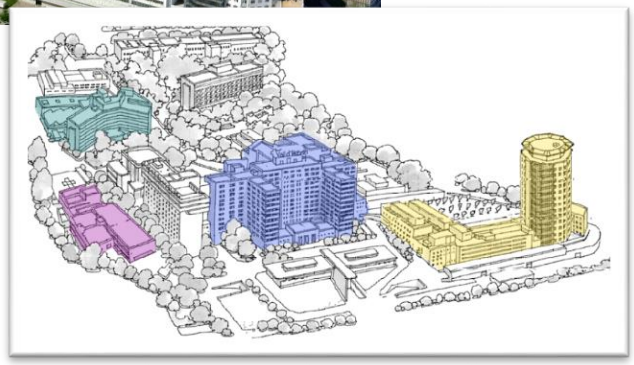
Promoting integrated holistic care for rare diseases
Center of expertise example

Eduardo Tizzano

Area de Genetica Clínica y Molecular y
Unidad Funcional de Enfermedades Raras
Grupo Investigación VHIR Medicina Genetica
Campus Valle Hebron, Barcelona

The University Hospital Valle Hebron is a hospital of high complexity at tertiary level in maternal-infant and adult patients and is one of the biggest hospitals of Spain and Europe

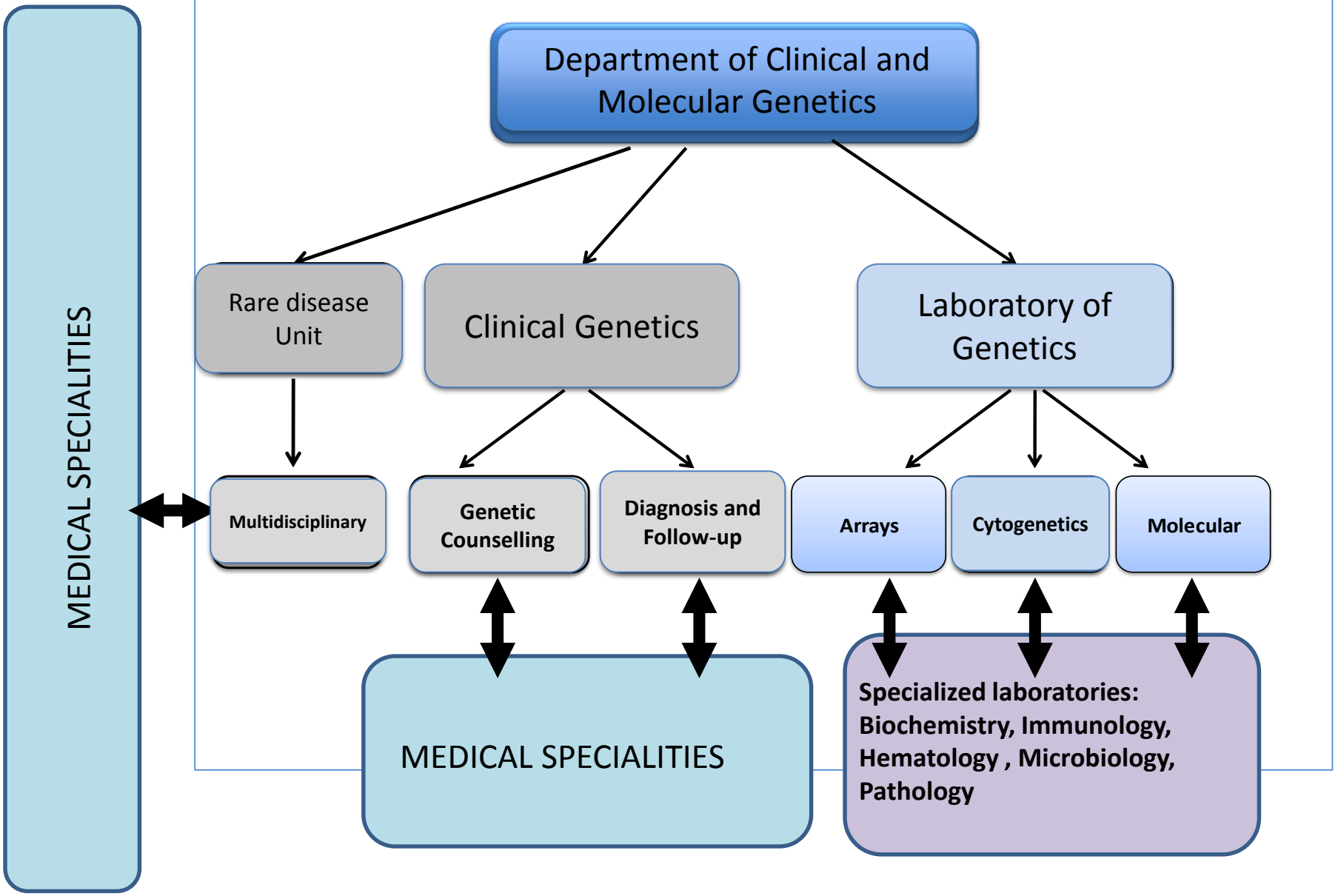


- **1952 |** Inaugurated in Barcelona;
- **2009 |** Research accredited by *Instituto de Salud Carlos III* as *Instituto de Investigaciones Sanitarias*
- **2013 |** Re-accreditated by ISCiii
- Nowadays it is the Public Centre with more activity in healthcare and research in Catalonia (clinicial trials, research projects) .

Annual Budget: 600 M€

7,000	1,150	390	50
professionals	beds	consultations	operating rooms

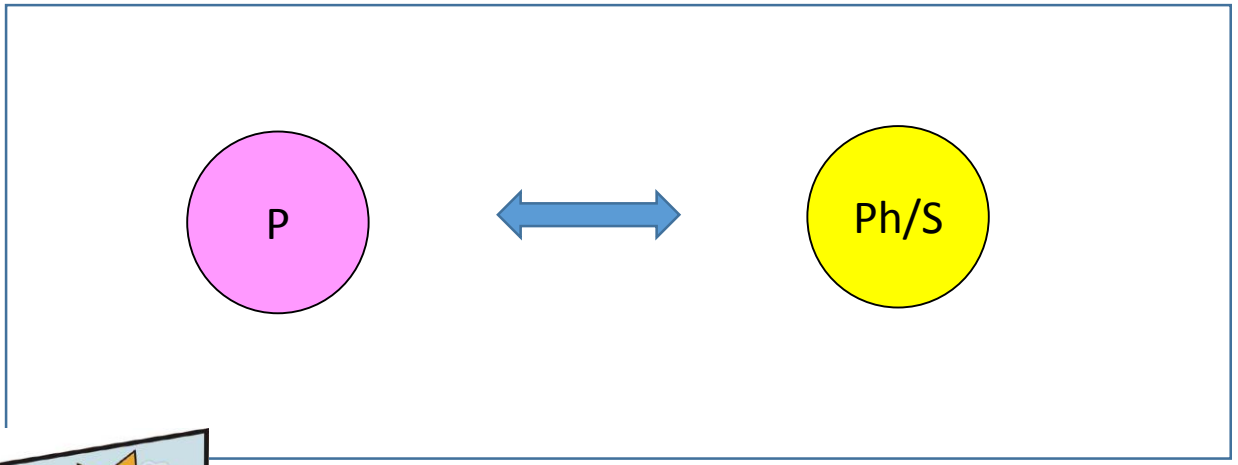
- **General (adults)**
- **Pediatrics, gynecology and obstetrics**
- **Traumatology & Rehabilitation**
- **Research**


