

ERN-EYE

Looking towards a holistic vision for Rare Eye Diseases

Hélène Dollfus* , Melissa Coriano* , Fouzia Studer* , David Keegan**

- *Hôpitaux Universitaires de Strasbourg – France
- ** Mater Misericordiae University Hospital / University College Dublin -Ireland

FRAMBU, 12TH APRIL 2018

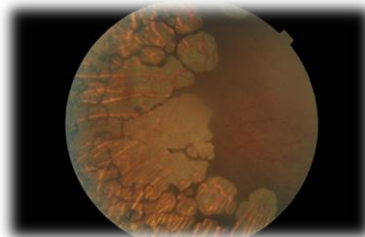
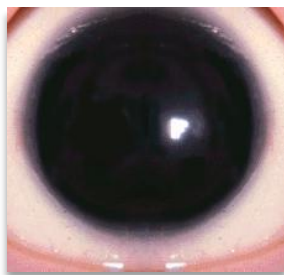


**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Eye Diseases (ERN-EYE)





Rare Eye Diseases

1/3 of RARE diseases affect the EYE

900 entities in ORPHANET & 30% are syndromic

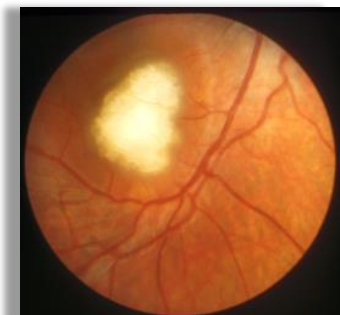
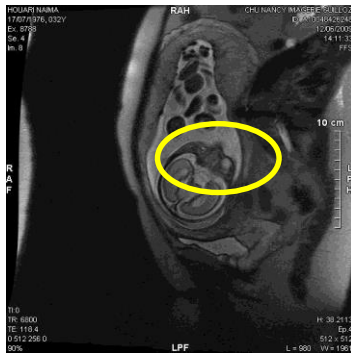
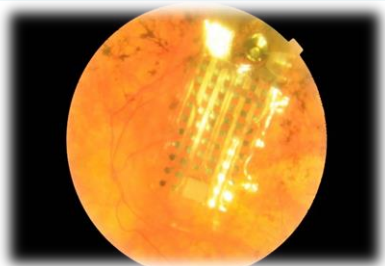
Occur in Childhood and Adulthood

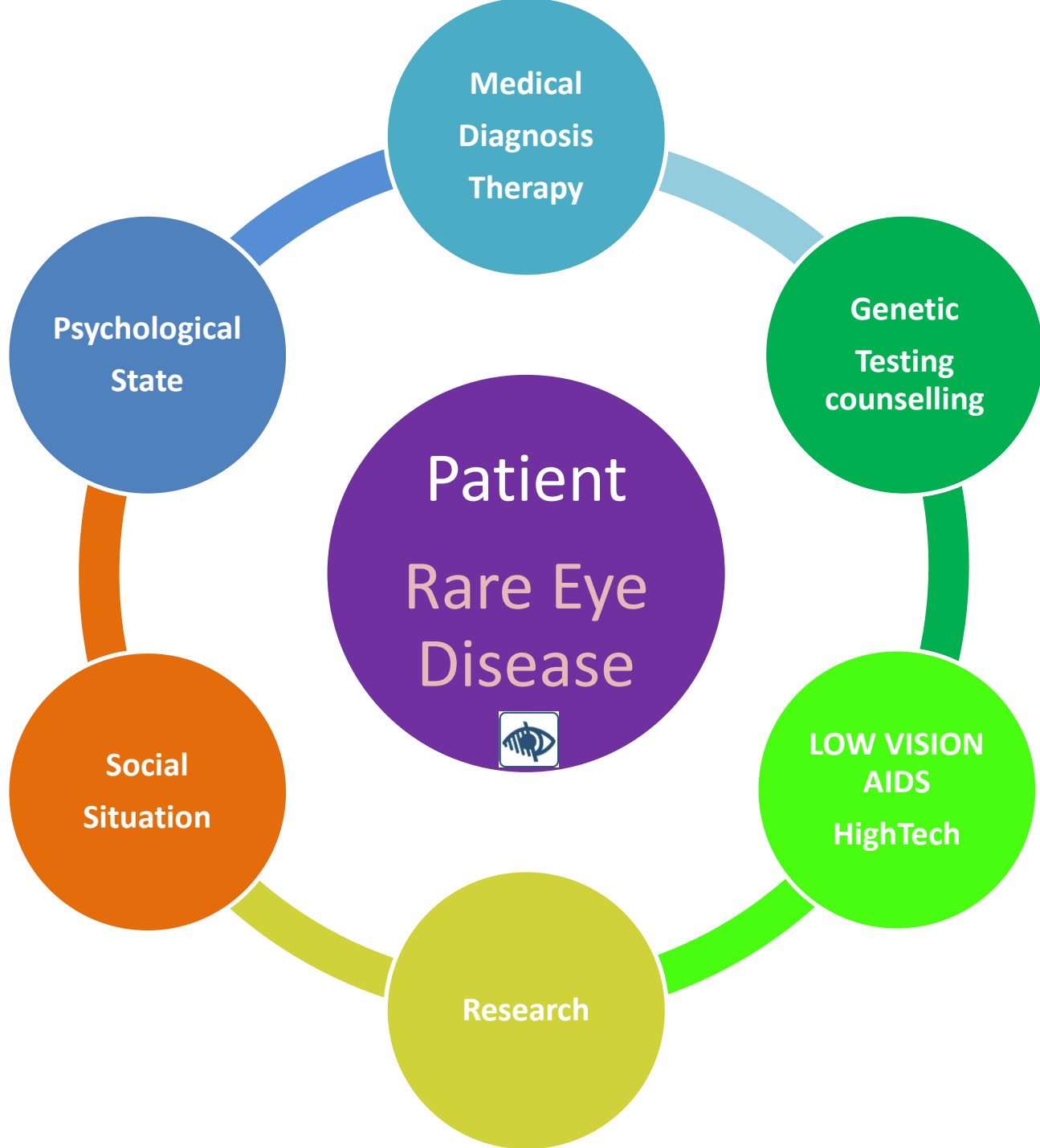
First cause of blindness for children and the young in the EU

