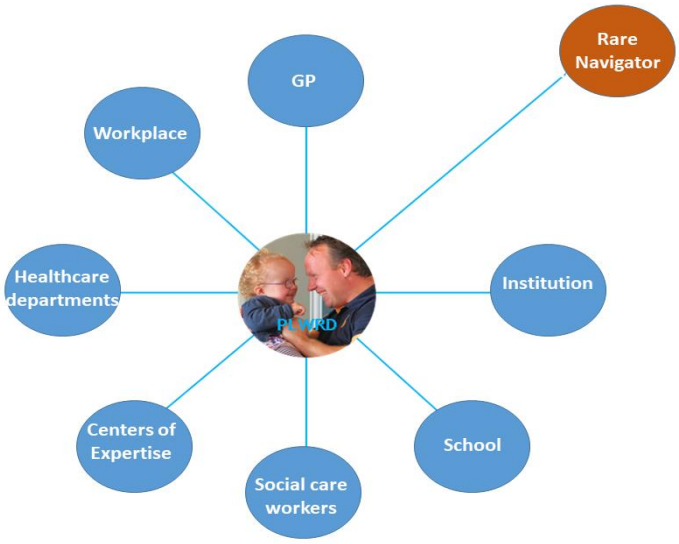
Navigators support and empower the wishes, priorities and decisions of vulnerable PLWRD through the principle of helping them to help themselves. Rare Navigators can help ensure that patients don't fall through the cracks so they can follow their treatment plans and improve their health literacy. In addition to the personal knowledge Navigators, Rare Diseases Denmark provides an education and continuously support program for the Navigators.

Evaluation

To study the impact of receiving support from a Navigator, all the PLWRDs involved in the project, answer the WHO-Five Well-being Index questionnaire in the beginning and in the end of the navigation.

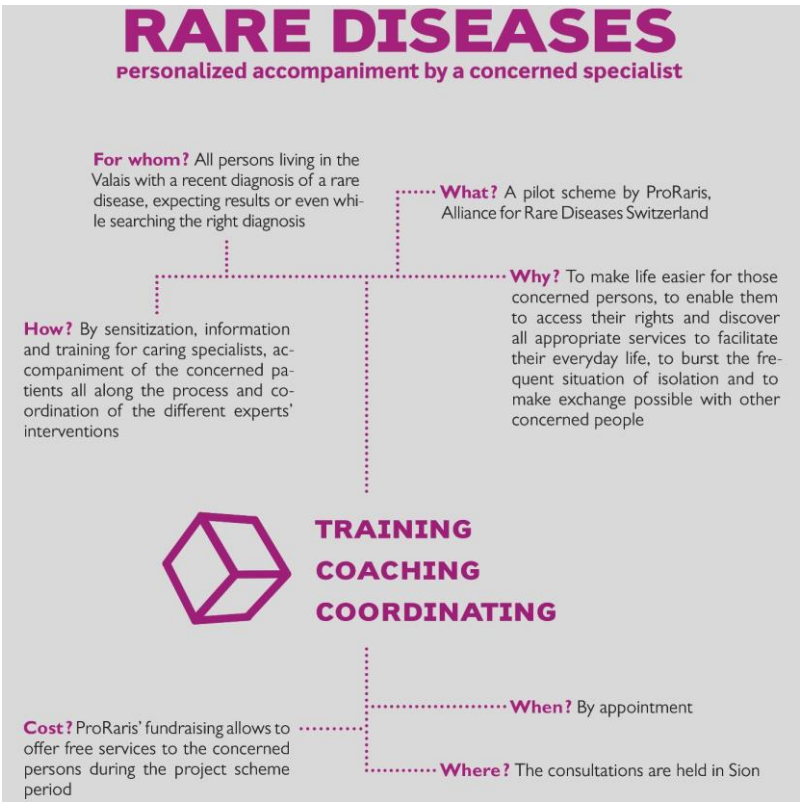
Facts

Originally the Patient Navigation concept was developed in the US to improve outcome in vulnerable populations by eliminating barriers to timely diagnosis and treatment of cancer and other chronic diseases. Rare Diseases Denmark has made it probable that PLWRD´s is particularly vulnerable in the health- care system because of the complexity and rareness of the illness. Standard procedures do not work for these patients and they need help navigating the systems.



Navigators play a role, in the lives of the PLWRD, for maximum 12 months. The navigation depends entirely on the individual's needs and the complexity.

1.2.2. ProRaris – a pilot scheeme of the Alliance for Rare Diseases Switzerland



1.2.3. Evaluation and management file for the pacient with neuromuscular pathology applied by the Parent project - Romania

Working tools: Evaluation and management file for the pacient with neuromuscular pathology applied by **Parent project** within the implementation of a case management project to ensure an organized, rigorous, effective and coherent multidisciplinary and interinstitutional intervention for the child and his/her family.

The basic situation	The results of the Parent Project
• **10 medical chapters** on assessments, treatments, recommendations • A chapter of formal and informal education • The situation of the beneficiaries: ✓ Attends School/access/counseling teachers/colleagues ✓ Home Education/Support Teacher • Benefits: Allowances, Disability degree, rights and facilities (Subscription, Rovinieta, CFR Tickets) • Socialization and leisure - activities with the family and with the group	▪ 10 children and young people from Dolj County and neighboring areas benefited from the multidisciplinary management for DMD/DMB ▪ A team of specialists and volunteers with Training in applying the international standards for DMD/DMB care was formed ▪ 9 families have received training and counseling for patient care ▪ A training seminar was organized for 20 care people, 11 volunteers, 10 specialists, ▪ Two jobs were created; 8 specialists from the medical, social field were involved; 8 Voluntary Contracts were signed ▪ Partnerships have been signed with 3 County institutions

1.3. Case management

The second EU Health Program for 2008-2013, implemented by the Executive Agency for Consumers, Health and Food (Chafea), promoted the specific health promotion, the early prevention and diagnosis of rare diseases, as well as the approach of the inequalities related to health. By setting up and strengthening pilot referral networks, EU projects have helped to develop the efficiency, share good practice, set common standards, and promote evidence-based interventions.

As the provision of integrated services can improve the management of chronic conditions, the health program can help identify and classify the best ways to care for patients. The aid for chronic patients for an active and productive life presents a significant social and economic value. The EU projects analyze the ways to keep people in the workforce longer and to facilitate the return to work of those who want to resume their activities after diagnosis. At the end of last year the first European Reference Networks were

approved. The NoRo Center is part of the RO-NMCA ID network at national level and of the ITHACA Network at European level. 24 applications, 370 hospitals and nearly 1,000 healthcare providers from 26 countries (25 Member States + Norway) were submitted.

Europe's response to the complex challenges of the chronic (implicit and rare) diseases has to be integrated into several sectors and policy areas. The EU health programs will reflect the needs identified in the reflection process on chronic diseases, giving priority to promoting prevention and health and to managing diseases in the spirit of empowering the patients. This is an important step towards collaborative sustainability, sharing of expertise, shortening time from diagnosis to treatment, and holistic and integrated care for rare diseases.

Description of **case management services provided by the NoRo Center** within the INNOVCare project:

a) designating a case manager for the beneficiary;

b) the case manager's task is supporting the beneficiary and his/her family in identifying the needs, elaborating the action plan and implementing it;

c) at least 5 individual interactions and at least 2 group activities organized and coordinated by the case manager;

d) intervention with a total duration of 9 months, during which 2 questionnaires will be applied - 1 at the beginning of the intervention period and 1 at the end of the intervention period. A third questionnaire will be applied 9 months before or 9 months after the intervention;

e) the case manager guides the beneficiary to the services he/she needs, establishes contacts with these services, provides information about services, rights, advises and instructs the beneficiary and the family to better manage the challenges caused by the disease, develops together with the beneficiaries a kit for the communication with specialists;

f) the services being provided within the INNOVCare project, in the framework of research on the efficiency of case management services and of holistic approach for people with rare/chronic diseases, the beneficiaries accept the provision of the services under this contract, and they also accept the provision of data (to be transmitted anonymously to the project partners) for this study.

Vision: The case managers are recognized as experts and key actors in rare disease co-ordination teams for access to quality and efficient care and for empowering the patient and his/her family.

Definition: The case management is a participatory process of collaboration, evaluation, planning, counseling and information on care options that meet the conditions for fulfilling the needs of the patients and of their families by communicating and accessing the resources.

The role of the case manager from the patients' perspective:

- to have someone to ask for advice when they need it;
- information and finding experts;
- information on treatment funding and access to care, treatment and education;
- identification of jobs to ensure their independence;

The skills and the qualifications of the case manager:

The case managers need to be able to listen, observe and respond. For this they need specific skills: skills to wait, to specify, to confront, to personalize, to solve problems and to plan actions.

The required **skills** for a case manager: positive approach; effective communication; negotiation skills; knowledge of the functioning of the medical, social and educational system and of the rights of patients with rare diseases, of the basic notions in accessing services and benefits for people with disabilities, the importance of obtaining informed consent, confidentiality of data and the privacy/autonomy of the beneficiaries; ability to perform patient and family needs assessment; critical thinking and analysis; the ability to plan and organize the activities necessary to achieve the objectives.

Appropriate **qualifications** for case managers: social worker, nurse, psychologist, special education teacher, physical therapist, physician, legal adviser, genetic counselor, pedagogue, etc.

The family members of patients with rare diseases and the patients can acquire case management skills and can become important community resource, with an appropriate training.

Case management objectives:

1. The holistic approach to the medical, psychosocial, behavioral and spiritual needs of patients and their families;
2. Intensification of patient involvement in decision-making;
3. Extension of the interdisciplinary team of case managers to include community support persons;

4. Collaboration with service providers and facilitation of patient communication with service providers;
5. Improvement of patient safety.

The basic premise of case management is that when a person reaches the optimal level of well-being and functional capacity, everyone benefits: patients because they have an improved quality of life, support services, health care systems, health insurance systems. Case management serves as a means of achieving the well-being of the beneficiaries and their autonomy through advocacy, communication, education, identification of service resources and facilitation of access to services.

These changes will determine the credibility of the case management and will complement the current trends and changes in health care.

2. Rare diseases- General information

A rare disease is the disease that affects less than **5 people out of 10.000**. They are also called orphan diseases because they still largely lack treatment, recognition and proper care.

There are over 8.000 different diseases in the world, so they are very numerous, complex, little known by the population, the medical staff and the health system officials.

They can manifest themselves from birth or from early childhood. In over 50% of the rare diseases, the first clinical signs occur in adulthood and are generally accompanied by motor and/or sensory difficulties, which are serious and cause a major handicap.

Considered unprofitable by the pharmaceutical laboratories, they have little or no research, they have little or no treatment, and their diagnosis can last for several years. The economic and social support for these diseases remains defective.

80% of the rare diseases are genetic. They are generally hereditary and are passed on from one generation to the next. They can also be the result of a spontaneous mutation, without a similar case in the family.

Among the other 20% of the diseases are included rare cancers, autoimmune diseases, toxic and infectious diseases and others of various causes (environmental factors, etc.).

In Romania, the frequency of the rare diseases is 6-8%. As in the rest of the European countries, these diseases affect in our country about 1.300.000 people, of which approximately 1.250.000 patients do not yet have the correct or complete diagnosis, nor the appropriate treatment or care.

2.1. Rare diseases – General information

The rare diseases identified in Salaj County and their description are presented in ANNEX 1. More information about the rare diseases on www.orpha.net or www.edubolirare.ro.

2.2. Access to diagnosis

The first and most difficult obstacle that patients and their families have to overcome is getting a diagnosis: it is often the most desperate struggle. This struggle is repeated with every new stage of a rare evolutionary or degenerative disease. Lack of knowledge about their rare disease often puts the lives of patients at risk and also involves inappropriate, even harmful medication and treatment prescription.

All those affected by these diseases are faced with similar difficulties in searching for diagnosis, relevant information, and finding qualified specialists. The specific issues are also highlighted by the access to quality medical care, global medical and social support, effective connection between hospital and family doctor, but also by professional and social integration, such as independence. Those affected by rare diseases are more vulnerable psychologically, socially, economically and culturally. These difficulties can be overcome by appropriate policies.

Due to the medical and scientific knowledge, many patients are not diagnosed. Their illness remains unidentified. They are the people who suffer the most because of the difficulties in receiving adequate support. (Orphanet)

In the absence of the correct diagnosis, the medical services are unable to adequately treat the patient. All abnormal behavior associated with numerous rare diseases is often reproached to the mother, causing guilt and anxiety. Misunderstanding, depression, isolation, and anxiety are daily manifestations of most of the parents of children with rare diseases, especially in the pre-diagnosis phase.

The whole family will withdraw, will marginalize. The vulnerability is psychological, social, cultural and economic. In many cases the birth of a child with a rare illness leads to the separation of the parents.

The communication of the diagnosis is a moment with a strong emotional load, ″officially″ marking the change in the life of the patient and of his/her family. The acceptance of the diagnosis and the normalization of the negative emotional states are conditioned by the way this diagnosis is announced.

The minimum conditions to be met when announcing the diagnosis have been already established in 1993 by the World Health Organization.

Accepting the diagnosis and adapting to the disease is the first major obstacle the patient and his/her family will have to overcome. Among the **reactions that may occur** after finding out the diagnosis of rare disease, both in the patient and his/her family/guardian can be listed:

- *Mourning reaction* – The diagnosis can create a feeling of loss, loss of control over life
- *Denial* - The patient and his/her family can ignore the disease, hoping it will disappear after a while, hoping that the doctors were wrong. That is why it is very important to announce the diagnosis together with relevant information about the disease or in an open discussion.
- *Anger* - It can be directed against everyone around, the cause being the misunderstanding of why he/she has the disease and the others not. Parents can also feel rage against their own person, against the child, the partner, or even the medical staff.
- *Depression* - Sadness, fatigue, lack of energy and hope can occur in patients with rare diseases. Parents can also live the same negative feelings and build negative scenarios about the future of the sick person.
- *Fear, anxiety* - Fear is related to the different symptoms, to the reactions of others to the diagnosis, to the evolution of the disease or to other changes that the disease implies. This feeling can occur both in patients and in their parents, who may feel overwhelmed by the situation.
- *Blame* - It often occurs because of the erroneous belief. The patient or his/her family can believe that the disease is the result of their inappropriate behavior, that it is a punishment for their own sins or others.

The negative feelings, sadness, fear, confusion and other like are normal right after receiving the diagnosis. Their persistence for a long time or their high intensity requires special psychological involvement.

The needs of the patients and of their families are diverse and complex, both before and immediately after diagnosis. Interdisciplinary teams are needed to address these cases, to counsel and guide the family through the social and medical system in our country. The referral to services should be done by the case managers involving patient associations within support groups.

2.3. Determination of the disability degree – Social Services

Since most rare diseases involve a disability, this must be accepted by the person concerned and recognized by obtaining a degree of disability certificate.

Often, however, family members feel that asking for determination of the disability degree, stigmatize the patient even more and accept the situation without trying to look for other "treatments".

Below are the steps that the person with disabilities must follow in order to obtain a disability degree certificate in Romania:

- Submission of the file;
- Complex assessment;
- Submission of the file to the commission in order to receive a disability degree.

Case managers has to know all the legal and procedural details of obtaining a disability degree certificate.