Vall d'Hebron
Hospital
MEDICAL SPECIALITIES
Department of Clinical and Molecular Genetics
Rare disease Unit
Clinical Genetics
Laboratory of Genetics
Multidisciplinary
Genetic Counselling
Diagnosis and Follow-up
Arrays
Cytogenetics
Molecular
MEDICAL SPECIALITIES
Specialized laboratories: Biochemistry, Immunology, Hematology , Microbiology, Pathology

Clinical process management (CPM)

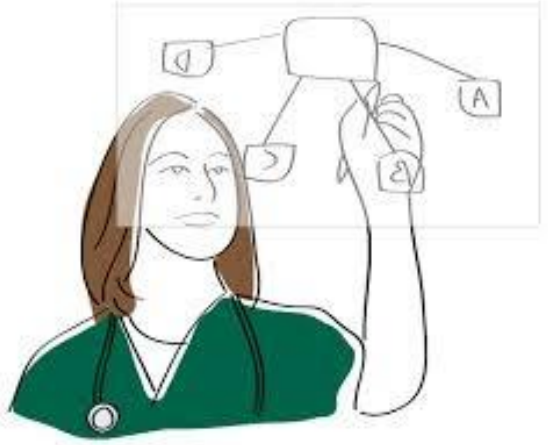
- Traditionally, clinical professionals dealt with patients in semi-isolation
- The concept of the patient as the center of attention of a tightly-interacting team of clinicians is relatively new
- Rare diseases are the paradigm to apply multidisciplinary approaches

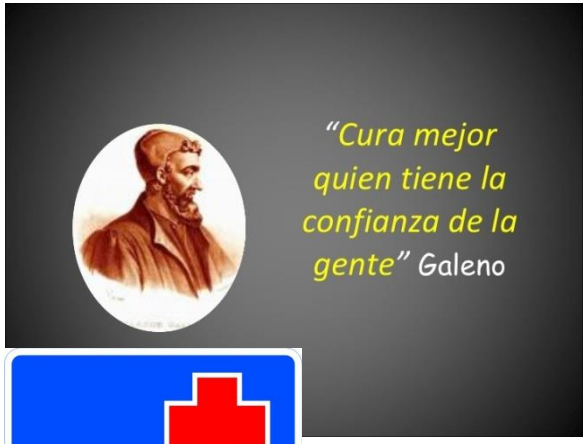
Traditionally monographic consultations Physician-Patient



New protagonists

CASE MANAGER





CM

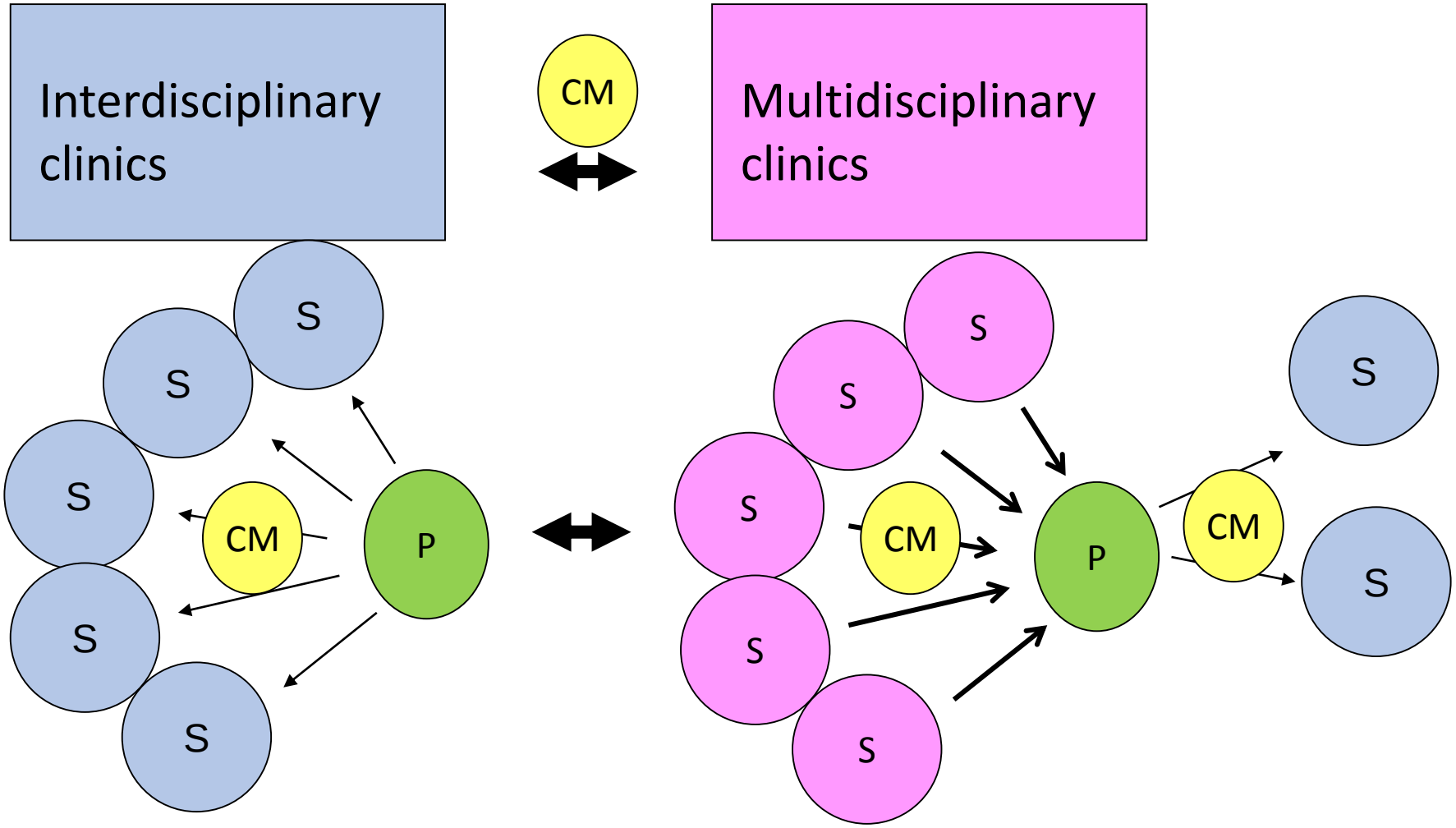
Ph/S

P/F

PHYSICIAN
SPECIALIST/EXPERT
COORDINATOR

PATIENT/ FAMILY

Towards implementation of integrated and multidisiplinary clinics with the collaboration of Case managers

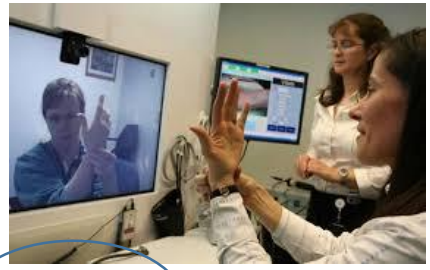


MULTIDISICPLINARY TEAM

- Coordinator or leader (pediatric *and* adult)
- Faculty members and specialists
- Case Manager
- Psychologist
- Administrative support
- Trained social worker
- Capacity to integrates medical, paramedical, psychological and social environment to offer the best to the patient.