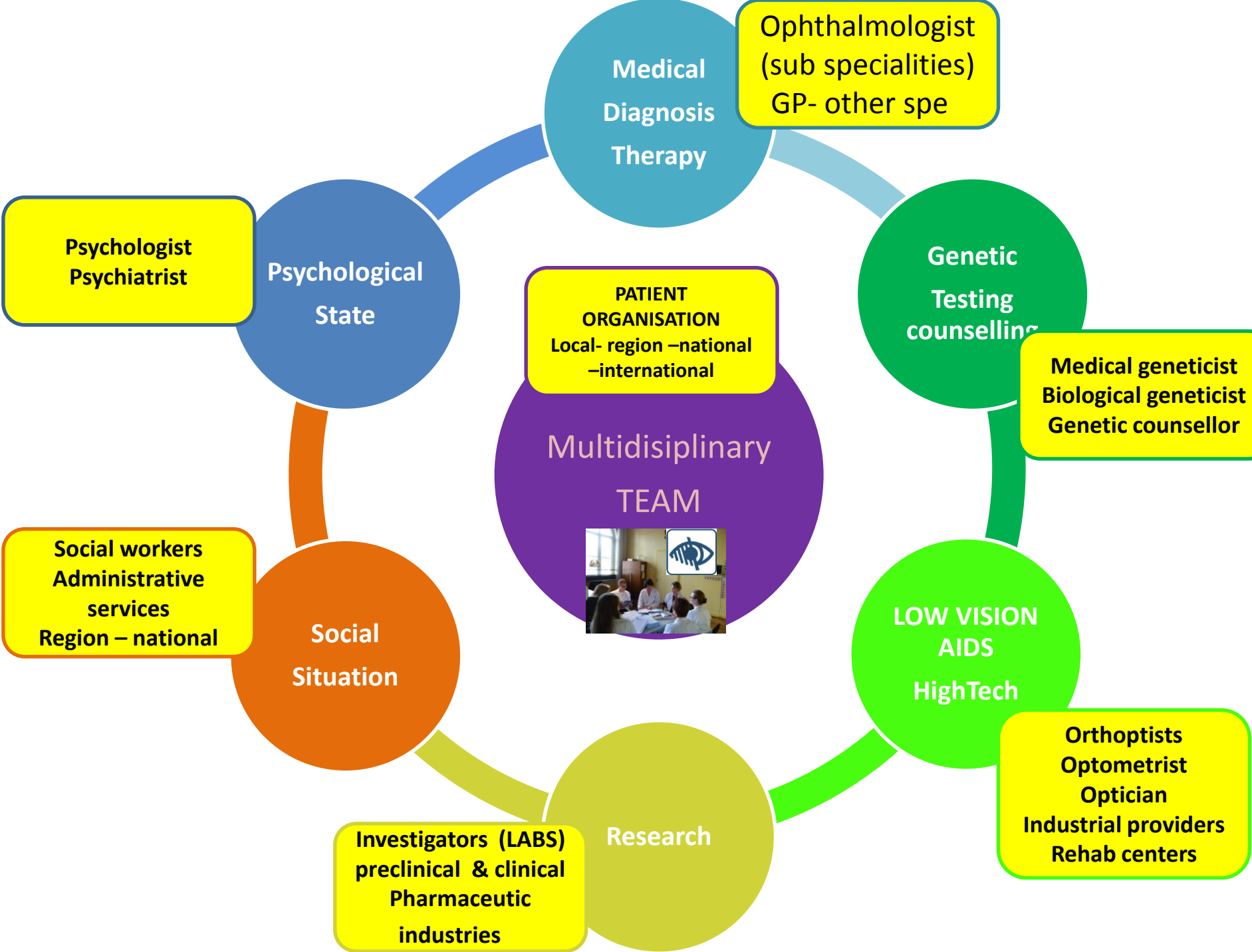
The leading cause legal blindness in early life in the UK (Liew et al, 2014)