Even though in the legislation in force things seem fairly simple to achieve, in reality, due to lack of staff, the family counseling on access to social, medical, educational services is completely missing. The recovery plans are completed with very little information. Families find out, from other families, where to go, what kind of services they can access in the community or elsewhere.

The determination of the disability degree of a person with disabilities produced by rare diseases is a family option, but specialists need to raise awareness of family members, that the possession of a disability degree certificate should not be seen as a burden. This means above all "facilities" that can lead to better management of financial and material issues, access to free social and medical services, etc.

Starting from the reality that the majority of the rare diseases do not yet have treatment, it is imperative to create and adapt the existing social services. Case managers has to know and involve in their network all the social service providers that can be involved in the management of their clients.

Patients also have problems accessing their rights and accessing information. There are problems in assessing the degree of disability, the evaluation not being uniform and the coding being incomplete. One solution could be the use by the evaluation committee of the people with disabilities of the Orpha Codes specific for each rare disease, as the ICD10 is incomplete from the point of view of the rare diseases.

2.4. Access to education

One fundamental right of any person is the right to equal opportunities in education, the right to be supported in making the most of their own potential. Through education, the person with disabilities produced by rare diseases has the necessary support to acquire certain knowledge, to form certain skills to help him/her integrate into society.

Although the right to education of the persons with disabilities is regulated by law and there are educational alternatives for children with disabilities, the reduced number of the

educational services and their geographical distribution limits their access to education. By far the most disadvantaged are the children with disabilities who live in rural areas where there are no schools for inclusive education or day centers to compensate for the lack of an official form of education. And where these services exist and are accessed, the number of hours of specific therapies and of other support services that the education system is able to provide in a formal and specialized context is limited and fails to meet the needs of the disabled child.

Currently, the educational practice in Romania has focused on including the people with disabilities in educational and normal living environments for the community or as close as possible to ordinary environments. In the spirit of an education for all and in order to increase the quality of life for the people with disabilities, the present concerns of the Romanian school aim at equalizing the chances of education and normalization. Equalizing educational opportunities requires that schooling of children with disabilities to be carried out, as far as possible, within the general education system, and that compulsory education includes children of all types and levels of disability. Ensuring equal opportunities is achievable if the school facilitates access for people with special educational needs to everyday life conditions as close as possible to normal conditions and to some additional services that are in line with the individual needs.

In Romania, according to Law 448/2006, people with disabilities have free and equal access to any form of education, regardless of age, according to their type, degree of disability and according to their educational needs. Persons with disabilities are provided with lifelong learning and lifelong professional training.

The education of persons with disabilities caused by rare diseases is carried out in the educational institutions included in the national education system and coordinated by the Minister of Education, Research and Innovation. There are several educational alternatives for the education of people with disabilities, according to Law no. 448/2006:

- Special education units/school centers for inclusive education;
- Individual integration in mass education units, including units teaching in the languages of the national minorities;
- Groups with special compact classes, integration in mass preschool and school facilities;
- Educational services with itinerant/support teachers:
- Home schooling until graduation, but no later than the age of 26;
- "Hospital" education during hospitalization;
- Educational alternatives.

Until 2012, the SECC was elaborating both the recovery plan and the educational plan, but with the order 6552/December 13, 2011, the educational plan returned in responsibility of the County Center for Resources and Educational Assistance - CJRAE.

According to the legislative order, for the evaluation, psycho-educational assistance, school orientation and for the professional orientation of the children, pupils and of the young people with special educational needs, the school and professional orientation and evaluation service was created, hereinafter referred to as SEOSP, within the resource and educational assistance county centers (CJRAE).

In the given situation and according to the order in force, the children with learning difficulties reach this service only at the request of the family, or after obtaining the disability certificate, if helped by DGASPC.

If the child has a disability certificate, the family will only fill out a simple request that the child be evaluated by SEOSP to obtain a school and professional orientation certificate. However, there are situations where children with learning difficulties are identified by teachers, school counselors and oriented together with their families directly to SEOSP.

Following the SEOSP assessments, the child with CES receives a school orientation certificate accompanied by a Personalized Intervention Plan, which may include the need for a support teacher, an adapted curriculum, or an orientation towards special education and occupational classes.

The implementation problems of this order remain quite a few:

- The responsibility for adapting the curriculum remains in the hands of the teacher from mass-learning, who is not verified by anyone, to what extent he manages to adapt the learning material as well as possible.
- Only starting with this year, within the special education, occupational classes have been set up (textile, tailoring and wood processing).

There are also many situations where the families of the children with learning difficulties cannot accept the situations they face, or for fear of stigma they prefer not to follow the legal procedures for obtaining a certificate of disability or of school orientation. These cases remain very difficult to manage by the teachers who need to work with them. The school councilors employed in the mass schools, in turn, are very few, assigned to several schools, with entirely different responsibilities. Under such conditions, the families of the children with learning difficulties or with special educational needs are not counseled/oriented towards these support services, which, due to lack of staff, are very difficult to carry out.

2.5. Employment

The issue of the professional qualification of the young people with disabilities and their integration into work is raised. In order to counteract the reluctance of the employers to people with disabilities, sheltered workshops can be set up to facilitate the transition to adult life and, implicitly, employee life.

Unemployment among people with disabilities is double that of the other categories of the working population.

In Romania, only 12% of the people with disabilities who have the age and ability to work have a job (compared with a 20% occupancy rate in Poland or 50% in Finland, Denmark, Luxembourg).

Current legislation (Law 448/2006) provides measures to encourage the employment of persons with disabilities by offering fiscal incentives, by setting a 4% quota for firms with more than 50 employees, by employment in protected forms (such as protected units and workshops) and which can also benefit from tax facility.

The social economy contributes in various ways to employment, directly by engaging in its own organizations: associations, cooperatives, foundations and more recently social enterprises and indirectly through the multiple functions it performs.

The social economy organizations contribute indirectly to employment growth by improving the employability of disadvantaged, low-skilled workers. The providers of employment and training services and sometimes the ones offering social services have a

very important role in implementing the active employment measures from the public employment or social inclusion policies.

The most representative type of protected unit belonging to the social economy are the organizations with intense integration activity; the protected units under this model, cooperatives, associations and foundations have a high rate of employment (usually above the average employment rate of 7 people with disabilities) and aim to extend their number as far as possible. These types of protected units have a high level of diversification of activities and are interested in accessing any type of tax facility or source of possible funding from local or central public budgets or any other type of private funding to develop their activities and to provide a relatively low level of worker productivity. In addition to supplying jobs, the social economy organizations with significant integration activity provide employees with a range of complementary services such as qualification courses, psychological or legal advice, recovery services, transport services, or fully or partially fund certain medical interventions for the employees with disabilities; only in these organizations we notice cases of employed persons who subsequently found a job on the labor market outside the protected unit.

The Protected Units are social economy entities known at European level as *Work Integration Social Entreprises* WISE or *Vocational Entreprises*.

The beneficiaries stipulated by Law 448 of 2006 are disabled persons who can be employed according to their professional training and work capacity, certified by the disability degree certificate, issued by the evaluation commissions from each county of from the districts of the city of Bucharest. For the purposes of Law 448 of December 18, 2006 and only in the context of employment, the persons with disability degree III are included in the category of people with disabilities.

The Authorized Protected Unit is the public or private market participant, with self-management, where at least 30% of the total number of employees with individual labor contract are disabled persons.

The protected workshop - is a licensed, protected unit without legal status, with self-management, where disabled people carry out training, development and skills development activities; it can work in community locations, day centers, residential centers and special education units.

A protected workplace is a space adapted to the needs of people with disabilities in the form of protected employment.

There are no additional costs associated with the employment of a person with disabilities. Moreover, the following expenses are deductible from the calculation of taxable profit: expenses for adaptation of the workplace, expenses for the purchase of equipment used in the production process by the disabled person, expenses for the transport of disabled persons from home to workplace, expenses for the transport of raw materials and of finished goods to and from the home of the disabled person employed for work from home. (Law 448/2006, Article 83, paragraph a and b).

Other deductible expenses related to the work integration process of a person with disability are: specific training, employment orientation and employment of disabled people expenses that are deductible from the unemployment insurance budget which the unit pays to the budget of the state. (Law 448/2006, Article 83, paragraph c)

Employers can benefit from state subsidies when hiring a disabled person if the employer is not legally obliged to hire disabled people. It will receive for one year the minimum wage per month, for every employed person with disability, provided it keeps the employee for at least 2 years (Law 76 of January 16, 2002, as amended, Article 85, paragraph 2).

Moreover, employers who hire indefinitely graduates with disabilities are exempted from paying the unemployment insurance and receive monthly, for 1,5 years, the following amounts:

a) 1 minimum gross national salary for the secondary school or arts and craft school graduates;

b) 1,2 minimum gross national salary, for the high school or post-secondary school graduates;

c) 1,5 minimum gross national salary for university graduates. (Law 76 of January 16, 2002, as amended, Article 80)

Physical or legal persons employing disabled people may establish protected units. The protected units may be:

a) economic agents with legal personality, regardless of the form of organization and ownership, which have at least 30% people with disabilities with individual labor contract of the total number of employees

b) sections, workshops or other structures without legal personality within the economic units or within the non-governmental organizations, which have their own accounting and at least 30% people with disabilities with individual labor contract of the total number of employees

c) family associations, associations or non-profit foundations established by at least one person with disabilities, as well as the disabled person who is authorized, according to the law, to carry out independent economic activities, provided that he/she pays the social security contributions stipulated by the law.

2.6. Integration into society

Living with a rare illness brings multiple implications in all aspects of life for both the sick person and the parents and other family members. After diagnosis, there is a difficult period in which the life of the sick person and of his/her family is changing, the system of values is shaken, what seemed to be fundamental until then can become an accessory and complex feelings can arise: fear, denial, shock. Depression, anxiety, refusal of the diagnosis are in fact an unconscious mechanism of adaptation to the suffering caused by the situation of the sick person. It is the phase in which the situation of the sick person and that of his/her family may seem hopeless. But it is not like that, alternatives and options are always there, they must be sought, found and valued.

For this it is important that the sick person and his/her whole family accept the diagnosis, find the inner resources and the energy necessary to cope together easily with everyday life and to know from the beginning the most appropriate ways to adapt the environment to the potential of the sick person. This involves many questions to be answered, such as: "Which family member or other people will spend more time with the sick person? To what extent will family members tolerate and accept the behavior of a person with disabilities? What kind of daily activities can the sick person have and what daily activities will he/she have to forget? What are the space "boundaries" in which they can be safe? Will he/she be able to attend school or work? Where will they find help? Are there any other people with the same disease? Is there treatment for the disease he/she suffers from? How will this person's life be later? How long will he/she live? What will happen to that person after the parents are gone?" and others. So most parents' fears are related to the possibility that, the sick person may never be truly independent.

The specialists involved in the case management must help the parents understand their children or the sick adults, must help them know information related to the diagnosis to be able to become co therapeutists along with the specialists during the activities and interventions in the recovery process. The parents and the whole family must know that they have been and remain the main support in the life of the sick person since receiving the diagnosis.

The parents also need help and support; they need meetings with parents of other people with the same diagnosis. These meetings have a profound therapeutic state because here is the place where parents can be listened and helped forming support groups for: overcoming the crisis situation after receiving the diagnosis; difficulties faced by parents in relation to the sick person; stress management; fears about the future of the sick person; changes felt as parents in terms of understanding, sensitivity, and ability to cope with the difficulties.

The family affected by the emergence of a rare disease also faces from a socio-economical point of view financial efforts that often exceed the possibilities and resources the family has. In most cases the emergence of the disease means loss of incomes or their limitation for both the sick person, and at least one parent, who is forced to give up work to take care and support the sick person. There are also situations where some families break up because of the appearance of a sick person.

Consequently, both people with rare diseases and their families need support to seek and access the resources needed for successful integration into social life.

3. Communication

The case manager uses communication as the main working tool. This helps him both in interacting with the client and his/her family, as well as with the network of specialists with whom he collaborates.

For the efficiency of his work, the case manager must be a good communicator. Here are some essential elements of the communication theory, which must be adapted to the situation, to the needs and possibilities of the clients and to the personality, style of the case manager.

The communication skills can be improved over time by always analyzing the communication processes that have had the expected results and those that have led to an unwanted outcome.

What is communication?

- Communication is the process of transmitting information, ideas, and opinions, from one individual to another or from one group to another group;
- It is a process by which an individual transmits stimuli in order to change the behavior of other individuals;
- It is the transmission of a message (ideas, feelings, information, and opinions) from one person (transmitter) to another (receiver).

3.1. Goals and objectives of the case manager communication

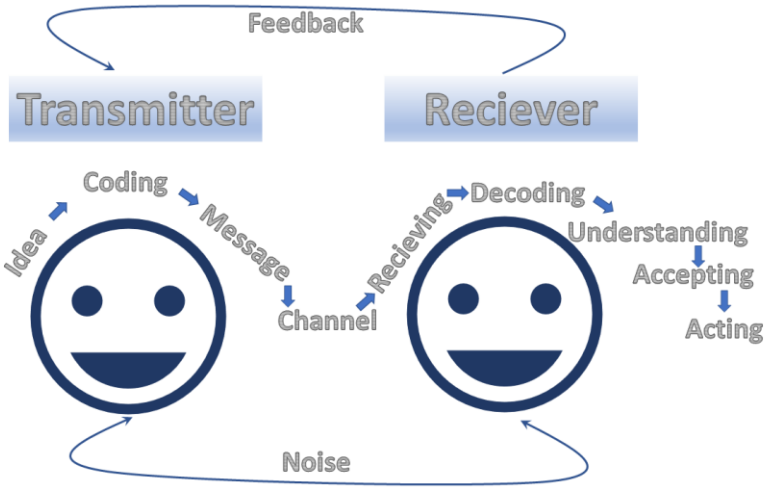
Communication' goals:
• Information;
• Training;
• Motivation/persuasion/encouragement;
• Procurement of information.

The objectives of the efficient communication:
• To be heard;
• To be understood;
• To be accepted;
• To provoke a reaction.

- *both of the patient, family members and of other specialists who are involved or should be involved in the management of the disease*

3.2. The elements of the communication process are:

- **Transmitter**;
- **Message**;
- Channel;
- **Receiver**;
- Feedback;
- Context.



Any communication involves **three basic elements**:

- the transmitter, where the communication initially starts,
- the message which is verbal, non-verbal, direct or indirect and
- the receiver, the recipient of the message.

The channel represents the environment through which information is passed from the transmitter to the receiver. This channel has a number of disturbing factors that have been reunited under the name of noise.

3.3. The stages of communication:

CODIFICATION: In communication, to codify means to translate thoughts, attitudes, emotions, feelings into words, touches, sounds, images. It consists in transcribing or transferring the respective message, information to be transmitted on a certain physical support belonging to an alphabet.

TRANSMISSION OF THE MESSAGE: consists in the transmission of the encoded message from the issuer to the receiver through various communication channels.

DECODIFICATION AND INTERPRETATION: refers to the deciphering of the transmitted symbols, respectively the explanation of their meaning, a formalized process in receiving the message. In psychological terms, decodification means understanding the message by the recipient.

FILTER: consists in the deformation of the meaning of a message due to physiological or psychological limits. Physiological filters caused by total or partial disabilities limit the ability to perceive stimuli and thus understand the message.

FEEDBACK: Ends the communication process. It can be positive or negative.