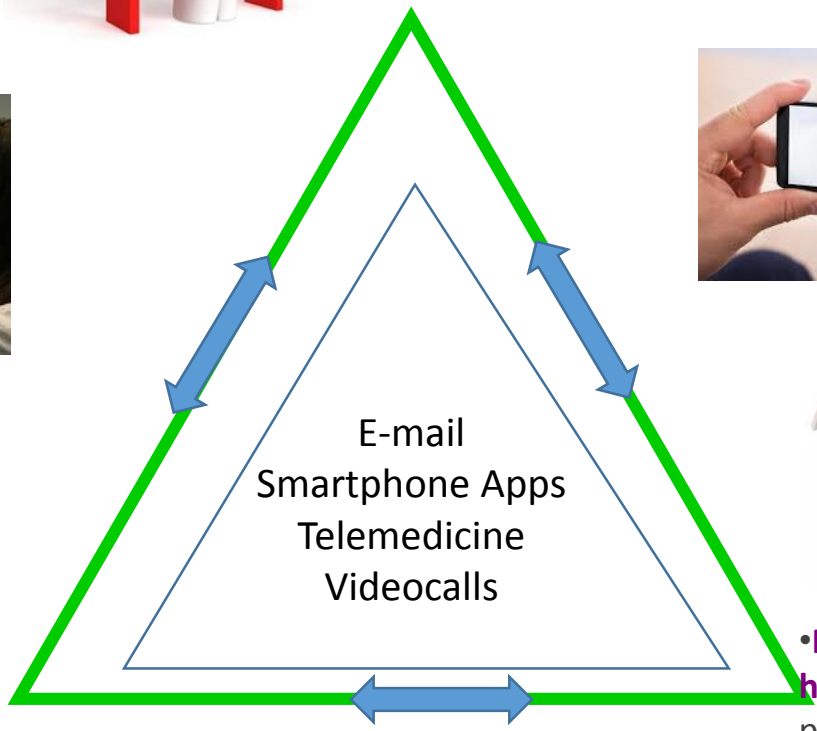
Multidisciplinary Multicentric Care

HOME



Hospital

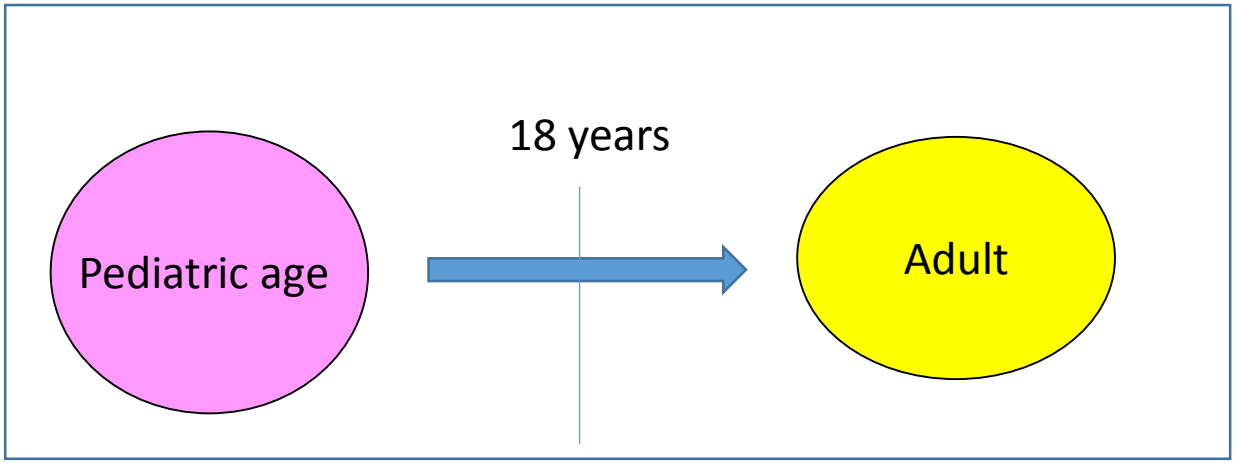
•**Specialized healthcare with strong expertise in RD** that has been formed by Complex Centers and tertiary reference hospitals.



Primary Care

•**Proximity or community healthcare**, constituted by the primary care services, by the specialized referential care (secondary care) and by the overall public resources from the education and social proximity areas from the patient's home.

Traditional **transfer** from pediatricians to adult physicians



ADOLESCENCE

INFANCY

Birth

TRANSITION

Adult life

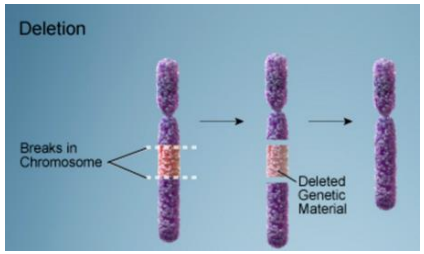

Proactive
Reactive

EXAMPLES OF EXPERIENCES IN A TERTIARY HOSPITAL

Vall d'Hebron
Barcelona Campus Hospitalari

Multidisciplinary assessment for Familial Cardiopathies

Multidisciplinary assessment facing a microdeletion syndrome



Multidisciplinary assessment facing perinatal congenital defects

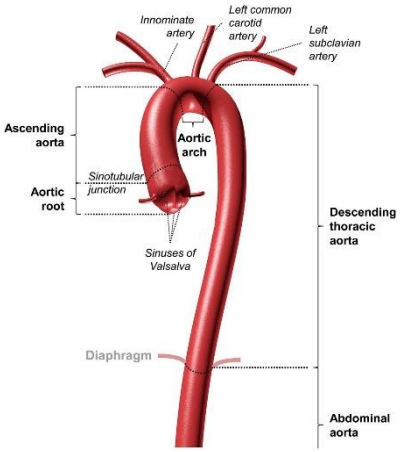
Description of the experiences

- **Profiles of the professionals that participate in the experience**
- **What was the reason to initiate this experience?**
- **Main objectives**
- **Development and evolution of actions and processes**
- **Results**
- **Reasons that facilitated to implement the experience**
- **Something wrong?**
- **How to improve?**

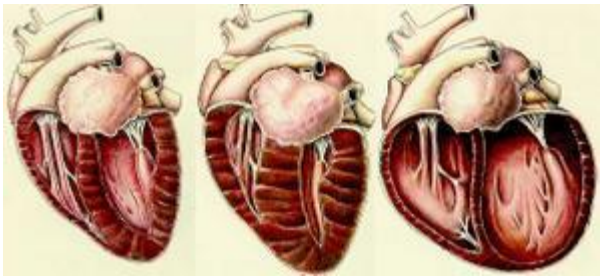
Multidisciplinary assessment for Familial Cardiopathies