Still lack of curative therapy for the majority of patients

Conditions are developmental / degenerative / concern the whole eye or a sector







What Patients Want:



Downloaded from <http://bjpo.bmj.com/> on April 8, 2018 - Published by group.bmj.com

Clinical science

Understanding the expectations of patients with inherited retinal dystrophies

Ryan Combs,^{1,2} Georgina Hall,² Katherine Payne,¹ Jo Lowndes,³ Sophie Devery,⁴ Susan M Downes,³ Anthony T Moore,⁴ Simon Ramsden,² Graeme C M Black,^{1,2} Marion McAllister^{1,5}



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www.nature.com/eye

CLINICAL STUDY

Identifying variation in models of care for the genomic-based diagnosis of inherited retinal dystrophies in the United Kingdom

¹Manchester Centre for Health Economics, The University of Manchester, Manchester, UK

²Manchester Centre for Genomic Medicine, Institute of Human Development, Faculty of Medical and Human Sciences, University of Manchester, Manchester Academic Health Science Centre (MAHSC), Saint Mary's Hospital, Oxford Road, Manchester, UK

³Manchester Centre for Genomic Medicine, Central Manchester University Hospitals NHS Foundation Trust, MAHSC, Saint Mary's Hospital, Oxford Road, Manchester, UK

⁴Centre for Medical Education, School of Medicine, Cardiff

Abstract

Purpose Advances in genomic technologies are prompting a realignment of diagnostic and management pathways for rare inherited disease. New models of care are being developed as genomic-based diagnostic testing becomes increasingly relevant within more and more aspects of medicine. This study describes current care models for the provision of a genomic-based diagnosis for patients with inherited retinal dystrophy (IRD) in UK clinical practice.

Methods A structured telephone survey, conducted (in 2014) with all 23 UK Regional Genetics Centres and a sample of specialist ophthalmology centres (n=4), was used to describe models of service delivery and current levels of genomic-based diagnostic testing. Quantitative data were summarised using descriptive statistics. Responses to open-ended questions were summarised using thematic analysis.

• Clinical:

Diagnosis,
Prognosis,
Inheritance pattern/family
planning, Upcoming research

• Practical support:

Registration
Signposting of services (e.g.
financial, education,
low vision and mobility support)

• Psychological support:

Help adjusting to diagnosis and
visual loss

1° Patients Clinical Expectations

- **Diagnosis, “name the disease”**

“ending of the odyssey”

Clinical - functional evaluation

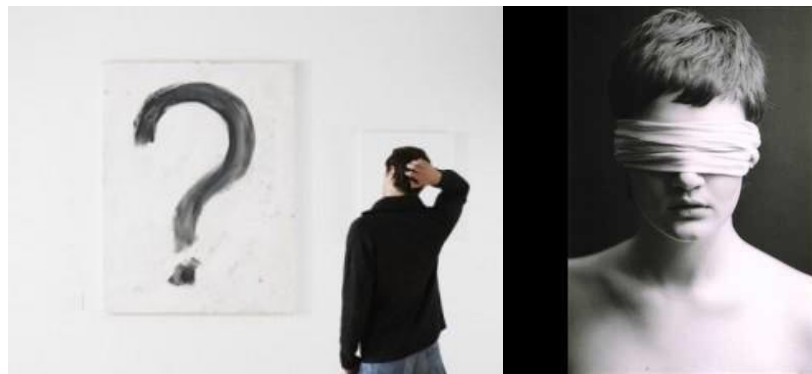
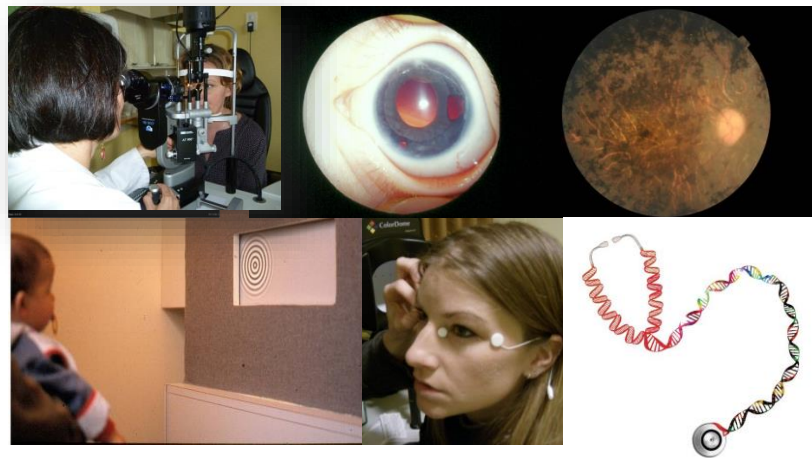
Genetic diagnosis (Panels, NGS):

RP >50% pathogenic variant identified

- **Prognosis, “what is going to happen”**

Some conditions are stable other will show progression ... slow or rapid ...