3.4. Efficient communication

The efficiency of the communication process is ensured by the following abilities (Lemeni, Miclea, 2004):

- identification of the sources of conflict and the use of specific conflict resolution strategies,
- active listening - the ability to capture both the content of the message and the emotions of the interlocutor, in order to understand the message as accurate as possible;
- assertive transmission of the message;
- the use of these skills in different communication contexts (dialogue, group communication, direct or mediated communication).

The development of these abilities is a complex and continuous process.

Barriers in communication

The communication skills are a proof of mutual respect. Ideally, every person should be able to listen to another person and show that he/she understands what the other wants to communicate. He/she should also respect himself/herself and express himself/herself in a non-aggressive manner.

See below some barriers in communication according to its origins:

Characteristics of the transmitter (E)

- contradictory or unconscious signals
- low credibility of the subject
- inability to communicate

Characteristics of the receptor (R)

- poor listening capacity
- prejudices related to the topic

Dynamics of interpersonal relationships between E and R

- semantics
- different statutes and degrees of influence
- different perceptions/opinions

Environmental factors

- Noise
- Defatigation

The effectiveness of communication depends on removing the barriers that may arise at each stage by planning the communication in a clear, accurate, concise manner.

This way:

The transmitter

To communicate effectively, he/she must first prove his/her credibility. He/she must know the receiving resources of the receiver and use expressive language accessible to him/her; send to the receiver messages that he/she understands and provide additional elements of understanding the message.

The message

If the message is too long, disorganized or contains errors, it is hard to understand. Messages also have emotional and intellectual components, which allow us to think about them and appeal to their motivational value. Messages must therefore be clear, explicit; convincing, appropriate, objective; concise, complete, good. They must have adequate content support.

Communication channel

Messages are transmitted through communication channels, which involve face-to-face meetings, phone and video conference for the verbal message, as well as letters, emails, reports for the written message.

The Receiver

Messages are sent to an audience. Undoubtedly, each person has in mind the actions and reactions they hope to awake in the audience. Do not forget that the audience comes in the process of communication with its own ideas and feelings, which will certainly influence the understanding of the message and the answer given.

Feedback

The audience will provide verbal and non-verbal feedback on the communicated message. Paying more attention to it is crucial to making sure the message arrived at destination.

Context

When communicating, we must keep in mind the context in which this process takes place (the environment, culture) and adapt our message accordingly.

It is important to keep in mind the time availability and the demands of the other people, especially in an over-crowded society like the contemporary one.

3.4.1. Reasons that prevent people from properly communicating with each other

1. Phobia of conflicts

The subject is afraid of conflicts or feelings of hostility expressed by other people. He/she believes that people who have good relationships with each other never argue. He/she also has the impression that close people will feel injured and will not be able to bear hearing what we feel or think.

2. Emotional perfectionism

The person believes that he/she should not experience negative emotions such as anger, jealousy, depression or anxiety. He/she requires to be always rational and with perfect control over his/her emotions, fearing to be perceived as weak and vulnerable. The subject is convinced that people will feel superior if they will find out what he/she really feels.

3. Fear of disapproval and rejection

The individual is so terrified at the thought of being rejected and ending up in solitude that he/she totally inhibits his/her feelings, accepting even others to abuse him/her, instead of manifesting his/her hostility by risking someone to get upset with him/her. He/she feels the exaggerated need to please others and meet their desires.

4. Passive aggressiveness

The subject represses his/her feelings of frustration and hostility instead of expressing them openly. In his/her relationships with others, he/she takes refuge in silence, trying to make them feel guilty instead of sharing his/her feelings.

5. Lack of hope

The person is convinced that his/her interpersonal relationships cannot be improved, no matter what he/she does, so he/she gives up trying. The subject also thinks he/she has tried everything and that nothing has worked or that his/her partner is too stubborn and too insensitive to change. As soon as the subject quits, things remain unchanged, and he/she comes to the conclusion that the situation is really hopeless.

6. Low level of self-esteem

The subject considers that he/she has no right to express his/her feelings or ask others for certain things. He/she thinks that he/she always needs to please others and meet their wishes.

7. Spontaneity

The person believes that he/she has the right to say everything he/she thinks or feels when he/she is upset. He/she thinks that any change in his/her style of communication is ridiculous and seems false.

8. "Thought Reading"

The subject thinks that others need to know what he/she feels and what he/she wants, without having to openly express his/her feelings and desires. This attitude gives him/her an excuse for hiding his/her feelings and nourishing resentment as others seem not to care about what he/she feels.

9. Maturity tendency

The individual is afraid to admit that he/she is mad because he/she does not want to give anyone the satisfaction of finding out that he/she hurt him. He/she is very proud of his/her ability to control his/her feelings and suffer in silence.

10. The need to solve the problems

When we have a conflict with someone, we do all sorts of attempts to solve the problem instead of openly sharing our feelings and listening to what the other person feels.

To this is added:

Criticism - Example: "You are to be blamed ..."

Labeling, Insults - Example: "How stupid! You speak like a peasant."

Sermon - Example: "You should...."

Counseling - Example: "If I were you ..."

Value Praise - "You've always been a good girl! You will help me, am I right?"

Differences of perception - Example: "Look how things are ..."

Command, order - Example: "Do what I say!", "Whatever is asked you do it!"

3.4.2. Cognitive barriers: Stereotypes and prejudices

A particular category of barriers to communication is cognitive barriers (stereotypes and prejudices).

Stereotypes are beliefs about the characteristics, attributes and behaviors specific to members of a social category. What is specific to stereotypes is the lack of sufficient knowledge about the members of the group. In other words, stereotypes are a generalization based on minimal knowledge.

Prejudices are negative attitudes, which involve unfavorable generalizations in relation to members of a particular group. Prejudices have 3 components: a cognitive one (stereotypes), a negative affective one and a behavioral one (discriminatory behavior).

For case managers, preparing for communication helps better transmit correct messages, and analizing past communication situations and identifying the barriers help in better communicating in the future.

3.4.3. Active listening

Listening (receiving the message) is the first step in establishing an effective communication relationship. Active listening is done in the following ways:

- establishing visual contact with the person we are talking to,
- use of minimal response and encouragement,
- focusing on the speaker and what he/she says,
- reflecting feelings,
- asking questions,
- paraphrasing, summarizing.

a) Establishing visual contact with the person we are talking to

The visual contact with the interlocutor shows that we are attentive to the discussion. When we want to start a conversation with someone, it is important to establish visual contact and interrupt the work we are involved in. Continuing the activity is a sign of lack of interest, it conveys that the other's message is not interesting.

b) Use of minimal response and encouragement

It automatically appears in our conversation either in non-verbal form (to nod, "yes"), or verbal form ("I understand," "I know what you mean"). It tells the speaker that the person with whom he/she is talking is paying attention (Child: "I do not like painting because I dirty my hands." Case Manager: "Hm, I understand what you mean").

c) Focusing on the speaker and what he/she is saying

It is very important when we listen: to focus our attention on the ideas and feelings transmitted by the other person, to avoid thinking about our own problems, not to change the subject.

d) Reflecting the feelings of the other

Reflection has the role of explaining and clarifying the emotions of the interlocutor at one certain point.

Example:

Case manager: Simona, how are things with your mother?

Beneficiary: Well she is still not finding work ... and school is starting and I need notebooks and more other things.

Case Manager: I think you are quite tense and agitated.

e) Paraphrasing

Paraphrasing involves reformulating what a person says when the case manager wants to make sure he/she understands what the person is telling him/her. Paraphrasing does not involve the evaluation or interpretation of what the interlocutor said.

Example:

Beneficiary: Every time my dad promises me that he will come to see me playing and always something more important comes up. They are never there when I need them.

Case Manager: As I understand you're angry because your parents are not around you when you need them, and because your father promises you some things and does not carry out his promises.

Active listening guide:

- **Empathize.** Put yourself in the other's shoes and try to understand what he/she feels.
- **Be aware of the feelings or emotions** expressed by the speaker and of what he is trying to communicate.
- **Validate other people.** This does not mean that you agree with other people, only that you have heard the person and understood his/her position.
- Paraphrase or rephrase with your own words what the speaker said to make sure you understood what was said.
- **Clarify the situation.** Ask questions to get more information about the problem.
- **Collect information** to help you better understand why the speaker acts or feels in a particular way.
- Try to identify the **interests.**
- Be calm!
- Use the person's name when you answer them. **Customize.**
- Keep an appropriate conversation tone.

f) Summarizing

The case manager summarizes from time to time the information provided by the beneficiary. Summarization has several roles: such as fixation and clarification of information.

g) Question enunciation

There are two types of questions: open and closed questions.

Closed questions are questions that generate specific answers, in terms of "yes" or "no", or answers that offer little information such as: "How old are you? Do you like school?".

These questions are used when we want to get some concrete information or when the other has difficulties expressing himself/herself.

Open questions allow the exploration of relevant content and emotions. Such questions can be: "*Can you tell me what you liked most about today's activity?", „How do you feel?", „What can you tell me about your school?*". Therefore, the answers of the interlocutors are much richer from an informational point of view.

3.4.4. The importance of nonverbal communication

It does not always matter just what you say, but especially how you say it – considering the look, the posture, the whole body language, the external appearance at the moment of the communication and the voice.

Even if verbal language is done with words much of the message is nonverbal. Research shows (Lemeni, Miclea, 2004) that, in communication, nonverbal language has a weight of about 85%.

Nonverbal communication allows us to send an independent message and accentuate a verbal message that we transmit.

Some examples of messages sent entirely by nonverbal language:

- to wave to a friend ↔ hello
- hit the table hard ↔ nervousness
- smile ↔ joy
- frown ↔ dissatisfaction, caution
- to cover the mouth ↔ to silence
- to nod/to toss one's head ↔ consent/dissent
- two fingers up ↔ the desire to respond
- thumbs up ↔ everything is ok, etc.

The nonverbal indices are very strong. They give us information about the interest and understanding that our message poses to others.

The main **non-verbal indices** are: gestures; visual contact; facial expression; clothes and personal space.

Nonverbal communication guide

- Be careful of your own and surrounding non-verbal indices.

- Be careful of gestures, because they can both complement and contradict your message.

- Visual communication is important in communication.

- Closely related to visual contact are facial expressions that can reflect attitudes and emotions.

- Posture also helps you to communicate your message more effectively. (Examples: walking with your shoulders down ↔ sadness, kneeling ↔ request).

- Clothing is important. Its adequacy shows respect for the values and conventions of the organization in which you are integrated.

- Be aware of other people's personal space when communicating.

3.4.5. Assertive communication

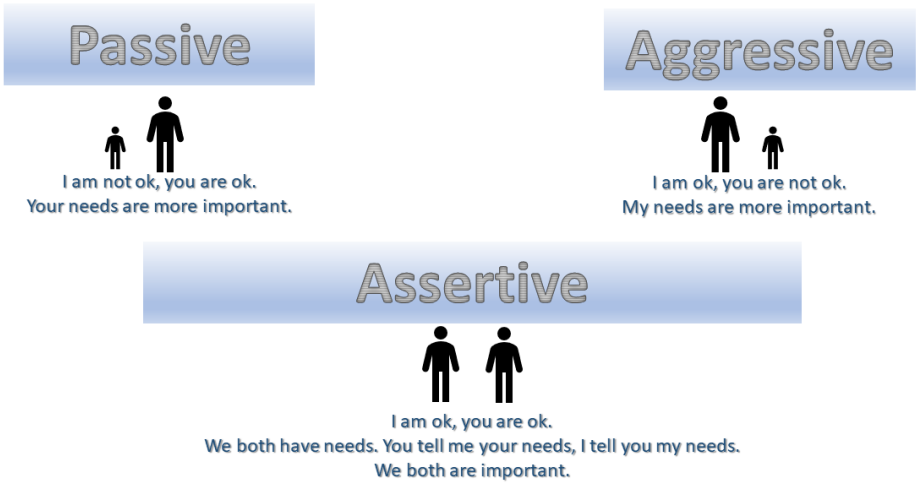
Assertive communication has developed as an effective way to respond to conflicting interpersonal situations. Lack of assertiveness is one of the most important sources of social inadequacy. **Assertiveness** is the result of a set of attitudes and behaviors that have as long-term consequences the improvement of social relations, the development of self-confidence, the respect of personal rights, the formation of a healthy lifestyle, the improvement of the responsible decision-making abilities and the improvement of conflict management abilities.

Assertiveness is the ability to express our emotions and beliefs without affecting and attacking the rights of others. The learning of assertive communication skills is facilitated by contrasting it with the two opposite behavioral patterns, passivity and aggressiveness.

Passivity (passive behavior) is the answer of a person who tries to avoid confrontations, conflicts, who wants everyone to be satisfied, without taking his personal rights or desires into account. These people feel hurt, frustrated, irritated, but without trying to express their discontent with others.

Aggressiveness is a behavioral reaction by which you blame and accuse the other, you violate the rules imposed by the authorities, you are insensitive to the feelings of others, you do not respect your colleagues, you think you are always right, you solve the problems by violence, you think those around you are often unjust with you, you are sarcastic, and you often use critical communication, you think that your rights are more important than others, you are hostile and angry.

The relationship between the three types of communication is presented in the picture below:



Consequences of assertive, passive and aggressive behaviors:

Passive	**Assertive**	**Aggressive**
The problem is avoided	The problem is discussed	The problem is attacked
Your rights are ignored	Your rights are upheld	Your rights are upheld without regard to the rights of others.
You let the others choose for you	You choose your activity	You choose your activity and that of others.
Distrust	They trust in themselves	Hostile, they blame, accuse
You see the rights of others as being more important	You acknowledge the right you and others have	Your rights are more important than others

Assertive style is not a fixed recipe, which you can use at any time and with anyone. But it is a method of being honest with ourselves and with the others. A way of telling what bothers us without hurting each other's feelings.

Giving an assertive message involves 3 steps:

- **expressing feelings,**
- **presenting the problem (what bothers you?),**
- **presenting your own solution to the problem.**

3.5. Developing co-operate communication skills

Benefits of using a co-operate communication style:

More achievements, more fun, or a *better coordination of the personal activities* with the *activities of those important for you*. The better you understand what others feel and want, and the clearer the others are about our goals and feelings, the more sure we are that all of us are on the same path.

More respect. There is a great deal of mutual imitation in everyday communication. Example: I shout, you start shouting too. If we adopt an attitude of respect and understanding towards the conversation partner, we invite him/her to do the same thing for us.

More influence. When we are honest and responsible, it is easier for us to involve other people in our work and to obtain agreements that will please everyone.

More comfortable in conflict situations. Because each person has different talents, more can be achieved if people work together and accomplish in groups what they could not achieve by themselves. But each person also has different needs and views, so there will always be conflicts in living and working with others. By understanding the message in the conversation, we can solve problems and mediate conflicts.

Less worry. Conflict situations and our actions against others have lasting echoes in our mind and body. If we are cooperative even in unpleasant situations, we will have a good mental state.

Closer relationships with others.

Healthier life.

Why co-operate skills do not develop on their own?

To co-operate requires a greater mental effort than to impose, to command what you want. Our way of communication is deeply rooted in our personality. For example, a person

who has been criticized and not listened every time he/she has said something might develop an aversion to the discovery of his/her thoughts and feelings.

We are usually too busy in observing others and the world around us to notice our own communication errors. Learning new communication skills takes time because we are surrounded by a flow of negative examples.