Session 1: Promoting integrated holistic care for rare diseases

Three main groups of rare disorders in Cardiogenetics

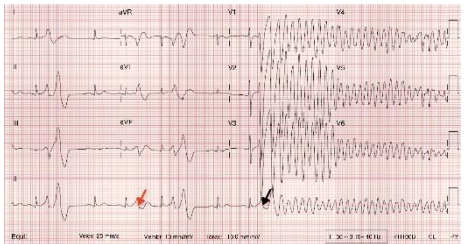


- **Syndromic and non-syndromic aortopathies.**

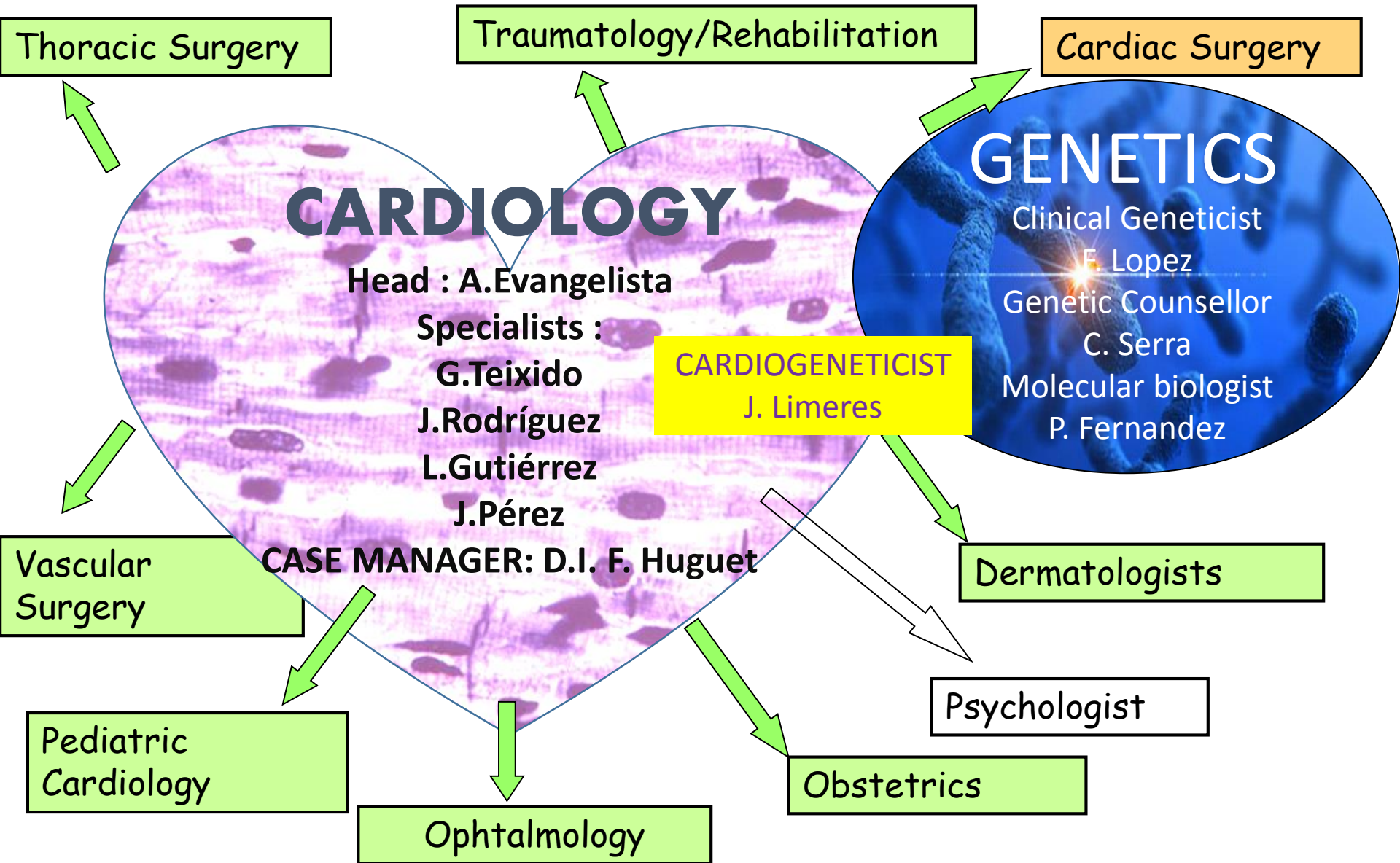


- **Familial cardiomyopathies**

- **Familial channelopathies (inherited arrhythmias)**



Profiles of the professionals that participate in the experience



What was the reason to initiate this experience?

- Starting in 2008, due to the motivation of the professionals involved in the diagnosis, management and follow-up of specific pathologies (Aortic pathology, Arrhythmias), with a high risk of debuting with serious and / or lethal manifestations, and often with a familiar presentation.

Main Objectives

- Comprehensive care for families affected by heart and aortic pathology to optimize resources and to facilitate access to the services required.
- Consolidate an expert group in the detection, monitoring and management of these patients and their families
- Improve the presymptomatic detection of patients through the systematic study of relatives at risk
- Promote research and scientific dissemination related to pathologies
- Create a tissue bank

Development and evolution of actions and processes

- Collaboration with Cardiology Service.
- **Case Manager from the beginning (Nurse)**
- The Unit works in the Cardiology section, Cardiology consultations, Ultrasound and ECG, Genetic consultation and Genetic Counseling.
- Pediatric Cases in Genetics Area
- Molecular team for interpretation of genetic test/NGS
- **Training a Cardiologist in the Genetics Department to implement the figure of CARDIOGENETICIST**

Results

- 700 families referred for Aortopathies.
- 500 families referred for familial myocardiopathies and channelopathies.
- More than 2,800 patients visited /registry
- Bank of tissues and samples.

- The Unit has become a reference Center for Catalonia and other regions in Spain in the management of familial cardiopathies and aortopathies, and is being an example of functioning and joint effort to manage and derive complex pathologies.

Reasons that facilitated to implement the experience

- The implementation of this Unit has been possible thanks to the interest of the Cardiology Service, to the existence of a Genetics Service capable of assuming clinical consultations and counselling to families, and **performance and interpretation of genetic studies in a multidisciplinary team** essential for the diagnosis and management of these pathologies.
- The success of the Unit is linked to the existence of a recognized profile of a specialized nurse as **Case Manager**

Something wrong?

How to improve?

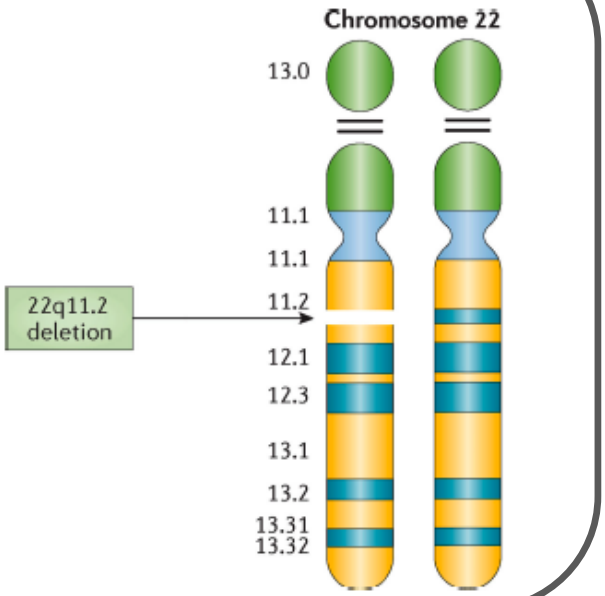
High demand
Waiting list
Overload of the consultations
Lack of psychological support to families and patients

- Incorporation of Professionals to shorten the waiting list
- Derive some controls to Centers of origin.
- Psychological support for patients