- Ophthalmic evaluation & functional evaluation are different
- “When will my child lose completely vision ? What does he/she see? ”
- “Will I get blind?”
- “ When will it be completely dark ?”
- “Can I still drive ?”

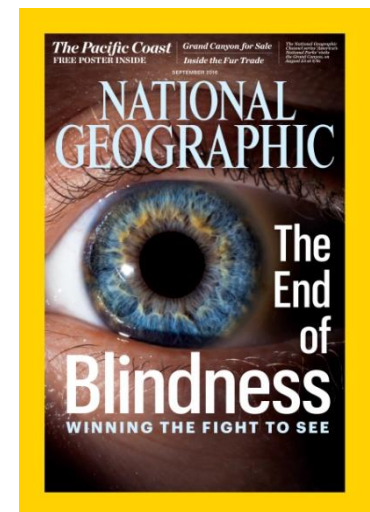
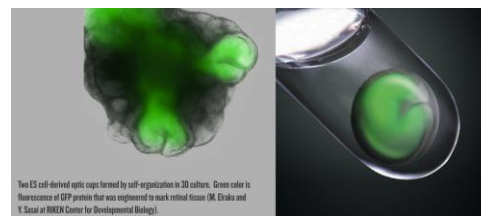
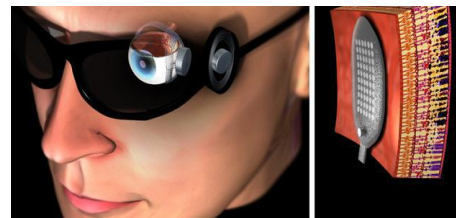


1° Patients Clinical Expectations

- Inheritance pattern/family planning,
- => genetic counseling
- => procreative options



- Upcoming research: what's up ?
Can I participate?



2. Practical support

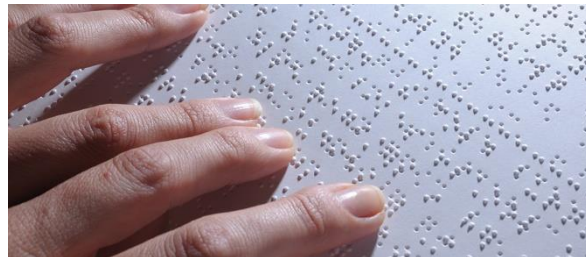
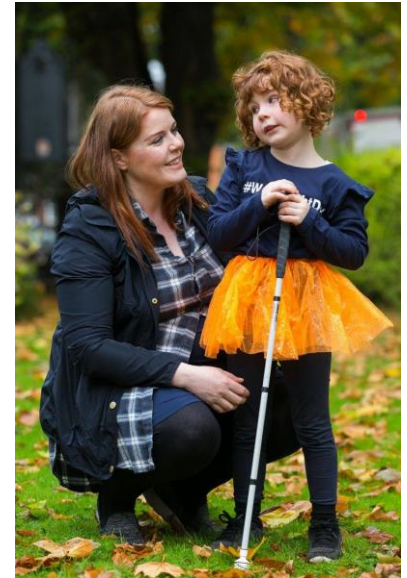
Loss of independence

=> REGISTRATION

=> Signposting of services

e.g. financial, education, work

low vision and mobility support



**Chiens guides d'aveugles ou
d'assistance uniquement**

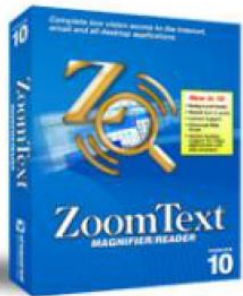


**2005-2015 : Accessibilité pour tous,
10 ans sans y jeter un œil !**

=> LOW VISION team



=> HIGH TECH SUPPORT TO VISUALLY IMPAIRED AND BLIND PATIENTS



3° Psychological support

- Help adjusting to diagnosis and visual loss to a **unique individual** with his own situation

- Blind child (LCA) and his mother, ...
- The young adult with NO Leber , ...
- The child with Stargardt disease, ...



- **Symbolism of visual loss – Loss of independence**

Grieving process

- Denial => I have no loss
- Depression = > I have all lost because I have lost vision ...
- Reaction => I have only lost that ...



3° Psychological support

- **Professionals** to care for psychological support
- Follow up +++ , Psychologists, Psychiatrists, Nurse, ...
- **Patient organizations** (international, national, regional, local)
- **Group therapy**
- **Telephone assistance**
“**Need to talk**” project for the Royal National Institute of Blind People (RNIB) project (Republic of Ireland, Northern Ireland, Scotland)

“I have been struggling with sight loss for 30 years, trying to live in a sighted world where no one really understood. Now, since receiving counselling, I’ve a whole new lease of life. I am now filled with hopes and dreams and my world is a much brighter place filled with endless possibilities.” – Helen



PROMS for RED : generic or specific ?