3.5.1. Suggestions on developing a more co-operative communication.

a) Listen more carefully and responsibly

It is important to listen and understand what you hear, even if you do not agree with the point of view, before expressing your own point of view. In order to get more attention from the conversation partner, in tense situations, you must first pay attention to him/her. Listen and give a brief restatement of what you have heard (especially feelings) before expressing your own needs or positions. The type of listening recommended separates "*understanding*" from "*approval or agreement*".

b) Explain your conversational intention and make invitations to consensus.

To help your conversation partner to cooperate with you and to reduce possible misunderstandings, start important conversations by inviting your conversation partner to join you. Successful "speakers" usually start a special conversation with a preface like, "*I would like to talk to you a few minutes about ... (subject) (example: organizing a trip). When would be the right time?*"

c) Express yourself as clearly and completely as possible

Do not hurry and give your listeners more information about what you are experiencing using a wide range of affirmations in first person singular. One way to get more out of the listener's empathy is to express as much as possible of the five dimensions of the experience you have lived.

Dimensions of the experience	*1. What do you see, hear or feel through other senses?*	*2. What emotions do you feel?*	*3. What interpretations or desires support these feelings?*	*4. What actions, information or engagements are required?*	*5. What will be the result in the future?*
Example	"When I saw 2 persons were late…	I was worried …	Because I was afraid the whole schedule would be messed up…	And I want to ask you to call and see what are they doing..	So we can start in due time without messing up the schedule.

d) Convert the complaints and the criticisms (to yourself and to others) into specific requests. Explain these requests.

To get more co-operation from others, whenever possible, formulate the desired requests using a specific, action-oriented, positive language, instead of a general one. Thus: the partners will focus their attention on the present situation; will cooperate more easily; will imagine new solutions. If you receive complaints or you are being criticized, try to convert these statements into action-oriented requests.

e) Formulate open and creative questions

Our common questions with YES/NO answers are rather barriers than facilitators for communication. To encourage the conversation partner to express his/her feelings it is useful to ask open questions.

f) Express more appreciation

To build more satisfactory relationships with those around you, you should express more appreciation, delight, encouragement, and gratitude. Because life is constantly asking us to solve problems, it is very easy to notice much easier the problems and the defects.

g) Consider effective communication an important part of everyday life.

Practice effective communication in a variety of situations, so it will become a habit, a "second nature."

3.5.2. How do we deal with difficult people?

a) Communication with passive-aggressive subjects. These people, instead of telling us they are angry, they refuse to talk to us. In such situations, Burns recommends *empathizing*; instead of insisting that the person speak, it is better to ask in a friendly tone why he/she refuses the dialogue. Active listening techniques are also useful in addressing passive-aggressive subjects because they lack the assertive self-expression skills. They are upset and do not know how to express their true feelings.

Passive, anti-aggressive technique. If the person still refuses to speak, we accept the idea that he/she is not a suitable person for dialogue. We will insist that some communication is necessary and we will insist for it to happen later when the dispute partner is in a better mood.

b) Communicating with aggressive (hostile) subjects Instead of refusing communication, our interlocutor can become aggressive, answering sarcastically or in a high tone. In such a situation, most of the subjects adopt a defensive attitude; they step back or feel hurt and critique themselves. The appropriate attitude in that situation is to listen and discern what is real in the other's words, as well as to identify the feelings of the latter.

c) Communicating with a stubborn and quarreling person. The biggest mistake we can make in such a case is to try and convince the other that they have to listen to us. Such an approach never has good results. If we want to get someone’s understanding, it is necessary that we first give him/her our understanding. First of all, we need to ask ourselves why our partner is so stubborn and quarreling.

d) Communicating with a hypercritical person. Effective communication when someone criticizes us involves the use of communication skills on one hand and, on the other hand, making changes within our system of attitudes and values. The criticized person feels humiliated, frustrated and tends to become defensive. Moreover, he/she does not recognize this and the defensiveness is increased. Instead of doing so, it is appropriate to admit openly how we feel "I feel hurt by your words and I want to defend myself."

e) Communicating with people with unreasonable demands. One of the reasons we feel exhausted and guilty is that we cannot refuse requests from others. The real problem is not the request that people around us ask us, but the fact that we do not know how to refuse them. One of the most effective methods is that of postponement. If we consider that we are overtaken or that the request is unreasonable, we must learn to say "no".

f) Communicating with a person who complains permanently. Persons complaining permanently do not want any advice but just want to be heard. In such situations, the disarmament technique must be used, through which we will try to agree with them rather than "help them" and immediately they will calm down and feel better.

3.6. Conflict resolution

The Latin word "conflictus" (violent interference) involves disagreement, discord and friction among group members: interaction where words, emotions and actions compete to produce troublesome effects. (De Visccher, Neculau 2001).

Conflict is a normal phenomenon in all relationships and groups. The conflict itself is neither good nor bad. Rather, the attitudes and reactions of those involved make it constructive or destructive. Yet people generally perceive conflict as something negative and feel uncomfortable in its presence.

If a conflict is good or bad it depends on how its management is done. Generally, if the outcome of a conflict is positive, then the conflict is considered "good" and if the outcome is negative, the conflict is considered "bad". In the absence of conflicts, progress is impossible to achieve. Throughout our lives, we are involved in a lot of conflicts.

There are several types of conflicts:

- **Intrapersonal**: conflict with oneself (for example, a person who cannot make decisions);
- **Interpersonal:** conflicts between two or more individuals (for example, a contradictory discussion between friends or between students and teachers, child and parent, etc.);
- **Intragrupal**: conflicts within a group (for example, between company employees);
- **Intergroupal**: conflicts between two or more groups (example between gangs, between students, etc.);

Strategies to resolve conflicts

Negotiating conflicts involves developing skills and practicing them in everyday life. So without claiming that we offer recipes, we will present the skills that are considered to be effective in resolving conflicts.

The win/win approach - the conflict is no longer seen as an attack or a defense, but the emphasis is on co-operation.

This approach has as main statement, "I want to win and I want you to win too." The rule is: "I'll do X for you if you do Y for me" (where X and Y are the needs of the two sides).

The win-win approach implies:

- Returning to the needs of those involved
- Recognizing the individual differences
- Willingness to adopt a position based on shared attitudes and information
- Attacking the problem not the people.

Creative responses - involves turning the problems into opportunities, involves seeing the good part of the situation. Our attitudes color our thoughts. Sometimes we are not even aware of how they color our world. Let's call these attitudes "hats": What hat will you wear every day? Do you see the difficulties of choosing as a problem or as a challenge?

Empathy - refers to our ability to put ourselves in the other people's shoes, to understand their emotions, thoughts and behavior.

Appropriate assertiveness - Assumes affirmation of one's own person without attacking others. Details about assertiveness have already been given in earlier chapter.

The power of cooperation - We recall the cooperation involves exploring, clarifying the details, finding possible solutions, redirecting to the positive aspects, use of the abilities described above to understand the needs and concerns.

Emotion Management - When it comes to managing emotions, we must take into account the purposes lying at the basis of the behavioral problems (often conflict-causing). These are:

- Searching for attention - "I feel part only when I'm noticed"
- Power - "I count only when I'm in control when no one orders me"
- Revenge - "I'm important only when others suffer as I suffer"
- Inadequacy - "I will not be hurt again, if I manage to persuade others not to wait too much for me"

Willingness to resolve situations - In order to resolve the conflict situation, it is important to analyze and resolve the following issues first:

- Suppressing needs - example Failure to recognize their own needs
- Unsolved personal histories – example: If someone's rights were violated in childhood, he/she might react violently when his/her rights are violated as an adult.
- Unacceptable qualities – example: Someone who does not accept his/her own anger may not even accept the others' anger.

Conflict Map - helps us to make a more objective image of the conflict.

- Briefly define the problem in neutral terms.
- Write all the people important to the conflict
- Write the needs of each person or group. What motivates them?
- Write for each person the fears, concerns or anxieties.
- Be able to redefine the problem as its understanding evolves.

Developing options

Clarification tools

- "Chunking" - splitting the problem in small steps
- Re-investigation - more information, expanding resources, constraints
- Setting goals - what result do we want?

Generating tools

- The obvious solution - to which all parties say "yes".
- Brainstorming - without censorship, without justification, without debate
- Consensus - Build a solution together
- Lateral thinking - are we practical, creative?

Negotiating tools

- Keep the current arrangements
- Exchange - what is easy for me to give and valuable for you to receive?
- Test and error - try one option then another
- Establishing alternatives - what will happen if we cannot agree?
- Meeting the consequences - what will I do if we cannot agree?

Selection

- Is it built on the win-win approach?

- Does it meet everyone's needs?
- Is it feasible?
- Is it right?
- Does it solve this problem?
- Can we stop at one option or we need some tests?

Negotiation - Basic principles in negotiation

- Strongly attack the problem and not the person
- Focus on needs not on positions
- Insist on common points
- Be creative about options
- Make clear commitments

Mediation - involves asking for help from a third person when the problem cannot be solved by those involved. The mediator may be a professional or any other person with communication skills who is not involved in the conflict.

Expanding the perspective

Here are some suggestions to broaden our perspective on how to approach the conflict:

Respect and valuate the differences - people have different points of view that may be true depending on the angle they are viewed from.

Analyze the problem in the long run - how will the issue or relationship look over a substantial period of time?

Take a global perspective - since the actions of the group members are interconnected, we are interested in the significance of our actions in interaction with others.

Be open to the idea of changing and assuming risk.

3.7. Networking

The network of specialists from the various services that the case manager's client needs is essential and constantly transforming.

The case manager has the task of knowing these services, initiating and keeping in touch with the specialists in these services, both in order to solve the situations with the beneficiaries they work with, as well as to facilitate future communication for the beneficiaries they will have introduced to these specialists.

Collaboration with local, county and national authorities supports the formation of the case manager's resource network.

It is very important for the case manager to use in the communication process the appropriate communication methods with the different interlocutors (client, family members, institution staff, external collaborators). Each interlocutor, influenced by membership groups but also by personality differences, has a different style and other need for communicating with the case manager.

The goal pursued in communication by the Case manager can be:

- knowing the client;
- assessing the needs of the client and of the family members;
- assessing the development opportunities;
- establishing the client's field of interest;
- training and education of the client;
- skills training;
- affective relationship between the client and the case manager;
- informing the specialists from the client's management network on the evolution of the client.

The Case Manager's most important skills are:

- Active listening;
- Attention;
- Understanding the client's specific needs;
- Communication and easy establishment of relationships;
- Analysis and synthesis;
- Flexibility;
- Adaptability;
- Self-confidence;
- Accuracy;
- Evaluation;
- To win the client's trust.

4. Increase family resilience. General information.

4.1. What is family resilience?

Resilience is a concept that has been used more and more often lately.

Resilience refers to the ability to overcome any unpleasant events, to adapt to change, and to recover from the difficult experience that has taken place. Resilience is the ability to overcome major problems of any kind. Resilience is the process of adapting to adversity, traumatic events, tragedies, threats or significant sources of stress such as family and relational issues, health or workplace problems, financial stress.

Resilience is not extraordinary but ordinary. People usually demonstrate the ability to adapt. Therefore, some people have the ability to recover easily from an emotional point of view and to rebuild their lives after traumatic events.

Being resilient does not mean that a person does not experience any difficulties or stress. Emotional pain and sadness are common in people who have suffered major injuries or traumas in their lives. In fact, the way to becoming a resilient person is likely to involve considerable emotional stress.

Resilience is not a feature that people have or not have. It involves behaviors, thoughts and actions that can be learned and developed by anyone.

Resilience refers to the positive adaptation to difficult traumatic conditions, keeping mental health intact.

It is a quality that every human possesses natively. The basic components of resilience exist in each of us: optimism, ability to solve problems, adaptation to the environment, communication skills or the power to handle difficult situations, the ability to identify solutions alone.

4.2. Factors that contribute to increase family resilience

Many studies show that the main factor in resilience is supportive relationships both inside and outside the family. Relationships based on love and trust offer role models, encouragement, but also assurance in strengthening a person's resilience.

Other factors that are associated with resilience include:

- the ability to make realistic plans and take action to achieve them.
- a positive self-esteem, and trust in strengths and abilities.
- problem solving and communication skills.
- the ability to manage strong feelings and impulses.

All of these are factors that people could develop in them.

Developing resilience is a personal journey. People do not react the same for traumatic or stressful events in their lives. An approach to building this capacity that works for a person may not work for another. People can use a variety of strategies.

But most of the time people fail to develop their resilience skills by themselves, and when it comes to people with disabilities, things are even more difficult. The case manager can support the patient and the family in increasing and strengthening resilience.

However, there are also ways to cultivate and strengthen resilience, to train our genetic qualities to better prepare ourselves to cope with major issues.

The factors that determine the resilience of a person cannot be fully identified. Among the useful qualities are: a strong personality, optimistic thinking, cognitive flexibility, adaptability, solution orientation, etc.

Very important is the social support, the good relationship with family and friends who can help in difficult times.

Some psychologists believe that religion, faith, spirituality are the main, undeniable source of human resilience, and they are convinced that this is from where the inner power, that is actually stronger than any external pressure comes from. Of course, many other factors can be considered.

It must be said that even a resilient person feels hardship and stress. No one has immunity. But the resilient person reacts differently. The resilient person has the ability to overcome difficulties, to learn from unpleasant experiences, to come back every time stronger, wiser, and more prepared for potential future shocks, understanding that events of this type are part of life.

The brain is like a muscle. It must be trained. Resilience is a sum of abilities. They must also be trained.

Psychologists have developed a resilience development plan to identify and cultivate skills that are favorable to the personality profile of the survivor.

The main objectives of this plan are as follows:

- Supporting a positive image of oneself with the identification of the qualities and life moments successfully overcomed;
- Establishing a purpose and a sense of life;
- Forming a circle of friends and supporters;
- Behavioral optimism and flexibility;
- Keeping in touch with life issues;
- Achieving a balance in all areas of life: health, work, marriage, affectivity;
- A varied daily program, with meetings, reading, shows, entertainment;
- Avoiding strong emotions in confronting difficulties;
- Observance of rest schedule, bedtime, holidays, etc;
- Establishing a future plan.

Unlike other countries where the families of the children with disabilities produced by rare diseases receive the necessary support, starting from the communication of the diagnosis (communication carried out in the presence of both spouses, having an interdisciplinary team supporting the family at such times: psychologist, social worker and a patient or person who has gone through such situations and managed to overcome them), in Romania it does not happen like this. Hence, the many problems faced by families later on.

The psychological resilience of the families with children with disabilities caused by rare diseases means the ability to recover the mental functions after a strong stress caused by the announcement of the disability, family trauma, severe living conditions, job loss for one of the parents, prolonged unemployment and other similar situations. Psychologists have questioned how some people manage to get over these difficult moments, to restore their

previous mental state and even to become stronger. They have drawn up a personality profile of the survivor. Some skills developed in difficult conditions have been identified and that many of those who have experienced difficult times in life become good professionals, find the best solutions to life's problems, do not victimize themselves, trust themselves, have active social life, are optimists, have friends and have a perspective project.

4.3. Empowering patients and their families

4.3.1. What does patient empowerment mean?

The World Health Organization defines the empowerment of the patient as a process by which people gain control over the decisions and actions that affect their health and must be seen both as an individual process and as a collective process.