Multidisciplinary assessment facing a microdeletion syndrome

Session 1: Promoting integrated holistic care for rare diseases

22q11 deletion Syndrome



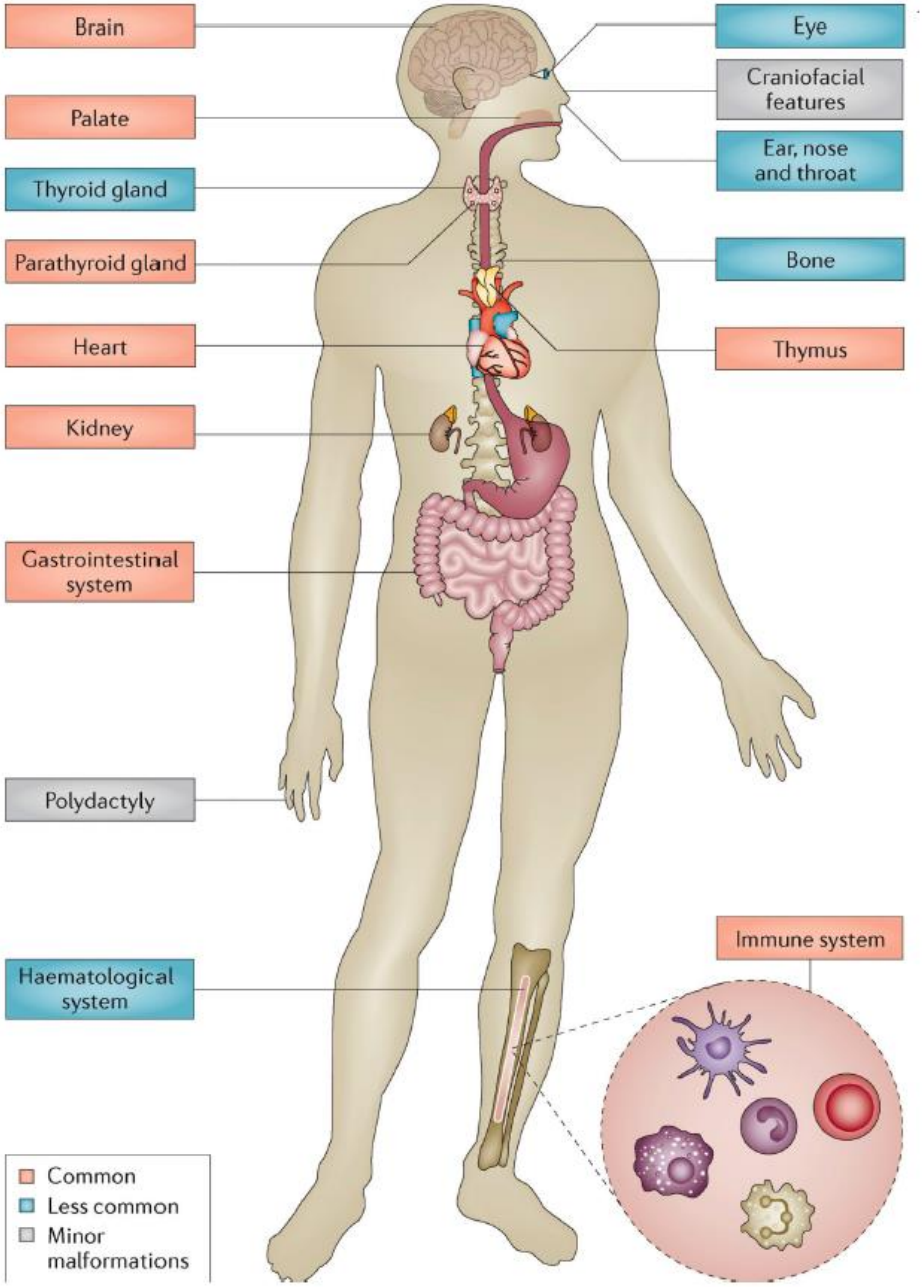
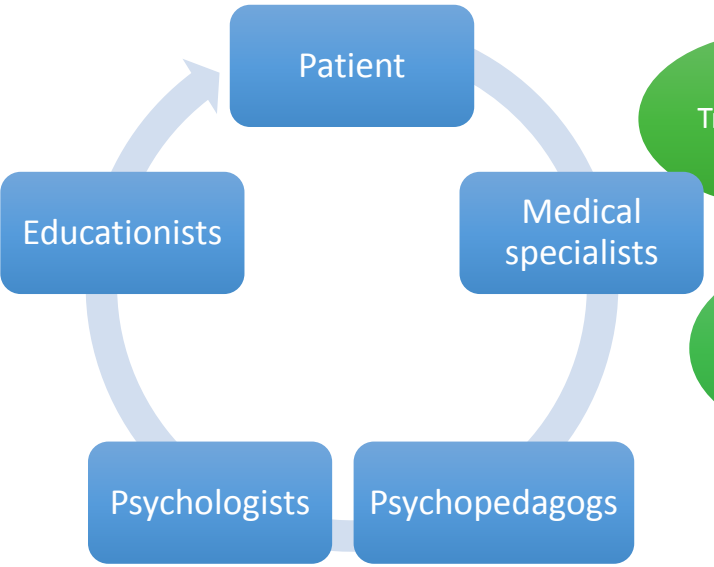


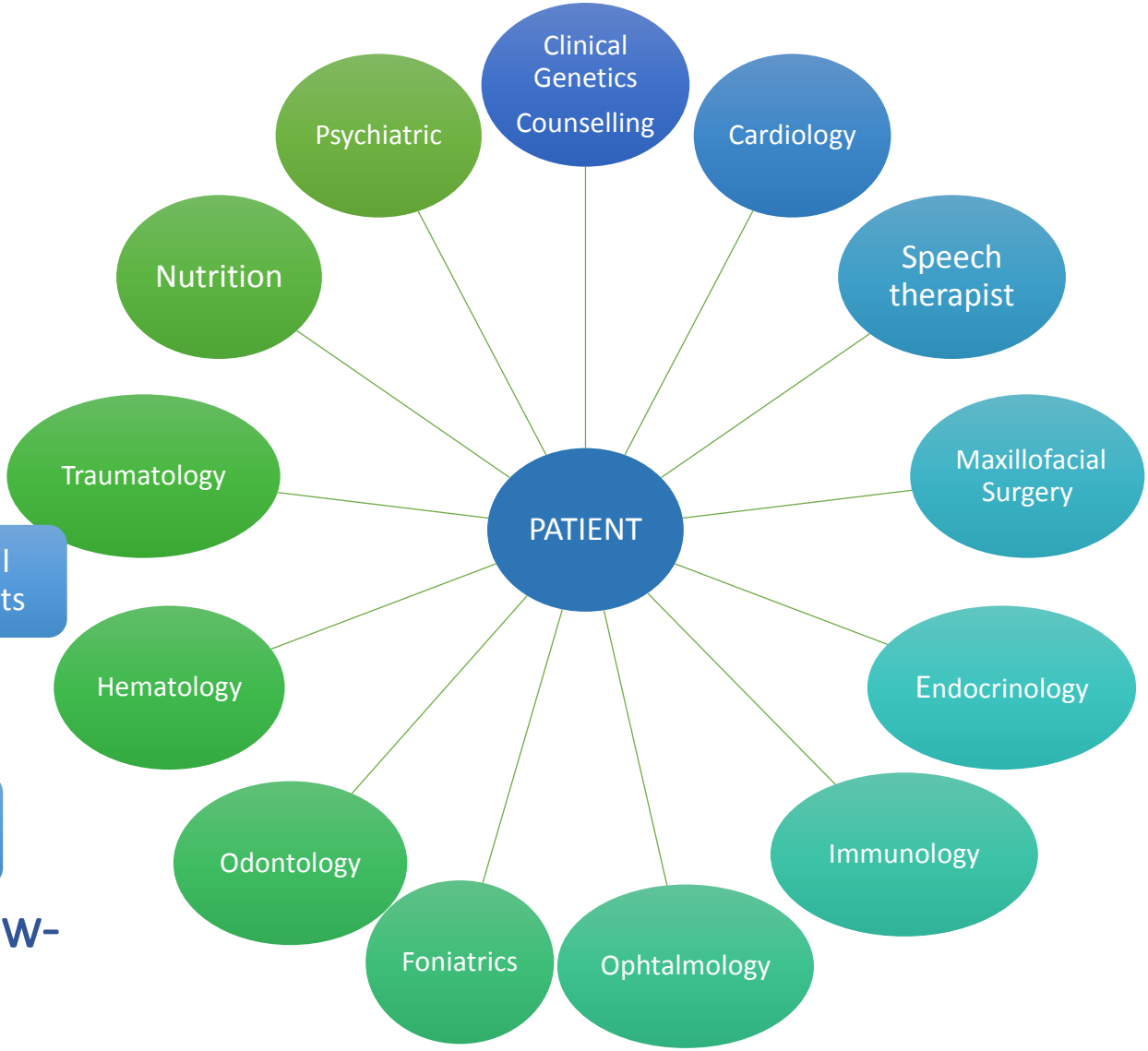
Figure 5. Organ and system involvement in 22q11.2 deletion syndrome