- (1) Can make important life decisions in an informed way (*decisional control*).
- (2) Has sufficient information about the condition, including risks to oneself and one's relatives, and any treatment, prevention and support available (*cognitive control*).
- (3) Can make effective use of the health and social care systems for the benefit of the whole family (*behavioural control*).
- (4) Can manage one's feelings about having a genetic condition in the family (*emotional regulation*).
- (5) Can look to the future having hope for a fulfilling family life, for oneself, one's family, and/or one's future descendents (*hope*).

Delivering practical solutions relating to available state benefits entitlement, adaptations and mobility for visually disabled people are of crucial importance to enable those affected by RD to *maintain financial, social, practical and educational independence*.

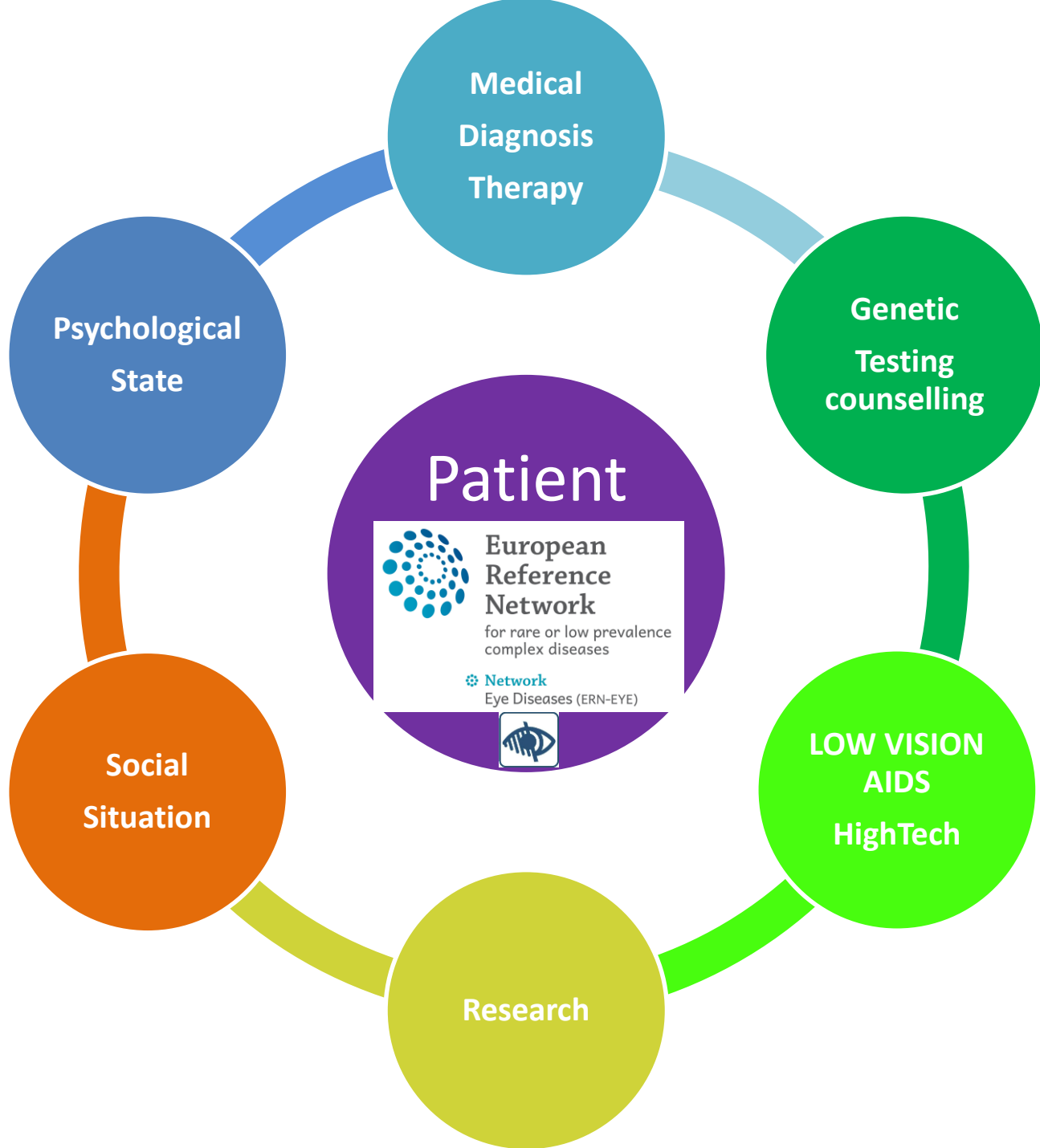
Loss of independence has been identified previously as a key issue for people with visual disability. Identification of independence as an important outcome distinguishes current findings from previous research relevant to clinical genetics.