An empowered patient:

- Knows and understands his/her real health condition and its effects on his/her own body;
- Feels able to participate in the decision-making process together with the specialists who treat him/her;
- Feels able to make informed decisions about the treatment;
- Understands the need to make the necessary changes to his/her lifestyle for the correct management of the disease;
- Takes responsibility for his/her own health and calls for assistance only when necessary;
- Searches for information, evaluates it and uses it.

An empowered patient will better understand how to navigate between the many players in the medical system, including family, doctors, health insurance, pharmacists, decision-makers, etc. When the patient does not trust himself/herself and does not know where to go, which are the next steps to take, which is the behavior to follow after diagnosis, an empowered patient is the one who will feel able to ask the right questions in order to receive the information that he/she needs.

In the process of empowering patients, 4 components are considered fundamental:

- Understanding the patient's own role;
- Acquiring sufficient information to enable the patient to get involved together with the healthcare provider;
- The skills the patient has;
- The existence of an environment that will facilitate this process.

The case manager can support patients by:

- encouraging the active participation of patients and their careers in choosing management options to improve the quality of life;
- guiding the patient in seeking and obtaining the necessary information on treatment options so that the patient can actively participate in the choice of treatment (to guide him/her towards specialists and services);
- making them understand that they can be active participants and not passive receptors in the care process and should also be well informed about all aspects of their state of health and the stage of the disease, in order to achieve the maximum benefit in the context of their social actions.

A key objective in the patient empowerment process is to educate patients about policies that might affect their health care services in the future. No matter where we live in the world, inequities in access to diagnosis, care and treatment are a reality.

The empowerment is a multidimensional process that helps people gain control over their own lives and increases their ability to act on issues that they consider important.

The empowerment is also a process by which people improve their capacity to identify their own resources to live well and navigate through the health system.

The empowerment includes self-esteem, self-knowledge, trust, coping skills and medical knowledge. The empowerment is not a simple process, and it is not necessarily a linear process.

A patient may feel empowered in a certain context and non-empowered in another context. During his/her journey, the feeling of being empowered may change, of course, depending on the person and on the environment the patient encounters.

An empowered patient has control over the management of his/her own health in everyday life. They take action to improve their quality of life and have the necessary knowledge, skills, attitude and self-knowledge to adapt their behavior and work in partnership with others when necessary to achieve optimal well-being.

Empowerment interventions aim at equipping the patient with the ability to take decisions on his/her health status as long as he/she want to become co-manager of his/her own disease in partnership with his/her doctor and to develop self-confidence, self-esteem and coping skills to manage the physical, emotional and social impact of the disease in everyday life.

Civic involvement in the health system is both a right and a duty.

People have the duty and obligation to individually and collectively participate in the planning and implementation of their own health care. Patients have a specific expertise that results simply from being patients, and this is a valuable source of knowledge based on experience. The patient's knowledge results from the fact that they live with the disease day by day and from the permanent contact with the health system.

Besides being a right, patient involvement is recognized as having benefits for the development of therapies and health services with a positive impact on patients and society. Patient engagement in health policies and programs will lead to services that will provide real value for patients and will reduce unnecessary services.

4.3.2. Health literacy

It is a key aspect of empowering the patient. Empowerment is more than becoming an educated/informed patient. Accurate information and resources are fundamental tools for empowering the patient.

Health literacy has been defined by the World Health Organization as the cognitive and social ability of people to have access to health information, to understand this information, and to use it to maintain a healthy lifestyle. Health literacy allows people to understand how to use that information to influence the way they care for their health.

Literacy in health should also be more important in the medical system in Romania, where, as elsewhere in the world, patients face difficulties in understanding vital medical information for them and for the management of the disease.

The case manager can play an important role in this - he can help the patient find the answers to the questions he/she has, help him/her look for and find the correct information, or who to turn to get answers to his/her questions.

Source: WHO. (2009) Background Note: Regional Preparatory Meeting on Promoting Health Literacy [Internet]. A ECOSOC.

In order to make truly informed decisions about health and treatment, it is vital that patients have access to all relevant information in an easily understandable format. Based on his/her knowledge, the patient is then able to process, appreciate, and apply the information to his/her own personal circumstances. Patient information needs vary according to age, socio-economic status, gender, beliefs, adaptation preferences and strategies, etc.

4.4. Announcing the diagnosis

Informing patients and their families about the diagnosis is a heavy burden for any physician. The way the diagnosis is communicated will have a major impact on the physician-patient relationship and on the patient's attitude towards the disease and treatment measures.

The announcement of a diagnosis generates various reactions, from shock to refusal in believing and especially accepting it. In the professional literature, there are described 5 stages or psychological reactions that people with incurable diseases and their families are going through after finding out the diagnosis:

- denial;
- anger;
- negotiation;
- depression;
- acceptance.

DENIAL is a normal reaction, after the initial shock of finding out the diagnosis, typical and necessary. It is the simplest mechanism to deal with the situation. The patient or his/her parents refuse the validity of the diagnosis.

ANGER usually follows denial, helping relieve the suffering. The disease is perceived as an unjust and undeserved punishment. The person seeks a guilty person responsible for his/her situation: anger can be directed to family members, health professionals, to their own person, etc. This manifestation proves a beginning of integration (the person begins to understand that he/she has a disease).

NEGOTIATION usually involves a change of behavior or a specific promise in return for a longer life. Negotiation is often in secret with God, recognized as the author of life, and it is a stage in which the person invests in various future projects.

DEPRESSION occurs when the disease has a negative evolution and if repeated admissions or surgical interventions (as the case may be) have no results.

The individuals often become solitary, isolated, and desperate; they feel useless, without future, without desires. Depression occurs when the person is aware of the losses brought by the disease and begins to feel sorry for himself/herself. Sometimes hope can persist in this phase of the disease, hope that a miraculous treatment will be invented.

Many times ACCEPTANCE is, first a rational reality and after an emotional reality, it is the phase characterized by emotional calm, without too big fear, joy or sadness. Acceptance is the phase in which a person accepts to live with the disease and wants to fully live each moment.

The case manager can intervene and support the person by showing him/her some ways to overcome crisis situations or by sending him/her to the abilitated persons who are able to support him/her.

Methods to overcome crisis situations:

- Communicating with family members and friends to receive the necessary help and support;
- Requesting guidance and assistance from a specialist (psychiatrist, psychologist);
- Requesting spiritual support (priest);
- Accepting the situation by adopting adaptation behaviors: keeping/maintaining the friends network, listening to music, watching movies and musical/theater performances, walking in nature, various occupational therapies, reading books with cheerful or interesting subjects;
- Communicating with other people that are in the same situation can often be a support for the patient. He will understand that he is not the only one in this situation. Success stories of other patients can be real emotional support and can cause the patient to see the future with optimism.

4.5. Personal development and self-confidence

4.5.1. What is personal development?

Personal development includes activities and experiences that have as ultimate goal the improvement of awareness, development of talents and personal skills, ***improvement of the quality of life,*** and contribution to achieving personal aspirations and dreams. (Wikipedia)

Personal development is the personal and continuous process by which we change ourselves for better each day. No one is perfect and no one has been born wise, but we can make an effort to gain self-knowledge, skills and new behaviors so that our personal and professional life can be fuller. Every person should know what he/she wants from life and what he/she needs in order to fulfill his/her wishes.

The first step would be raising awareness that we need something extra. Then define exactly what we want to improve:

- maybe we do not know how to set our goals;
- maybe we do not have a very good picture of ourselves and this holds us back;
- maybe we have some beliefs that sound like barriers or maybe communication is deficient. It is important to have clear what we want to improve and then see how we can do it.

There are several ways we can improve the quality of life, the relationships, health, career, spirituality. The strategy may include reading specialized books, attending events or different meetings on areas of interest, attending a course or coaching sessions.

Changing some habits and opening up to another perspective - are provocative and possible. To succeed we need to change. We, not others! We need to get out of the comfort zone and not fool ourselves that what we know is enough. The world is in constant change, and if we want to succeed the solution is to keep pace, to be better tomorrow than today.

Through personal development, we acquire new knowledge and we broaden our own mental map. And in this way we are more prepared for new situations related to work, relationships, stress, etc., that will arise in life.

4.5.2. What is self-confidence?

Nathaniel Branden said that self-confidence means, above all, an experience. It means seeing that you can cope with everyday challenges. It means trusting your ability to think, learn, choose, make decisions, adapt to changes ... And it means knowing that you deserve happiness. Having this basic confidence is a matter of survival. To recognize what feeds on self-esteem and what damages it is one of the challenges that must be faced to live consciously, lucidly. Because reality is sometimes different from what we think.

For the patient, self-confidence means learning how to deal with the disease every day. The case manager has the role to teach him/her how to better manage his/her disease and thus improve his/her lifestyle.

Patients are experts through the experience they have, living with the disease every day. Their perspective on their own disease is unique: living with the disease every day, the patient learns how to manage it alone with the help of medical professionals, and out of necessity he/she learns how to navigate through the sanitary system to get the best care.

The case manager has the role of supporting and teaching the patient how to navigate through the socio-medical system, how to access the services he/she needs, so that he/she succeeds in better managing alone his/her disease and have an improved quality of life.

In order to learn how to manage his/her disease every day, the patient needs help from the team involved in the management of the disease - starting with the case manager who will help him/her identify his/her needs and then the available services will help him/her access these services and get in touch with the other specialists. The team of specialists should support the patient in managing the disease, as well as its physical, psychological, emotional and social impact. Through self-management, patients can develop the self-confidence, the self-efficacy and the skills they need to have control over everyday life and to improve their quality of life.

Self-management does not mean letting the patient deal alone with the situation. It is a holistic approach providing the appropriate support and the necessary tools according to the patient's individual needs and preferences. Self-efficacy refers to an individual's belief in his/her ability to learn and successfully perform a specific behavior. A strong sense of self-efficacy leads to a sense of control and to the desire to undertake new and difficult tasks.

5. Development and orientation

5.1. Independent life abilities

One of the objectives of case management is to improve the functioning and well-being of the beneficiary by focusing on high quality services in the most efficient way, to address complex needs and to improve the quality of life of the beneficiary.

In order to achieve this goal, ***one responsibility of the case manager is to identify and direct the beneficiary towards certain independent life skills training services, indispensable skills for the harmonious personal development and for the accumulation of skills and abilities to live independently.***

The concept of "independent living ability" is defined as "a set of knowledge, behaviors, attitudes and performances required for a quality life in any psychosocial context." It is, in fact, the set of skills a person needs to make the right decisions to fulfill the aspirations for a quality life.

In order for the intervention to be efficient and beneficial for the beneficiary, the case manager follows and identifies an independent quality life skills training service that strengthens the beneficiary's capacities for personal development, for handling and solving problems and increases the abilities of the beneficiary to interact and to function in the community, all respecting the values and the personal goals of the beneficiary.

The case manager can guide and train the beneficiary in developing independent living skills.

The case manager is responsible for monitoring the training the independent living skills of the beneficiaries.

The training service, to which the manager directs the beneficiary, can use methods that engage the participation of the beneficiaries in the learning process, which promotes responsibility for this process. Methods that highlight both the learning process and the learning content and product. They also focus on affective, as well as cognitive learning.

The development of independent living skills for the beneficiaries is a life-long process. In the education system, it must be provided for all age groups, according to the needs of the individuals and of the groups. Education must be coherent, continuous and must stimulate development.

In their training, the beneficiaries go through different modules focused on areas of independent life skills, such as: daily living habits (cooking, meal preparation, house management, etc.), personal care (personal hygiene, health, etc.), community housing and resources, social development, money management, work and study skills. The beneficiaries go through these modules, both theoretically and in an interactive way, thus having the chance to acquire the skills necessary for an independent life.

The modules must be personalized to suit every beneficiary, children and young people regardless of gender, ethnicity, culture, sexuality, capacity/incapacity and social circumstances. The beneficiaries, children and young people, have an enormous personal and social potential. Both they and the adult beneficiaries confront themselves in this world with complex difficulties that are changing rapidly, and society needs healthy, fulfilled, responsible and participatory citizens.

Methodological principles for the development of independent living skills

- To accept the idea that the skills the beneficiaries do not have can be developed through exercise, will.

- Attention to how participants are motivated. They should not perceive activities as mandatory.
- Take into account the significance of psychological and biological age.
- Apply education for lifelong learning in day-to-day work.
- The training for independent life should not be limited to the institutionalized environment, but it should happen in different environments (school, community, street, local, child and youth center, etc.)
- The education for lifelong learning recommends the application of participatory teaching and learning methods.

5.2. Daily life management

Among the case manager's attributions is also the monitoring of the level of the beneficiary's skills training on day-to-day management. This includes home management and leisure management, to facilitate integration into society's norms.

Home management

Order and cleanliness should be two strong points of a person's home. When the house is well organized and maintains cleanliness, in fact in every season there is a general cleaning to get rid of objects that are no longer useful, it can be saved a lot of space, which can then handle new pieces of furniture or new decorations.

Disorder causes ***additional stress*** in that it ***drains energy***, causing a feeling of fatigue. Of course, an organized, clean and ventilated space has the opposite effect, meaning it lowers stress, energizes and helps the person feel comfortable. In addition, the more clean the house gets, the less germs will multiply. Constant disinfection of objects, dust removal and house ventilation are simple gestures that keep the house clean, but it also takes into account the health of the person.

Moreover, maintaining order and cleanliness in the house allows time to be organized in a more efficient way. If order and cleanliness are maintained daily, a few tens of minutes, instead of a whole day a week, the person saves time, which he can dedicate to other activities.

Free time management

Free time management is very important when it comes to organizing the activities according to their importance, as well as according to the independence of the activities. In other words, activities that depend only on the person concerned and mandatory activities, which depend on other people, such as teachers, doctors, etc.

Beneficiaries must be able to distinguish between important, necessary or even mandatory activities, such as: going to school/work, going to the doctor, therapy/recovery hours, etc. and the relaxation activities such as playing, reading, watching a movie. After completing the module, the beneficiaries must be able to categorize their activities according to their importance and priority.

5.3. Health management

In addition to day-to-day management, the beneficiary needs to know health management methods: how to physically develop, how to maintain personal hygiene, how to eat healthy, as well as the important aspects of his/her treatment.

Physical development

Developing physical abilities in people with disabilities involves the development of some abilities, which will contribute to the improvement of the general physical condition of these persons or of certain aspects such as: training the muscular system, reducing spasticity, scoliosis etc., all of which are strictly necessary for the social and psychological integration of the beneficiaries and, last but not least, for the adoption of a more independent way of life. Thus, these people will feel useful to society and to themselves, becoming citizens active in solving community problems and integrated personalities. Developing physical abilities in people with disabilities is a continuous process of study and training. It is a process by which beneficiaries are helped to:

- Exercise different forms of physical education during rest, study, or service time;
- Practice various exercises to develop the muscular system and to correct the sphere of movement.

Personal hygiene

Personal hygiene aims at maintaining a clean state of the body, of the clothing and of the environment. Proper personal hygiene is the first step towards a healthy life because it includes practices that prevent the occurrence and spreading of diseases.

Poor hygiene affects the person's health, but also the relationships with other people, by putting them aside. Beneficiaries need to know how to use hygiene products (for example toilet paper, soap, shampoo, brush, comb, toothbrush, toothpaste, dental floss, deodorant, sanitary napkins/packs, shaving utensils) how to wash their body and hands after using the toilet, and also how to properly use the hygiene products to keep the hair, teeth, nails and body clean.