McDonald-McGinn DM, et al. Nat Rev Dis Primers; 1: 15071. doi: 10.1038/nrdp.2015.71.

Profiles of the professionals that participate in the experience



Multidisciplinary follow-up



What was the reason to initiate this experience?

- **Increasing number of patients diagnosed during the last years**
  (from prenatal to adult life)
- **Variability of non classic symptoms**
- **Better knowledge and awareness of the disease**
- Intention to improve communication among all specialists
- Proposal to families to meet together in the hospital

Main Objectives

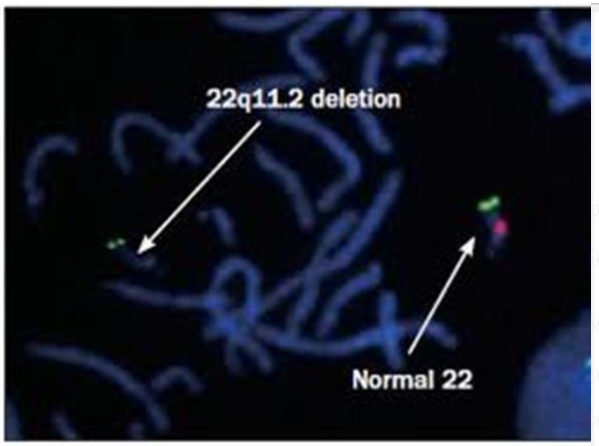
- **Improve care to patients and relatives**
- **Improve coordination of medical visits**
- Prevention and management of some manifestations
- Interact with families
- Ensure a correct diagnosis and genetic counselling
- Creation of a follow-up protocol

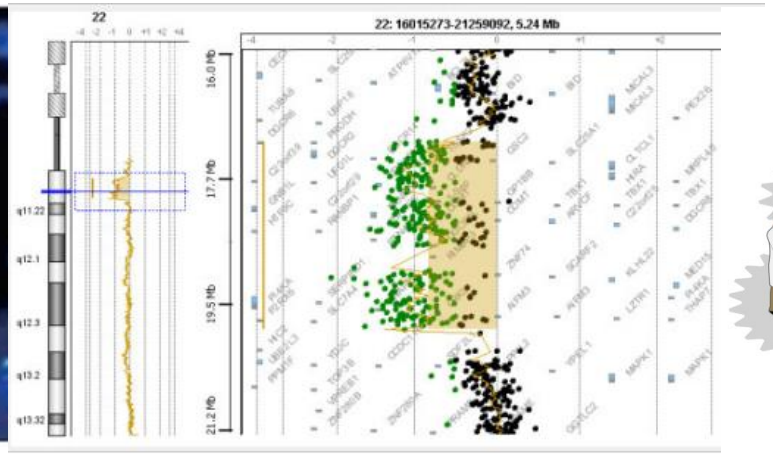
Development and evolution of actions and processes

Year 1996
- **First diagnosis**
- FISH

Year 2008
- Implementation of **array-CGH**
- Increase and improving diagnosis

Year 2012
- **Implementation of multidisciplinary follow up**
- Improvement of care





Results

- First diagnosis in 1996 → **143 patients**
- Initial multidisicplinary follow-up in 2012
- Active follow-up of 116 patients
- Recontact of some adult patients to continuous follow-up
- Specialist committee to evaluate patients and define protocols
- Activation of Catalan Society 22q:
  - Offering of a meeting space for members
  - Active participation in the annual world day at the zoo, at scientific conferences and family gatherings

Reasons that facilitated to implement the experience

- Improved awareness of other specialists increase diagnoses
- Provision in the same hospital of the different specialists prenatal - pediatric - adult
- **Activation of 22q Catalonia Association**, facilitating a meeting space to meet, collaborate in the organization of family gatherings, conferences, world celebration ...
- Creation of a **multidisciplinary monitoring protocol**
- **Laboratory and clinics working together: studies are carried out and interpreted within the same center**

Something wrong?

- Attempts to unify the same day for the different specialists is yet difficult (several visits in the same day)

How to improve?

- Devoted case management
- Increase psychological support
- Work in the transition process

- Clinical and Molecular Genetics (Teresa Vendrell and Anna Mª Cueto-González)
- Pediatric Cardiology (Queralt Ferrer) Adults (Congenital Heart Disease Unit)
- Pediatric Endocrinology (Maria Clemente León) Adults (Anna Casteras)
- Maxillofacial Surgery (Montserrat Munill and Nicolas Sierra)
- Pediatric Psychiatry (Nuria Gómez) Adults (Gemma Perramon)
- Psychology (Raquel Vidal)
- Pediatric Immunology (Andrea Martin)
- Adults Pediatric Ophthalmology (Silvia Alarcón)
- Adults Otorhinolaryngology (Marc Pellicer)
- Neuropediatry (Anna Felipe) Phoniatrics (Mercedes Velasco and Anna Maria León)
- Speech therapy (Tania Puignou)
- Pediatric Hematology (José Luis Dapena)
- Nutrition (Susana Redecillas)
- Traumatology (Cesar García Fontecha and Marius Aguirre) Rheumatology (Consuelo Modesto)
- Sleep disorders (Elena Jurado)