PROMs developed for evaluating generic clinical genetics may thus not capture all patient benefits relevant to RD.

McAllister M,
The Genetic Counseling Outcome Scale: a new patient-reported outcome measure for clinical genetics services.
Clin Genet 2011



Ryan Combs et al
Understanding the expectations of patients with inherited retinal dystrophies
BJO 2011





ERN-EYE MEMBERS -29 HCPs

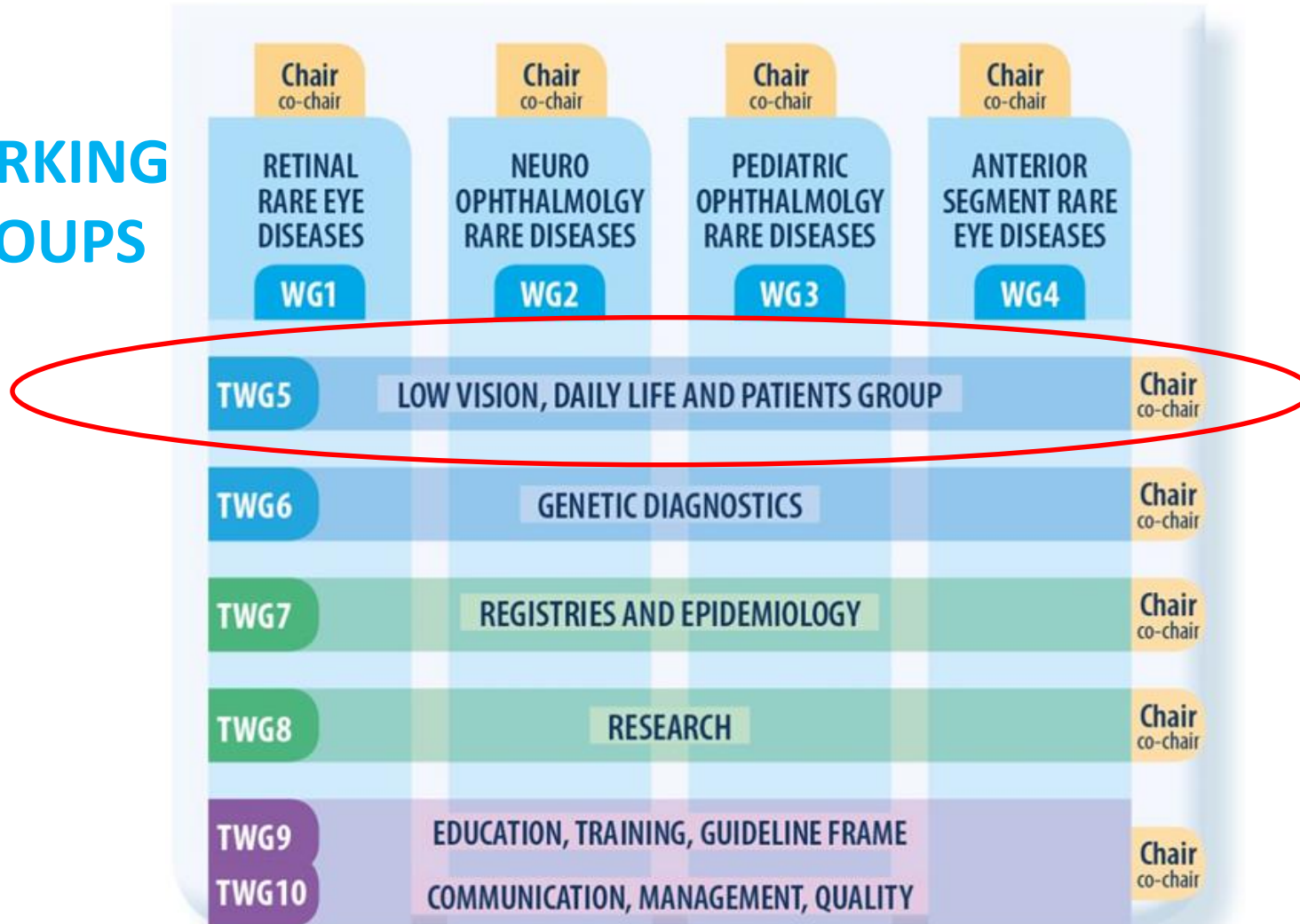
13 Member States

- Belgium
- Czech Republic
- Denmark
- Estonia
- France
- Germany
- Italy
- Latvia
- Lithuania
- Netherlands
- Poland
- Portugal
- United Kingdom