Healthy diet

A healthy diet means eating various foods belonging to the basic food groups: proteins such as meat, eggs and vegetables; dairy products; fruits and vegetables; cereals as well as bread and pasta; fats and sweets. Although it seems simple, it is not always easy to establish a proper diet for each individual. Sometimes, beneficiaries eat more from one (preferred) food group avoiding other food groups or opting for convenience over quality.

A healthy diet requires a plan, a goal, and the effort to include a variety of foods in the meals. Most people do not consume enough nutrients because they do not include in the meals a corresponding amount from each food group.

So it is important to pay attention not only to what is being consumed, but also to what is removed from the diet. For this, it would be advisable to develop a diary to record everything that a person eats and drinks during a week.

Attention should be paid both to the quantity of food one eats and to the food groups it belongs to. It is not necessary to eat a minimum amount of food each day from each food

group, but to eat the recommended amount over a week. Some small changes are enough to balance the diet. In most cases, many important nutrients are removed from diet.

Treatment

The management of treatment consists in the beneficiary's awareness of the importance of administering the treatment correctly, in understanding the importance of medication administration. It is also necessary for the beneficiary to understand that the correct administration of the treatment, as well as the observance of the medical visits and of the advice from the physician are vital for the management of his/her own health.

5.4. Money management

How much we spend, how much we save, what are the things we need and what are the things we can buy are decisions we take almost every day.

Budget Management

The service identified by the case manager must be effective in explaining to the beneficiary what money means, what its role is and how it is won. To facilitate the learning process, it is possible to use interactive shopping games in which the beneficiary has to give money for toy to receive the desired objects, to teach him/her the idea of exchange underlying commercial relations and to convey the idea that for every object he/she wants from the store, money is needed.

When the beneficiaries have understood these concepts, they can move to the next level of the training, in which the beneficiary aims to understand the advertising mechanism and learn to distinguish between needs and desires, among the products that make him/her more harm than good and, above all, how to resist the impulse to spontaneously buy various products. In this respect, the case manager can consult together with the pacient various money management guides or other valuable tools that simply explain to the beneficiary the importance of managing their money.

Savings management

The beneficiary needs to know the concept of "saving". It is necessary for the beneficiary to be helped to understand that when he/she wants a more expensive object, he/she can gather a part of the amount of money, by saving from his/her income or by earning extra money from various small services rendered to other people he/she can accomplish according to the skills or abilities that he/she possesses.

An effective win-win exercise can be implementing a savings program to start with the beneficiary, trying to improve himself/herself the ability to manage money, in order to have a model that the beneficiary can follow. Personal experiences can be shared with the beneficiary to motivate the beneficiary to maintain the program (examples: Tell him/her how you decided to buy a cheaper product, or how you decided to postpone a particular purchase or how long it took you to save enough money for what you wish).

5.5. Social development management

Personal development includes activities and experiences that have as ultimate goal to improve awareness, to develop talents and personal skills, to improve the quality of life, and to help achieve personal aspirations and dreams.

Communication

Communication is a set of actions that have in common the transmission of information in the form of messages, news, signs or symbolic gestures, written texts, etc. between two people, called interlocutors, or more formally, transmitter and receiver. The term of communication is related to our existence as people, then to society, because human beings and communication are interdependent. It is a dynamic process, in a permanent transformation. Society exists because of communication. Society means community and it is seen as a process involving participation from the members of the society.

Interpersonal relationships

Interpersonal relationships can be defined as those social relationships between two or more people generally characterized by "minimal use of stereotypical labels, the existence of unique and idiosyncratic rules (…) and a high degree of information exchange". In general, interpersonal relationships are between two or more people who are in proximity positions in which they interact or influence each other.

Interpersonal relationships establish values, rules of conduct, patterns of behavior, interaction styles. Psychologists believe that interpersonal relationships are closely related to happiness, to physical and mental health. Relationships are very important for any of our daily activities, but they can also be a source of psychological tension and unhappiness when conflicting. Friendship, love and marriage, relationships with parents and children are the main types of interpersonal relationships.

Living together the events plays an important role in the development of human relationships, has a "contagious" character, systematically cultivated and channeled by social practices.

The need for affiliation

People cannot live isolated from each other. The need for affiliation refers to the need of people to have frequent contacts with others. It forms the basis of the development of interpersonal relationships.

The need for privacy

A useful distinction for the study of interpersonal relationships is that between the need for affiliation and the need for privacy. The former refers to the desire to establish as many social contacts as possible, and the second refers to the preference for warm and close relationships.

Loneliness

In everyday life we are inclined to think that loneliness means the lack of social relationships. We are alone when our relationships with others are very few and inadequate. Lonely individuals are usually shy and have a low self-esteem. They are very often

depressed, have negative opinions about others and are socially cautious: they avoid interpersonal situations in which they may be rejected by others.

Interpersonal attraction

The term attraction refers to a motivational state: the desire to interact, to have relationships with a certain person. It is the opposite of the term rejection (the desire not to initiate a relationship or to end a relationship) and different from indifference.

5.6. Personal development management

The concept of "personal development" includes various activities that contribute to our improvement as people, activities from working out to reading books about relationship, socializing, love, communication, creativity and any other field that touches man as a being.

Career and career guidance

Professional orientation is the activity by which a person can achieve a real and complete picture about himself/herself, about his/her own person in order to work, to perform an activity that offers satisfaction. In this way he/she has a high efficiency and productivity at his/her work.

The case manager should help the beneficiaries understand the methods by which they can find out which profession is closest to their personality so that they can integrate into the labor market but also find a job to like. For this, a career or a professional orientation test can be done.

Once a career or professional orientation test has been carried out, the beneficiary will find out which are the main trades that are closest to his personality. Following such a career or professional orientation test, the beneficiary will also receive an assessment of his/her interests finding out what his/her dominant interests are.

The interests are identified following professional orientation assessments, reflecting the types of activities that please us. For example, if the beneficiary likes to sell things or services, then he/she will be happy to work as a consultant/sales representative gaining also professional satisfaction.

In general, many people confuse the interests with the abilities of a person, but there is a very big difference between a person's interests and abilities. Following professional orientation meetings, it is possible to identify the dominant interests and the skills developed over time. Interests tell us what we enjoy, and skills what we can do, the experience, the knowledge or the training we have accumulated in a certain area. The more the interests and skills overlap, the more we are satisfied with what we do.

In life, interests have a continuous dynamics and there are certain factors that can influence them, so it is recommended that they be periodically analyzed through professional orientation tests. They are also an important source for getting to know yourself better.

5.7. Legislation – European & Romania legal frame as refferance

In **ROMANIA** the rights of persons with disabilities to social care are regulated by Law no. 448/2006 (updated in 2016) on the protection of persons with disabilities, republished. It is granted in the form of ***social benefits*** and ***social facilities*** to persons holding a valid certificate of disability degree.

The right to social care in the form of social benefits is granted ex officio to persons who have certificates of disability degree and who are already registered in the institution ***or*** on request for persons who submit their file for the first time. The right to social care in the form of social facilities is granted on request.

Law no. 448/2006 (updated in 2016) on the protection of persons with disabilities, regulates the rights of persons with disabilities in various fields such as:

- health and recovery;
- education;
- house;
- culture, sports, tourism;
- transport;
- tax incentives;
- social services and benefits;

In addition to the rights provided by this law, persons with disabilities have a number of exemptions, benefits and facilities, regulated through different decisions or articles of other ordinances, laws that refer to persons with disabilities.

The case manager can consult a legislative basis, where it is necessary to direct the beneficiary towards different services or facilities to which he/she is entitled.

Legislative basis:

Law 448/2006 on the protection and promotion of the rights of people with disabilities - republished (updated in 2016);

Decision no. 268/2007 approving the Methodological Norms for the enforcement of the Law 448/2006 on the protection and promotion of the rights of persons with disabilities;

Decision no. 89/2010 of 05/02/2010 amending and supplementing the Methodological Norms for the application of the provisions of Law no. 448/2006 on the protection and promotion of the rights of persons with disabilities, approved by the Government Decision no. 268/2007;

Order no. 707/538/2014 amending and supplementing the annex to the Order of the Minister of Labor, Family and Equal Opportunities and of the Minister of Public Health no. 762/1992/2007 for the approval of the medical-psychosocial criteria on the basis of which the degree of disability is established;

Law 263/2010 on the unitary pension system (updated in 2016);

Decision 257/2011 approving the Norms for the application of the provisions of Law no. 263/2010 on the unitary pension system;

Order no. 1.418 of October 11, 2010 on the approval of the Procedure to establish the disability degree in order to register for disability pension and of the Procedure to verify the disability degree;

Decision no. 155 of February 23, 2011 for the approval of the criteria and norms of clinical diagnosis, functional diagnosis and assessment of the work capacity which set the I, II and III disability degree;

Law no. 76 of January 16, 2002 on the unemployment insurance system and on the stimulation of employment (updated in 2016).

Methodological Norms of February 20, 2002, implementing Law no. 76/2002 on the unemployment insurance system and on the stimulation of employment.

Government Decision no. 680/2007 for the approval of the Methodological Norms on the granting of free inter-urban transport to persons with disabilities;

Decision no. 884/2009 for amending and supplementing the Methodological Norms regarding the granting of free inter-urban transport to persons with disabilities, approved by the Government Decision no. 680/2007.

Order no. 62/2007 of 23/04/2007 for the approval of the Instructions on the permit for urban transport with means of surface transport for persons with disabilities and its model.

Order no. 223 on the implementation of the unique format of the parking card for people with disabilities.

The EU has a strong mandate to protect and promote the rights of persons with disabilities and to improve their social and economic situation. The Charter of Fundamental Rights of the EU states that "the EU recognises and respects the right of persons with disabilities to benefit from measures designed to ensure their independence, social and occupational integration and participation in the life of the community". It also prohibits any discrimination on the basis of disability.

Since January 2011, the EU is a party to the **UN Convention on the Rights of Persons with Disabilities -** the first legally binding international human rights instrument to which the EU is a party. This represents a strong European commitment to promote, protect and ensure the full and equal enjoyment of all human rights by all persons with disabilities. Its articles provide clear guidance on the steps that are to be taken to ensure that development policies and programmes in any sector area (eg. education, health, justice, employment) are inclusive of persons with disabilities.

In order to help implement the Convention, the Commission adopted the **European Disability Strategy 2010–2020,** which focuses on eliminating barriers in eight main areas: accessibility, participation, equality, employment, education and training, social protection, health, and external action. With regard to external action, the Strategy aims to promote the rights of persons with disabilities, including in its development programmes and humanitarian aid and in international fora.

In the context of the 2012 Strategic Framework on Human Rights and Democracy, **the EU Action Plan for Human Rights and Democracy (2015-2019)** focuses on a number of priorities and key human rights challenges. Disability will be specifically addressed under the challenge "Cultivating an environment of non-discrimination".

In November 2016 the European Commission put forward its proposal for a new European Consensus on Development, a blueprint for aligning the Union's development policy with the 2030 Agenda for **Sustainable Development**. In order to ensure that no one is left behind, the proposed Consensus reiterates the EU's commitment to a rights-based approach to development cooperation. It also stresses the commitment to promote the rights of persons with disabilities and work to ensure their participation on an equal basis.

EU framework focused on rare diseases:

Communication on rare diseases: Europe's challenges COM (2008)

http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf

Recommendation on an action in the field of rare diseases (2009/C 151/02)

https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

6. Personalized intervention procedures / working methodology

6.1. Working Methodology in case management of persons with rare diseases and their families

Case management of persons with rare diseases is the set of techniques, procedures and working tools that provide advice to beneficiaries to improve their quality of life. (In the professional terminology of the case management, the professional who offers advice is called case manager**, and the person with a rare disease, or the beneficiary of the services, is called** client**).**

This is done through 3 well-defined stages: the initial assessment phase, the intervention phase and the final assessment stage.

Starting from the psychosocial dynamics of the **client, in the given situation - the person with a rare disease, the case manager** defines a set of objectives and means appropriate to the process of social change he thinks necessary, building in this respect an intervention project.

For a more complex assessment of the situation of the person with a rare disease, the case manager will use psychosocial investigation methods and techniques such as: the observation method, the documentation technique and the interview methodology.

The information obtained through the observations, supplemented with the data gathered through interviews and the analysis of the documents, will provide the case manager with the necessary material to investigate the case and clarify the client's problems in order to design a specialized intervention.

Given that the interview will be used at all stages of the case management, we will outline below how it can be used throughout the process.

Through the initial interview we will review whether the person with a rare disease needs the professional case manager intervention. In this case, the potential client, the person with a rare disease, will become a real user of the counseling service.

Each interview tends to have *a direction and a structure* (beginning, middle, and end). So the case manager needs to reflect on each interview and always review the content of the interview.

The introductory phase of the interview - involves questions or discussions of a general nature, for an emotional preparation of the client - the person with a rare disease. The most important skills are the ones to ask questions (open or closed) and to listen actively.

In the introductory phase, the questions follow:

- the exploration of the problem and of the client's situation;
- the client socialization in the role of the interviewee;
- the encouragement of the client to share factual and emotional information;
- assistance to the client to feel respectable and comfortable in the interview relationship.

The middle phase of the interview - the case manager will explain the purpose and objectives of the interview. The following skills are required:

- to explore and accompany (nonverbal and verbal);
- to orientate towards change;

- to observe (verbal language, client's non-verbal language, visual contact, avoiding sensitive topics for the client, observing inconsistencies or shortcomings, stressful or conflicting circumstances);
- to listen (active, passive, etc.);
- to ask questions (closed, open questions, guiding questions, follow-up questions, approving questions);
- to focus, guide and concentrate;
- to guide the client to achieve the proposed objectives;
- to paraphrase;
- to summarize;
- to confront;
- to silence;
- to develop the climate.

Ending the interview – takes place when the client has overcome the problem or the professional cannot work with him/her. The case manager will have to have the skills to prepare the client for this step, to summarize, to know how to identify the clients who express their concerns only at this stage.

In the interview, the case manager needs to make sure that he is understood by his client; the case manager will have a professional behavior, will respect the confidentiality of the information provided during the interview.

Successful interviews do not depend on the content (what was said) or whether or not the client has obtained what he/she has asked for. A significant outcome for the first interview is that the case manager is perceived as someone able to understand the client's concerns and feelings about the difficulties he/she is going through.

The success of the case coordination is based on the focus on the details of the interaction between the one who helps and the one who is helped. For this**, counseling occupies a pivotal place.**

The British Counseling Association defines the concept of counseling as "the fact when a person regularly or temporarily acts as a counselor, explicitly offers and accepts to give time, attention and respect to another person who will be temporarily in the role of the client" (Membership Notes, 1990). The task of counseling is to give the client the opportunity to explore, discover and clarify his/her ways of living with more resources and greater welfare.

Case managers need to be able to **listen, observe, and respond**. For this they need specific abilities: skills to wait, to specify, to confront, to personalize, to solve problems and to plan actions.

According to Nelson Jones (1983), counselors must have the following qualities:

- empathy or understanding, the effort to see the world through the eyes of the other;
- respect (trust in the other's ability to solve the problem);
- objectiveness or specificity (the person being counseled to be able to reduce his/her confusions);
- self-knowledge and self-acceptance;
- authenticity in the relationships with the clients;
- congruence (the verbal language fits the nonverbal one).