Vall d'Hebron
Barcelona Campus Hospitalari

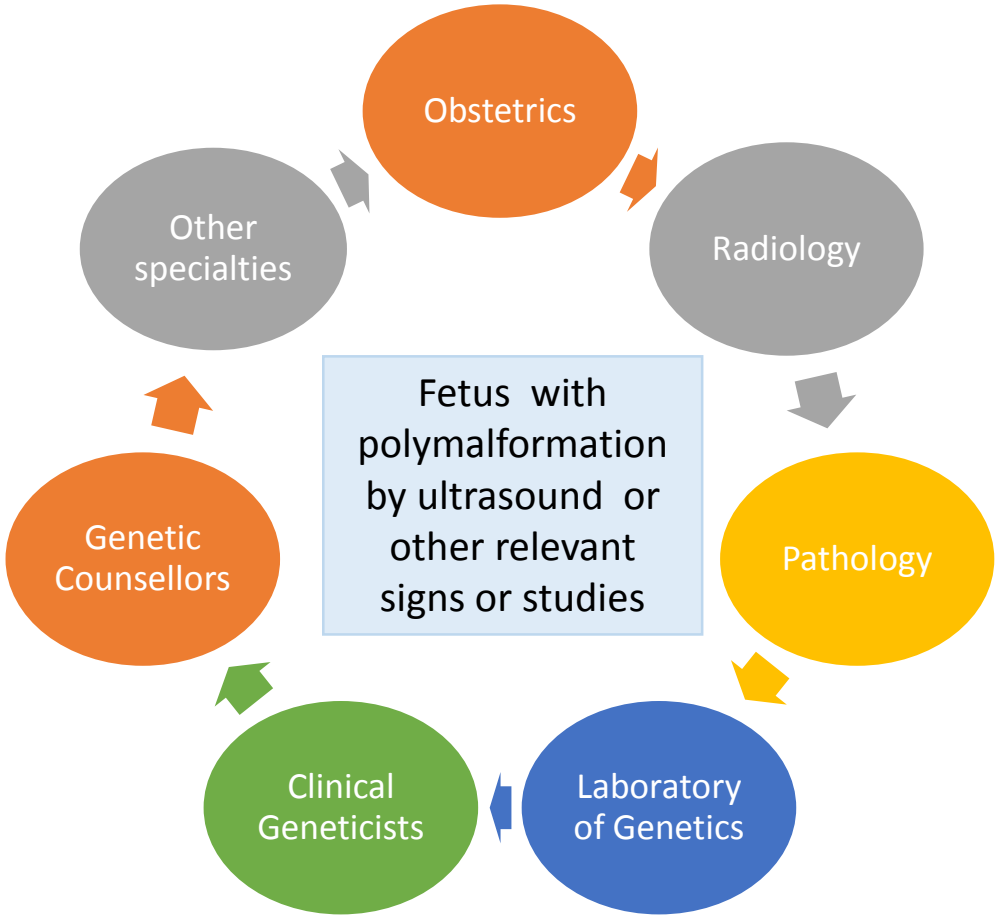
Multidisciplinary assessment facing congenital defects

Session 1: Promoting integrated holistic care for rare diseases

Aspects of relevance when a congenital defect is detected in pregnancy

- Face the loss of a desired pregnancy
- Anguish feeling of repetition
- Lack of categorical fetal diagnosis
- Recurrence risk is unknown
- Maternal-fetal link in 2nd and 3rd trimester (visualization of the fetus in ultrasounds, fetal movements, delivery)

Profiles of the professionals that participate in the experience



Specialists: Neonatologists, neurologists, cardiologists, nephrologists, etc.

Involvement of Specialists

Vall d'Hebron Barcelona Hospital Campus

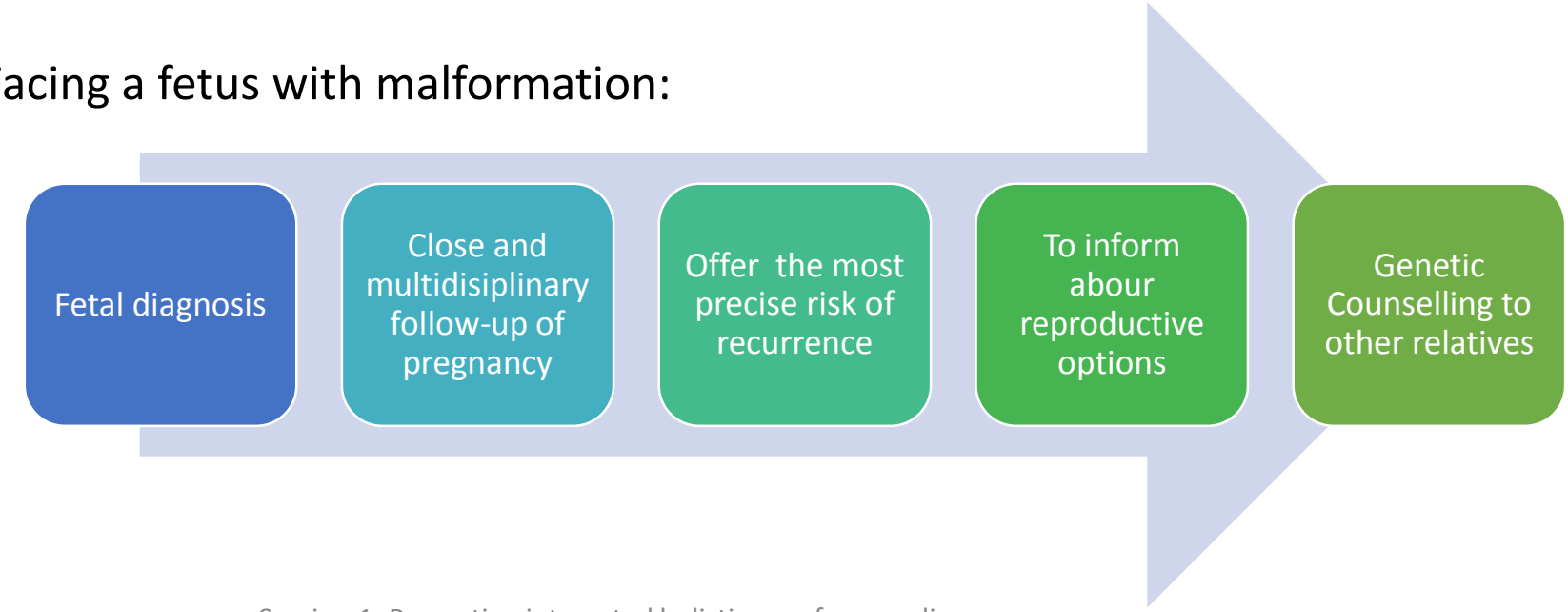
- Fetal Medicine Unit
- Clinical and Molecular Genetics Division
- Pathology Department
- Radiology Department
- Other specialties

What was the reason to initiate this experience?