WORKING GROUPS



Schedule of Services in Ireland

Services/Products	Available in Ireland											Available World Wide					
	NCBI	Arbour Hill Braille Unit	Assist Ireland	Child Vision	Fighting Blindness	Irish Guide Dogs	M.I.S.T.	Assoc of Housing for the Visually Impaired	National League of the Blind	The Anne Sullivan Centre for the Deafblind	Visiting Teacher Service	Visually Impaired Computer Society of Ireland	United States	United Kingdom	Australia	Denmark	
Website Last Updated	22-Jun	NA	20-Apr	06-Jun	15-Jun	27-Jun	20-May	Dec-15	Dec-16	22-Mar	20-Mar	NA					
Early Intervention				✓						✓			2	✓	✓	✓	✓
Education Programmes				✓									2	✓	✓	✓	✓
Rehabilitation Training/Therapies	✓			✓				✓			✓		4	✓	✓	✓	✓
Orientation and Mobility	✓			✓		✓							3	✓	✓	✓	✓
Braille Training	✓			✓				✓					3	✓	✓	✓	✓
Braille Production	✓	✓		✓									3		✓		
Daily Living	✓			✓		✓		✓		✓	✓		6	✓	✓	✓	✓
Residential Training/Living				✓		✓		✓		✓			4	✓	✓	✓	✓
Kitchen Skills	✓					✓		✓					3	✓	✓	✓	✓
Customer Service Skills	✓			✓		✓							3	✓	✓	✓	✓
Employment Service	✓			✓		✓							3	✓	✓	✓	✓
Money and Finance Services													0		✓		
Technology Help Groups	✓			✓	✓							✓	4	✓	✓	✓	✓
Community Integration Prgrms	✓					✓		✓	✓				4	✓	✓	✓	✓
Activity Centers	✓			✓				✓	✓				4	✓	✓	✓	✓
Local Support Worker	✓							✓		✓			3	✓	✓	✓	✓
Counselling	✓				✓								2	✓	✓	✓	✓
Group Therapy					✓								1	✓	✓		✓
Support Groups	✓				✓		✓						3	✓	✓	✓	✓
Access Audit Services	✓					✓							2	✓	✓	✓	✓
Funding and Benefits									✓				1		✓		✓
Radio													0		✓	✓	✓
Library	✓			✓									2	✓	✓	✓	✓
Books	✓		✓										2	✓	✓	✓	✓
Braille Accessories	✓		✓										2	✓	✓	✓	✓
USB Players	✓		✓										2	✓	✓	✓	✓
DAISY and mp3 Players	✓		✓										2	✓	✓	✓	✓
Trascription Tools			✓										2	✓	✓	✓	✓
Talking Pens/Labels	✓		✓										2	✓	✓	✓	✓
Paper (pen and braille)	✓		✓										2	✓	✓	✓	✓
Writing Guides	✓		✓										2	✓	✓	✓	✓
Scented Pens	✓												1	✓	✓		
Bright Labelers	✓		✓										2	✓	✓	✓	✓
Bumpons	✓		✓										2	✓	✓	✓	✓

Services/Products	Arbour			
	NCBI	Hill Braille Unit	Assist Ireland	Child Vision
Website Last Updated	22-Jun	NA	20-Apr	06-Jun
Early Intervention				✓
Education Programmes				✓
Rehabilitation Training/Therapies	✓			✓
Orientation and Mobility	✓			✓
Braille Training	✓			✓
Braille Production	✓	✓		✓
Daily Living	✓			✓
Residential Training/Living				✓
Kitchen Skills	✓			
Customer Service Skills	✓			✓
Employment Service	✓			✓
Money and Finance Services				
Technology Help Groups	✓			✓
Community Integration Prgms	✓			
Activity Centers	✓			✓
Local Support Worker	✓			
Counselling	✓			
Group Therapy				



LES MALADIES RARES SENSORIELLES

5 centres dédiés aux MR ophtalmologiques :

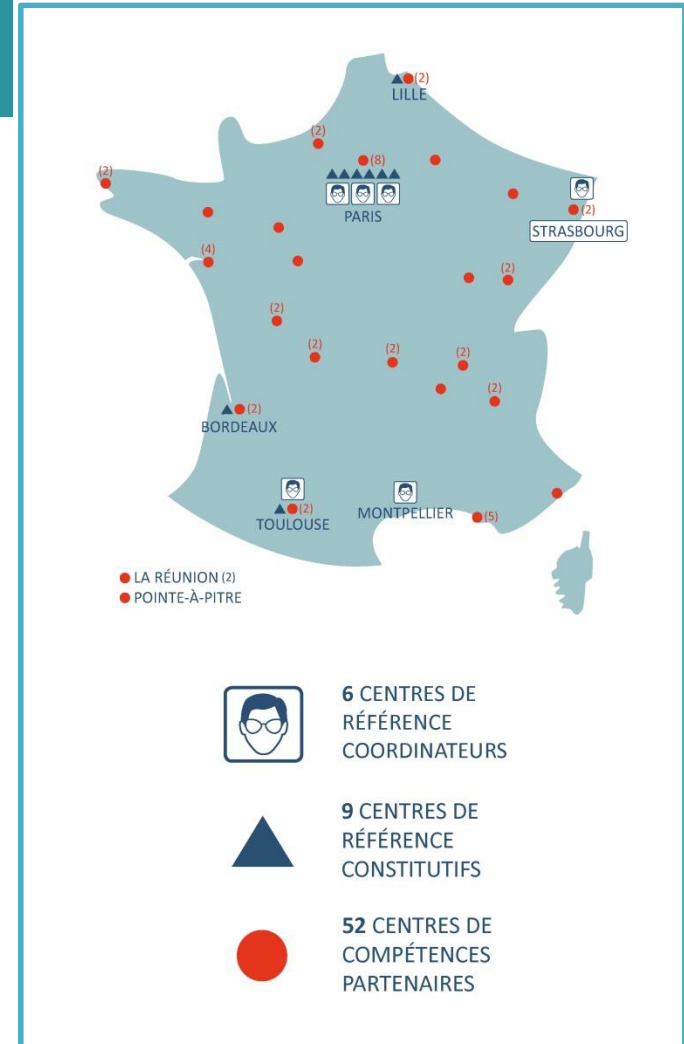
- **CARGO** : Pr H. Dollfus, HUS, Strasbourg
- **CRKN** : Pr F. Malecaze, CHU Toulouse, Toulouse
- **MAOLYA** : Pr I. Meunier, CHU Montpellier, Montpellier
- **OPHTARA** : Pr Bremond-Gignac, Hôpital Necker-Enfants Malades, Paris
- **REFERET** : Pr J. Sahel, XV-XX, Paris