The aim of counseling will be to help people in the development process, to help the individual become a person with full functionality, self-confidence, to develop an internal evaluation resource.

The conditions for conducting good counselling are **empathy, non-possessive "warmth", acceptance** and **authenticity.**

Empathy can be defined as "the entry/the stepping" into the inner world of another person in order to understand the thoughts, feelings, behaviors and meanings that the person attributes to certain events. At the same time, the counselor remains himself. Empathy does not mean identifying with that person. The level of empathy is closely correlated with the degree to which the client manages to explore his/her inner world.

The non-subjective "warmth" is expressed in general by: body language (posture, proximity, personal space, visual contact), words and speech (tone, word type, etc.). Any mismatch of words with non-verbal language creates confusion. The warmth and the proximity should be used with the utmost care. Someone very "cold", distant, cynical and distrustful can feel threatened in front of a person who looks "warm" and affectionate. Warmth and proximity are the attributes that facilitate the formation of the working alliance between the client and the case manager.

Authenticity is the degree to which the counselor is himself in the relationship with the client.

As the basis of communication, authenticity is at the same time a precondition for empathy that encourages the client to explore himself/herself and to be, in turn, honest and open.

Acceptance is another essential condition in counseling. It contains implicitly another condition, namely that of recognizing the uniqueness of the person. Acceptance is the framework for change. Counselors generally have different degrees of acceptance that may vary from day to day or from client to client. If we accept people as they are, in turn, they will accept us as we are.

6.2. Work tools used in case management of people with rare diseases and their families

Given that case management is carried out through the Romanian Prader Willi Association, the case managers will use part of the working tools validated by the association and registered through the ISO 9001:2015 procedure and some of the case management documents, but adapted to the needs of the project INNOVCare.

6.2.1. Initial evaluation stage

The case manager will fill out a ***Record Sheet*** and attach copies of the beneficiary's relevant documents (medical letter from the specialist doctor, documents proving the diagnosis, disability degree certificate and identity documents of the beneficiary and whether it is the case of the other family members/legal representative. When completing the record sheet, the written consent for the processing of personal data will be required.

Contract for the provision of social services. It will be filled out for a determined period of 9 months and will be signed by the beneficiary/representative of the beneficiary and the President of APWR. One copy is handed over to the beneficiary/representative of the beneficiary, and the second copy is attached to the Beneficiary's file.

Social Investigation - The role of the case manager is to evaluate and identify the needs of the person with a rare disease. This is done based on the *Social Investigation* that

can be done at the beneficiary's home or within the NoRo Center on the basis of the discussions with him/her.

This involves both collecting general client information (identification data, civil status, health status, family structure, etc.) as well as specific information about the medical, social history; information about the family and community system, about resources that may be involved in solving the case.

The social investigation will be focused on the delimitation of the problem, its awareness by the client, the identification of personal/family/community resources and the responsibility of the client for the involvement in the intervention process and the choice of the types of services that the client will benefit from.

6.2.2. Intervention stage

Following the analysis of the information obtained during the initial assessment stage, the case manager together with the person with a rare disease/his/her legal representative develops **the action** (intervention) **plan.**

The action plan contains the objectives, activities, deadlines and the persons responsible with carrying out the proposed activities. These will be recorded in the document and will be reviewed as often as needed.

The way in which the objectives proposed in the action plan will be met, together with the time dedicated to the activities carried out in this respect, will be recorded in the **meeting reports/action reports.** The meeting reports will be completed whenever a case manager meets with the client, and the action reports will be completed by the case manager whenever he/she takes an action in the client's interest (makes an appointment for a consultation, looks for a procedure for access to a particular type of service, communicates to the client the results obtained by telephone or vice versa.

6.2.3. Final assessment stage

The closing of the case is documented in the *Case Closing Sheet*, where the case manager records the manner and cause of the case closing, the case status at the end and the date of the closing.

Closing the case can be done under the following conditions:

- At the end of the contractual timeframe;
- If the client quits the counseling service for various reasons and refuses to collaborate with the case manager;

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Annexes

Annex 1 – Rare diseases

Duchenne Muscular Dystrophy (DMD)/Becker (DMB) (Code ORPHA- 262)

Clinical definition

Duchenne muscular dystrophy (DMD) is a genetic disease characterized by the degeneration of the striated muscle fiber that causes a progressive atrophy of most muscle groups, resulting in the installation of a variable motor disability depending on the shape and stage of evolution.

The disease has been known since 1868 for its rapidly evolving form (Duchenne) and since 1955 for its slow evolving form (Becker).

Clinical signs

The onset average age is 3 years old for the fast evolving form (Duchenne) and 13 years old for the slow evolving form (Becker). All intermediate stages can be observed. This onset translates into disorderly, difficult, tedious walking with frequent falls, different from the previous walking. Eventual running capacity will also be quickly lost. At this stage, molet hypertrophy can already be observed. Progressive difficulties arise in climbing stairs, walking becomes hesitant, with the risk of falling, difficulties in maintaining balance and in reclaiming from one position to the next. To limit the falls, the patient reduces his walking speed and adopts a cranked position that allows balance to be maintained (around the age of 9 for Duchenne). In the case of Becker's myodistrophy, the loss of ability to move occurs later (after the age of 30). After the loss of the ability to walk, the sitting position is marked by functional problems related to trunk static and to the difficulties associated with hand-mouth movements to eat, and to the alteration of the vital functions (respiratory, cardiac, digestive).

Becker muscular dystrophy occurs in various clinical forms. The onset age and the evolution of this disease are very varied. The weakening of the belt muscles and the hyperatrophy of the muscle mass are often not obvious. The severity of Becker's myopathy is related to a possible failure of the heart muscle due to heart failure and heart rhythm disorders. Walking disturbance may remain moderate over several years: loss of walking occurs before the age of 40 in only 50% of the patients. The evolution of the disease is very variable from one individual to another and even within the same family. There are many cases when the disease is atypical, being accidentally discovered, revealed by the increased muscle enzymes in plasma. Some "benign" forms only manifest through cramps that occur during effort. These forms may evolve over time to a cardiomyopathy that is independent of the severity of the muscle damage.

Hemophilia A and B (Code ORPHA – 448)

Clinical definition

Hemophilia A is a hereditary congenital disease, inherited as an X-linked recessive trait characterized by diminished quantitative or qualitative altered synthesis of factor VIII of coagulation (fVIII).

Clinical signs

Although it is a congenital disease, the hemorrhagic manifestations rarely occur in the neonatal period (6% of cases), the diagnosis usually being given after infancy (32% of cases) and in 30-40% of the cases at the adult age. 1-2% of newborns with haemophilia may have intracranial bleeding; the cephalhematoma may be suggestive in the newborn with a positive

family history. Clinical manifestations are more obvious once the child becomes more active and more exposed to the trauma.

The most common hemorrhagic manifestations are:

• Intra-articular bleeding (haemarthrosis) is by far the most characteristic manifestation for haemophilia; superficial muscular hematomas,

• abundant and repetitive haematuria, involving renal colic in 20% of patients,

• postoperative bleeding or bleeding after dental interventions, in the ENT sphere,

• digestive bleeding, pointing a ulcerative disease; meningo-cerebral bleeding,

• spontaneous bleeding in the interfascial space of abdominal muscles, especially in the ileophoid muscle, with frequent retroperitoneal and intraperitoneal extension

• prolonged bleeding at the venepunction place or the formation of hematoma at the place of the intramuscular injection.

Oculocutaneous albinism (OCA) (Code ORPHA – 998)

Clinical definition

OCA comprises a group of hereditary diseases characterized by pigmentation deficiency of the skin, hair and eyes.

The cells that synthesize pigment have two different origins. The cells from the retinal pigment epithelium, the anterior and posterior iris epithelium and the external ciliary epithelium originate from the prior primitive neuroectoderm, and the melanocytes from the irioma, ciliary and choroid stromal originate from neural crests. These types of cells can synthesize melanin starting from an amino acid - tyrosine. In this process of synthesis of the melanin pigment, a complex enzyme equipment is involved, which has as an essential component an enzyme called tyrosinase. The mutations occurring in the genes encoding this enzyme complex, and especially in the gene encoding tyrosinase, can lead to pigment deficiency in the skin, hair, and eye, so to various forms of albinism.

Depending on the gene in which the mutation occurs, there are four forms of oculocutaneous albinism, called OCA1, OCA2, OCA3 and OCA4.

Clinical signs

The four forms of oculocutaneous albinism have in common the hypopigmentation of the skin, hair and eyes, but differ in pigmentation degree, severity of the disease and the association with other symptoms.

OCA1, has beside the common elements of albinism, nystagmus, reduction of iridial and retinal pigment, foveal hypoplasia associated with reduced visual acuity, strabismus and reduced stereoscopic vision. The teguments are very depigmented (translucent) and the iris is light blue.

OCA2 is a form with variable degrees of pigmentation. The teguments may be light cream, and the hair may be light yellow, blond or brown. The degree of the iris and retina pigmentation, as well as the visual acuity is superior to OCA1.

OCA3, occurs more commonly in black people, being a "reddish" albinism. The synthesis of melanic pigment is more effective than in forms 1 and 2, which results in milder forms of albinism and visual anomalies in relation to the other types of albinism.

OCA4 has similar manifestations to OCA2, of which it is genetically different, but is more common in the Asian population.

Down Syndrome (Code ORPHA – 870)

Clinical definition

The most common and known genetic disease, produced by trisomy 21, is the first cause of mental retardation.

The incidence of trisomy 21 was estimated at 1: 650 breathing newborns.

The disease is more common in male children, the sex ratio being M:W 3:1

The newborn with trisomy 21 has less weight and waist than the gestational age parameters, muscular hypotonia, and reduced Moro reflex.

Conclusive physical aspect with brachycephalic head, flattened occiput, broad fontanels, round and flat face, epicanthus, oblique palpebral fissures, small nose with flattened root and small anteversate nostrils, macroglossia with lingual protrusion, implanted lower ears, short neck, short and flat hands, with brachydactylia and simian fold.

Clinical signs

The cytogenetic analysis is essential and mandatory in each case.

The cariotype can highlight:

Free and homogeneous Trisomy 21 (92-95%) - meiotic nondisjunction most common maternal (85-90%)

Trisomy 21 in mosaic (2-3%) - mitotic nondisjunction

Trisomy 21 due to robertsonian translocation between chromosome 21 and an acrocentric chromosome (4-5%)

Partial Trisomy 21 (1%) - partial duplication of the long arm of the chromosome 21.

Bartter Syndrome *(Code ORPHA – 112)*

Clinical definition

Bartter Syndrome is a rare inherited defect of Henle's ansa thick ascending branch from the glomerulus. It is characterized by low levels of potassium, low blood acidity - alkalosis and normal blood pressure. There are two types of Bartter syndrome: neonatal and classic.

Clinical signs:

In 90% of the cases the Bartter's neonatal syndrome is observed in 24-30 weeks of gestation with amniotic excess - polyhydraminos. After birth the child drinks and urinates excessively (polyuria and polydipsia). Life threatening dehydration may occur if the child does not receive enough fluids. Approximately 85% of children have excess calcium in urine (hypercalciuria) and kidneys (nephrocalcinosis), which can lead to renal lithiasis. In rare cases the child can progress to kidney failure.

Patients with classic Bartter syndrome may have symptoms in the first two years of their life, but are usually diagnosed at school age or later. Adults may experience polyuria, polydipsia and tendency to dehydrate. Those patients who develop vomiting develop physical retardation. Renal function is normal if the disease is treated, but occasionally it can end up with end-stage renal disease.

Phenylketonuria-PKU- (Code ORPHA – 716)

Clinical definition

Phenylketonuria (PKU) is a genetic metabolic disease in which the phenylalanine hydroxylase (pha) enzyme is missing or has a very low level in the blood. This enzyme is required for the metabolism of phenylalanine, an amino acid from food.

If PKU is not diagnosed and treated immediately after birth, phenylalanine accumulates excessively in the blood stream, where it is partially oxidized to phenylpyruvic acid, with a particularly toxic effect on brain tissues, leading to mental retardation and central nervous system damage.

Clinical signs

The symptoms of phenylketonuria usually occur within a few months after birth, in parallel with the rise of phenylalanine in the bloodstream of the baby, coming from the mother milk proteins or from the milk formulas.

Early symptoms of Phenylketonuria:

- the mold smell of skin, hair and urine;
- vomiting, diarrhea, irritability;
- skin drying, eczema, sensitivity to light.

The symptoms may become severe around the age of 4-6 months:

- tremors, hypertonia;
- growth and development retardation;
- episodes of epileptic attacks.

Approximately 90% of children with PKU have blond hair, pale skin and blue eyes.

Without establishing a poor diet in phenylalanine, children with PKU develop severe, profound, irreversible mental retardation.

Prader Willi Syndrome (Code ORPHA – 739)

Clinical definition

Prader Willi syndrome is a chromosomal abnormality secondary to the absence (deletion) of genes on chromosome 15 inherited from the father, or to the maternal disomy of chromosome 15 (both chromosomes 15 being inherited from the mother).

The characteristic features of Prader Willi syndrome are neonatal hypotonia, low stature, small hands and feet, obesity, mental retardation and hypogonadism.

Clinical signs

- severe hypotony during the infant period;
- feeding difficulties during the first months of life;
- excessive feeding;
- (hyperphagia = absence of satiety);
- progressively developed obesity;
- cognitive deficiencies;
- particular behavioral phenotype;
- hypogonadism;
- small stature.

Williams Beuren Syndrome (Code ORPHA – 904)

Clinical definition

Williams Beuren Syndrome is a rare chromosomal genetic disease characterized by:

- distinct facial features;
- moderate mental retardation;
- particular cognitive profile;
- cardiovascular abnormalities;
- elevated calcium in the blood (hypercalcaemia) and/or in urine (hypercalciuria).

Clinical signs:

- small height;
- special facies, with elfish look;
- starlike iris;
- thick lips, small and spaced teeth;
- systemic hypertension;
- supravalvular aortic stenosis;
- pulmonary stenosis, renal artery stenosis;
- harsh, thick voice;
- Medium IQ 56, very sociable behavior;
- hyperacusis;
- hypercalcemia, hypercalciuria.

Turner Syndrome (ST) (Code ORPHA – 881)

Clinical definition

Turner Syndrome is a chromosomal disease caused by complete or partial absence of chromosome X in all cells of the body or only in some cells.

The disease can be diagnosed as early as childhood, characterized mainly by smaller height than the one corresponding to the chronological age and *pterygium coli* (cutaneous fold on the lateral sides of the throat).

After puberty, the clinical diagnosis is much easier, based on the triad: hypo-height, primary amenorrhea (absence of menstrual cycles), and poor female secondary sexual characteristics.

Clinical signs at birth:

• female child shorter than normal;

• lymphadem (subcutaneous lymph accumulation) in the hands and feet;

• short neck with excess skin on the neck and/or *pterygium coli* (skin fold on the sides of the neck);

• large intermamelonary distances.

Clinical diagnosis during childhood is based on the identification of a major growth deficit, the child's height being much lower than the mean value corresponding to biological age.