- To achieve diagnosis of a fetus with malformations
- Need to allow the assesment of the couple
- To offer emotional support to the couple
- In case of interruption
  - To define the recurrence risk
  - Offer a follow-up in the next pregnancy
  - Death mourning accompaniment

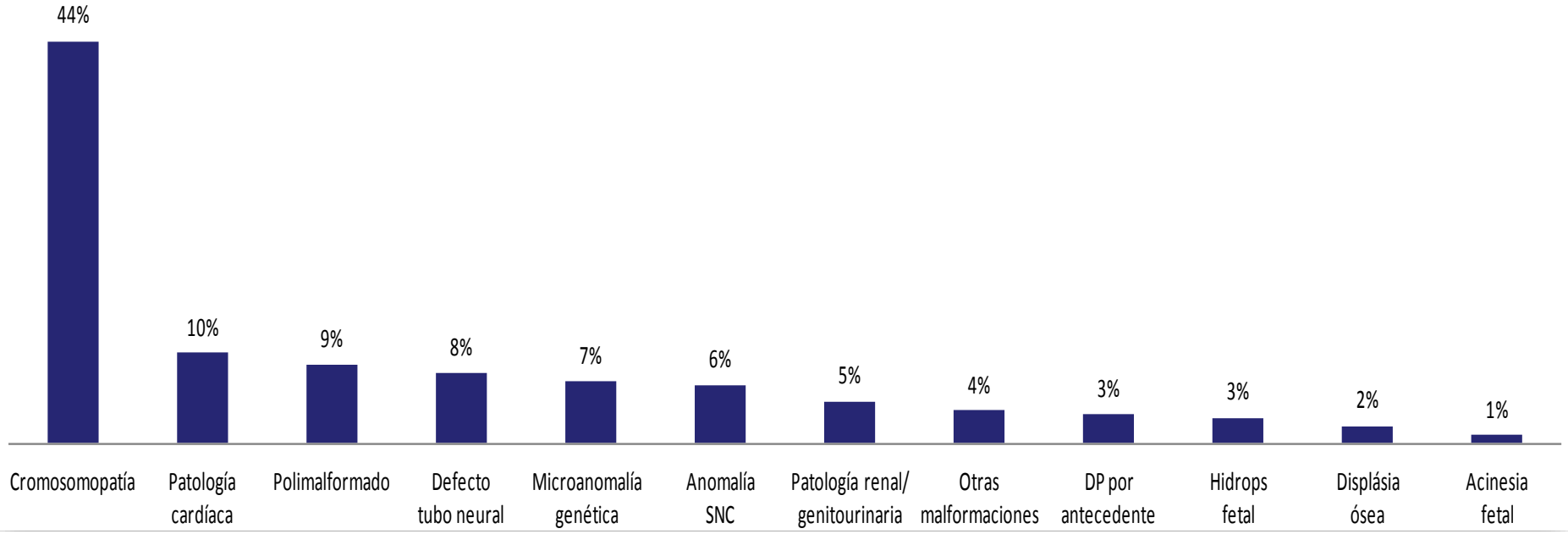
Main objectives

Facing a fetus with malformation:



Results

- From 2013-2017, 940 cases and consultations of post-termination of pregnancy, chromosomal anomalies was the most frequent indication of termination (44%).



Reasons that facilitated to implement the experience

- **Good predisposition and implications of all professionals involved**
- **Increase of requests for this type of situations**
- **Improving in the diagnostic tools**

Something wrong?

- In the beginning, certain difficulties to obtain the information coordinated with the rest of specialties s and at an appropriate time
- Sometimes homogeneity can lack information and the message transmitted to the couple

How to improve?

- Adapt better space (favoring the privacy of the couple)
- Offer psychological help throughout the process

Conclusion: even without a clear diagnosis, it is successful

- Post-termination of pregnancy consultation involve s a multidisciplinary collaborative work, **being essential to reach a diagnosis** that allows to offer a precise risk of recurrence.
- In cases without fetal diagnosis, it is vital to establish an empirical risk and inform about reproductive options that decrease this risk.
- **This type of consultation provides the opportunity for the couple to have a space in which to deal with emotional aspects related to the termination.**
- These aspects help to reduce the anguish/sorrow of the process, contribute to the management of the mourning of the lost pregnancy and provide information that helps to decide their reproductive options.

Something wrong?

High demand
Waiting list
Overload of the consultations
Lack of psychological support to families and patients

- Attempts to unify the same day for the different specialists is yet difficult (several visits in the same day)

- In the beginning, certain difficulties to obtain the information coordinated with the rest of specialties s and at an appropriate time
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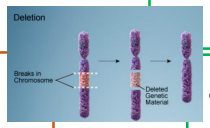
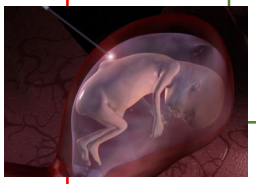




How to improve?

- Incorporation of Professionals to shorten the waiting list
- Derive some controls to Centers of origin.
- **Psychological** support for patients

- Devoted case management
- Increase **psychological** support
- Work in the transition process

- Adapt better space (favoring the privacy of the couple)
- Offer **psychological** help throughout the process

Multidisciplinary team for undiagnosed cases in the context of genomic/exomic approach

Clinical and laboratory specialists (according with pathology and manifestations)
Molecualr biologists
Clinical Geneticists
Genetic Counsellors /Genomic Counsellors
Bioinformaticians

XUEC Xarxa de Unitats Expertessa Clínica de Malalties Cognitius –Conductuals

Network of Units of Expertise in Intelectual Disability Disorders

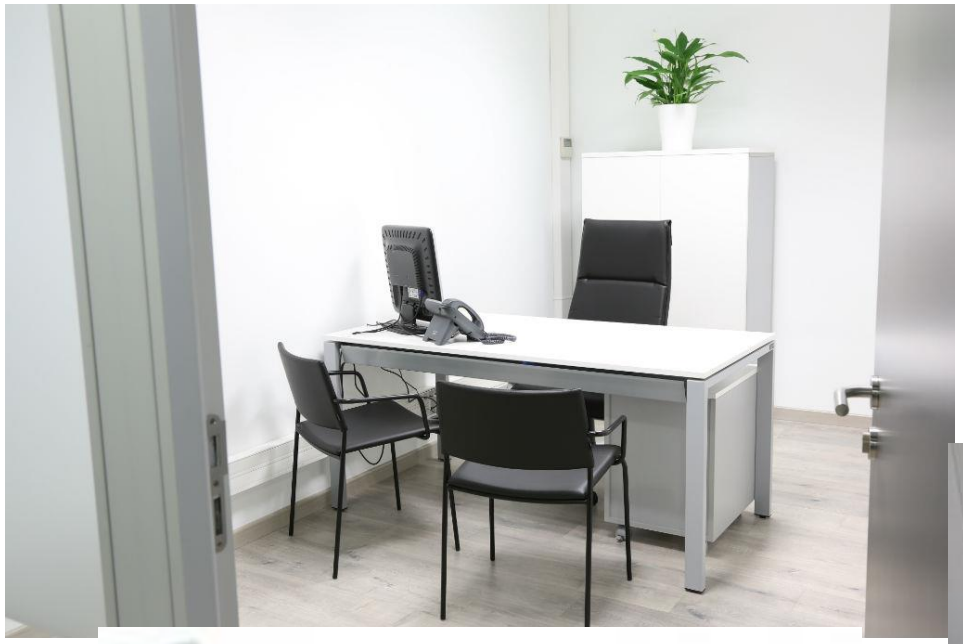
Created by the Health Department of Catalonia with the perspective to have similar thematic areas as ERNs

Objectives: coordinate and optimize the follow-up of patients with different IDD by expert centers

TALLER PARA PACIENTES CON ENFERMEDADES RARAS

MÁS ALLÁ DE LA PROPIA ENFERMEDAD

Interactive workshops for patients with rare diseases
“Beyond the own disease”



Better communication and counselling in a round table

Managing expectations of the patient

People on table. Communication.

Eduardo Tizzano MD, PhD e tizzano@vhebron.net
Department of Clinical and Molecular Genetics
Rare Diseases Unit
Hospital Vall d´Hebron
Medicine Genetics Group VHIR