Post-puberty clinical diagnosis of Turner syndrome is suggested by the following clinical signs:

• small height - less than 145 cm high;

• primary amenorrhea (lack of menstrual cycles);

• primary and definitive sterility (unable to get pregnant) because the ovaries do not produce eggs;

• poorly developed mammary glands;

• absent axillary pilosity;

• reduced pubic pilosity;

• external genitals (vulvar lips, clitoris) with infant appearance;

• uncharacteristic cranio-facial dysmorphia (mature aspect of the face, triangular facies, epicanthus, antimongoloid palpebral apices, ogival palatine arch);

• low hair insertion line on the neck, finished in *trident*;

• biacromial diameter greater than the bitherohrterial one (shoulders broader than the hips);

• *cubitus valgus* (outwards deflection of the forearm in relation to the arm);

• convex hypoplasic nails;

• numerous pigmental nerves (moles);

• auditory deficits due to internal ear abnormalities that favor recurrent middle ear infections;

• congenital visceral cardiac or renal malformations (30-40% of cases);

• intelligence is normal or at the lower limit of normal, with a decrease of the spatial perception and of the abstraction capacity.

Multiple myeloma (Kahler Disease) (Code ORPHA – 29073)

Clinical definition

It is a malignant proliferation, of unknown origin, of the plasmocites in the bone marrow. Multiple myeloma generally develops in people over the age of 60 years.

An abnormal increase in the production of a single type of immunoglobulin for a particular myeloma is observed in the patient's blood. Plasmocites secrete substances that gradually lead to the destruction of bone tissue. The disease can be found due to rebellious bone pain, anemia, or due to a significant increase in sedimentation speed, being able to exceed 1-00 mm per hour.

Clinical signs

The symptoms of the multiple myeloma depend on how advanced the disease is. In the early stages of the disease there can be no symptoms. When the symptoms occur, patients usually accuse bone pain, usually at the level of the ribs and back. Patients may also have bone fractures, weakness, tiredness, weight loss or repeated infections. When the disease is advanced, the symptoms may include nausea, vomiting, constipation, urination problems, tiredness and numbness in the lower limbs. These are not safe signs of multiple myeloma; they can be symptoms of other medical problems.

Multiple Sclerosis (Code ORPHA – 228145)

Clinical definition

Multiple Sclerosis (MS) is a chronic, immune-mediated disease that affects the central nervous system (CNS) - including the brain, spinal cord and optic nerves. Although it occurs most often in the young adult, it is known that MS occurs at the beginning of childhood or after 60 years.

Clinical signs:

- Early symptoms of multiple sclerosis:
- Blurred or double view
- Thinking problems;
- Missing or lack of coordination;
- Loss of balance;
- Numbness;
- Tingling;
- Weakness in an arm or leg;

There are not two people who show the same symptoms of multiple sclerosis. You may have only one symptom and pass a long time before you notice others. But for some people, the symptoms worsen in a few weeks or months.

Common symptoms of multiple sclerosis:

Abnormal sensations. People who suffer from multiple sclerosis say they feel the presence of "needles" in the body. They also have numbness, itching sensation, burns, stabbing or severe pain. Approximately half of the persons with multiple sclerosis have these symptoms. Fortunately, they can be managed or treated.

Bladder problems. Approximately 8 out of 10 persons have bladder problems, but they can be treated. Frequent urination and constipation are common symptoms of the disease.

Difficulties in walking. Multiple sclerosis can cause muscle weakness or spasms, which makes walking much more difficult. Balancing problems and numb feet make walking difficult.

Dizziness. It is normal for persons who suffer from multiple sclerosis to feel dizzy, but they will not have vertigo and will not feel that the room is spinning.

Fatigue. About 8 out of 10 people feel very tired. The feeling of fatigue normally occurs around noon and causes muscle weakness and drowsiness, having nothing to do with effort.

Tonic spasms. These usually affect the leg muscles. For about 40% of those affected, tonic spasms are an early symptom of the disease. In progressive multiple sclerosis, tonic spasms affect 6 out of 10 persons.

Sexual difficulties. Both women and men with by multiple sclerosis are less receptive to the touch and have a reduced sexual appetite.

Speech problems. Sometimes multiple sclerosis can cause breaks in speech. In more advanced stages of the disease, some persons also develop swallowing problems.

Thinking problems. Approximately half of the persons with multiple sclerosis have problems concentrating. However, multiple sclerosis does not affect the intellect, the ability to read or understand a conversation.

Tremors. Tremors may be minor, but in some cases, may affect the daily activity of the affected person.

Vision problems. Narrow vision is, in most cases, among the first symptoms of the disease. It usually affects one eye. The vision becomes blurred, gray or with a dark place in the center.

Epidermolysis bullosa (Code ORPHA – 304)

Clinical definition

In fact, the term "epidermolysis bullosa" is inappropriate because "epidermal" lesions are observed only in some entities. In the other entities there are deeper, junctional or dermal lesions. The term continues to be used because it has long been established.

In fact, there is a heterogeneous group (almost 30) of vesicular hereditary disorders, which concern the skin but also the mucosas, especially the oral mucosa. Vesicles occur especially in the area where there has been a traumatism of the skin. For some heat is a favoring factor. Others manifestate by spontaneous vesicles.

Today, these disorders are classified in IV main types according to the level at which the cleavage explaining the bubble occurs, according to the most important clinical signs, to the mode of genetic transmission:

I. Intraepithelial, non-cicatricial forms with dominant autosomal, autosomal recessive and X-linked transmission.

II. Junctional forms, characterized primarily by atrophic skin phenomena with recessive autosomal transmission

III. Dermolytic, atrophic and cicatricial forms with dominant and recessive autosomal transmission

IV. Acquired, nonhcritable forms

Clinical signs

Dermolytic forms are almost exclusively due to dominant or recessive mutations in the gene for "collagen VII, alpha-1-polypeptides" (COL7A1) gene located in 3p21.3 region; some have unknown location.

In the junctional forms, there are remarkable autosomal recessive mutations affecting genes located in regions 1q31, 1q32, 1q25 - q31, 10q24.3, 17q11 - qter, 18q11.2, others have unknown location

Intra-epithelial forms are due to various, autosomal dominant, recessive or X-linked mutations of some genes located in regions 8q24, 12q13, 17q12-q21, Xq27.3-qter

The key clinical sign is:

• The vesicle, which may appear neonatal or later, in different regions of the body, localized or generalized, on the skin and/or on the mucosas, may be permanent or transient, can heal spontaneously or leave wounds, sinuses and scars, etc. All of these depend on the clinical form of the disease, but especially on the level at which cleavage between the layers of the skin occurs.

Myasthenia Gravis (Code ORPHA -589)

Clinical definition

The "invisible" disease, myasthenia gravis is a chronic neuromuscular disease that manifests itself through severe fatigue in repeating a certain movement and weakness of the volunteer muscles of the body; the very term "myasthenia gravis" is also referred to as "severe muscle weakness."

Clinical signs

The diseases called autoimmune, including MG, are characterized by the fact that the body produces "abnormal" antibodies or antibodies that ″go crazy″ and attack the normal body structures that are no longer recognized as its own: in the case of myasthenia gravis, abnormal antibodies are produced against some proteins in the junction between the nerve

and the muscles - called receptors, which are in this way blocked. The nervous impulse is no longer transmitted to the muscles and they do not contract anymore.

MG is considered a rare disease. The frequency of the cases is 1 per 10000 persons, more often women aged 15 to 30. Men fall sick more often after age 50, but practically anyone can make the disease, even young children.

Bardet- Bield Syndrome (Code ORPHA 110)

Clinical definition

Bardet-Bield syndrome (SBB) is a genetic disease transmitted autosomal recessive with different clinical expression from person to person, even among members of the same family. It affects the intellectual development (mental retardation), the vision (pigment retinopathy), the limbs (increased number of fingers-polydactyly), the kidneys and the sexual organs (hypogonadism) and it also manifests through obesity.

The syndrome was named after Georges Bardet, and Arthur Biedl. Georges Bardet was a Hungarian doctor practicing medicine in Vienna and Prague and who described the case of two sisters suffering from pigment retinitis, polydactyly, hypogonadism and obesity. Two years later, Arthur Biedl of the University of Paris finds the same symptoms in two other sisters unrelated to those described by Bardet.

A similar condition was originally called Laurence-Moon-Bardet-Biedl syndrome, including the names of two English physicians John Zachariah Laurence and Robert Charles Moon. Currently, Laurence-Moon syndrome and Bardet-Biedl syndrome are considered to be two separate entities, the first having paraplegia without polydactyly and obesity.

Clinical signs

In many cases, the diagnosis of SBB is delayed, probably due to insufficient knowledge of the disease, as well as due to the gradual onset of the symptoms related to pigment retinitis and renal dysfunction (which determine the visit to the doctor).

• Eye damage may occur early or may occur later in the course of the disease. It is estimated that approximately 98% of the patients have visual damage by the age of 30 years. Many children with SBB will have myopia or strabismus before the onset of the visual failure.

• Obesity usually occurs during childhood, even in the first year of life, and its severity increases with aging. SBB obesity is not responding to diet and is often accompanied by non-insulin dependent diabetes and hypertension.

• Extremities defects. Polydactyly occurs in over half of SBB cases and is a very important sign for the diagnosis of the disease but may not be present in every affected family member. In addition, other extremities malformations have been described: brachydactylia (short fingers), partial syndactylia (affiliated fingers) or clinodactyly (angled fingers).

• Mental retardation is variable, often it may be moderate. There are cases of SBB that are perfectly normal at the intellectual level.

• Difficulties in learning, mainly related to mental retardation and visual disturbances.

• The waist may be affected (small height).

• Urinary tract malformations. Kidney failure is the most important cause of illness in SBB patients. There are numerous abnormalities in the urinary tract: cystic tubular disease, urinary tract malformations, chronic glomerulonephritis.

• Neurological disorders such as ataxia with coordination and balance disorders.

• Behavioral disorders: emotional lability, frustration, obsessive-compulsive tendencies, difficulty in concentrating.

• Speech defects: nason voice, the modification of the first consonant of words, which leads to difficulties in understanding, to which the limited vocabulary used also contributes.

• Dental abnormalities such as small teeth, hypoplasical enamel and short dental roots.

• Asthma – the association with asthma is seen in some patients with SBB with a certain type of genetic mutation.

• Fertility. Typically, men affected by SBB suffer from infertility due to genitourinary tract malformations and hypogonadism.

• Diabetes mellitus. Approximately 4% of those with SBB have non-insulin-dependent diabetes mellitus as a result of the development of severe insulin resistance.

Concluding, the clinical signs of SBB are:

Primary (for diagnosis, the presence of 4 of the following signs is required):

• Retinopathy, Polydactyly, Obesity, Learning Difficulties, Hypogonadism in Males, Renal Abnormalities.

Secondary (3 primary and 2 secondary criteria are required):

• Speech disorders, strabismus/cataract/astigmatism, brachydactylia/syndactyly, growing disorders, polyuria, polydipsia (increased urine and ingested liquids), coordination and balance disorders, extremities spasticity, diabetes mellitus, dental disorders, congenital heart disease, liver fibrosis.

Mastocytosis (Code ORPHA – 98292)

Clinical definition

Mastocytosis is a rare haematological disease with an incidence of 0.3 cases per 100.000 inhabitants per year. The mast cells are part of the immune system. These are produced by the bone marrow.

Mast cells are located in organs such as the skin, the marrow, the liver and the spleen.

Patients with mastocytosis have an abnormal increase in mast cell at these levels.

They produce and store various substances such as histamine, which by their action produce the clinical picture of mastocytosis.

This disease is not hereditary, but it is not impossible to have several affected people in a family.

Clinical signs

The release of the mediators stored in mast cells leads to symptoms such as:

-pruritus; redness; diarrhea; indigestion; nausea; abdominal pain; muscle pain; bone pain; osteoporosis; hypotension; anaphylaxis; heart rhythm disorders (tachycardia/bradycardia); headache; concentration issues; asthenia; vertigo; irritability; depression and fear.

Depending on the level at which the accumulation of a larger number of mast cells has occurred, the World Health Organization has classified mastocytosis into two different classes:

- Cutaneous mastocytosis;

- Systemic mastocytosis;

Cutaneous mastocytosis: is an exclusively dermatological disease (the accumulation of mast cells is in the skin), benign. It occurs in childhood, and most cases disappear in puberty.

Three types of cutaneous mastocytosis are described:

Maculopapular cutaneous mastocytosis (Urticaria Pigmentosa):

-the most common variant;

- characterized by: pink or brown spots and/or vesicles on the body, not on the face.

- Telengiectasia macularis eruptiva perstans (TMEP) - a rare type of MCMP, characterized by a large pink persistent bulge on the adult body.

Diffuse cutaneous mastocytosis:

-appearance at birth;

- the skin is thickened and bladders appear easily;

- it is a very rare form of the disease;

Mastocytoma:

- little common and occurs in childhood;

- characterized by an isolated node or by multiple nodes.

Systemic mastocytosis:

- myeloproliferative disease;

-Mast cells proliferate in at least one organ, most commonly in the bone marrow;

- usually occurs in adults;

-not a hereditary disease.

4 forms of systemic mastocytosis are described:

Indolent systemic mastocytosis (MIS);

-the most common form;

-does not change over time;

- rarely advances to another subtype: marrow mastocytosis (does not involve skin damage); systemic latent mastocytosis (shows the highest accumulation of mast cells);

Systemic mastocytosis with a blood alteration.

Aggressive systemic mastocytosis:

- presents the poorly functioning of at least one organ, rarely the skin.

Mastocyte Leukemia:

- exceptionally rare haematological disease;

-It never affects the skin.

Crohn Disease

Clinical definition

Crohn Disease is a chronic inflammatory disease localized at the wall of the digestive tract, potentially affecting any segment from the esophagus to the anus, but especially the distal portion of the small intestine (terminal ileitis) and the colon, characterized by transmural ulcerative inflammatory lesions that can cause complications such as strictures or fistulas.

Crohn disease is the disease of the young adult (15 to 30 years), but can also occur at older ages, with the same incidence in women and men.

The cause of Crohn disease is unknown. There have been proposed genetic, infectious, immunological causes, but none have been proven firmly.

Clinical signs

The main manifestations of Crohn disease are:

• enhanced intestinal transit - soft feces, several times a day - weeks-months persistence (chronic noninfectious diarrhea);

• recurrent abdominal pain;

• rectal bleeding;

• persistent fever;

• alteration of the general state;

• joint pains - frequent lumbar pains (back pains);

• weight loss.

Juvenile Idiopathic Arthritis (Code ORPHA – 85438)

Clinical definition

Juvenile idiopathic arthritis (AIJ) is a chronic disease characterized by persistent inflammation of the joints; the typical signs of joint inflammation are: pain, swelling and limited movement. "Idiopathic" means that we do not know the cause of the disease and "juvenile", in this case, means that the symptoms usually occur before the age of 16.

Clinical signs

Arthritis should be present for more than 6 weeks in order to exclude temporary arthritis forms, such as those that may arise from various infections. The term AIJ includes all forms of persistent arthritis of unknown origin with onset in childhood.

AIJ includes various forms of arthritis that have been identified (see below).

The diagnosis of AIJ is therefore based on the presence and persistence of arthritis and on the careful exclusion of any other disease by assessing the medical history, the physical examination and the laboratory tests.

For more information, visit https://www.printo.it/pediatric-rheumatology/RO/info/2/Artrita-Idiopatic%C4%83-Juvenil%C4%83.