1 centre surdité: Dr S. Marlin, Hôpital Necker enfants – malades, Paris

ASSOCIATIONS DE PATIENTS

Alliance Maladie Rares

- RETINA France
- Association syndrome de Wolfram
- Association Microphthalmie France
- Association Valentin Haüy
- Association contre les Maladies Mitochondriales : AMMI
- Association Gêneris
- Association Bardet-Biedl
- Association Infram'oeil
- Association Genespoir
- Association France choroïdermie
- Association Ouvrir les yeux
- Association KJER France
- Association Vision'Ere



Les structures médico-sociales

Carte interactive

Type d'atteinte :

Enfant
Ain

INSTITUT AFIS

Enfant, Adolescent, Adulte.
5 rue du Lycée
01000 Bourg en Bresse
Tél. : 04 74 22 66 85
Fax : 04 74 22 12 23

MDPH 01

Enfant, Adolescent, Adulte.
13 avenue de la Victoire
01000 Bourg en Bresse
Tél. : 08 00 88 84 44
Fax : 04 74 32 58 58
Email : mdp@cg01.fr
Site internet

SESSD

Enfant, Adolescent, Adulte.
Bât. Le Keynes, 13A, Ch du Levant
01210 Ferrey-Voltaire
Tél. : 04 50 40 41 82

1

ESMS pour enfants

▲ SESSAD

● Etablissement

ESMS pour adultes (champ CNSA)

▲ S.A.V.S.

● F.A.M.

■ M.A.S.

✕ C.R.P.

Situation des départements

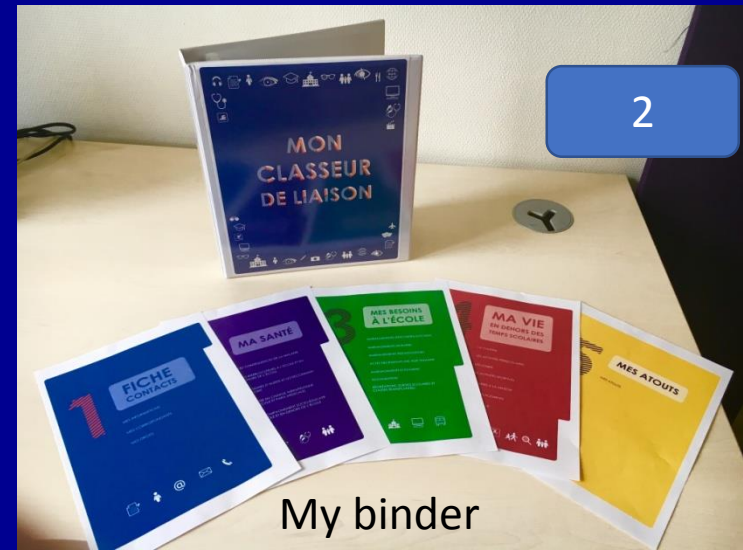
■ Départements dotés d'ESMS pour enfants

■ Départements dotés d'ESMS pour adultes

0 33 66

Source cartographique : Artique

2



My binder

tvst

DOI: 10.1167/tvst.6.3.12

Article

Optimizing the ULV-VFQ for Clinical Use Through Item Set Reduction: Psychometric Properties and Trade-Offs

Gislin Dagnelie¹, Pamela E. Jeter¹, and Olukemi Adeyemo¹; PLoVR Study Group^{1,*}

¹ Lions Vision Research & Rehabilitation Center, Department of Ophthalmology, Johns Hopkins University School of Medicine, Baltimore, Maryland, USA

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1. When receiving a printed page, how difficult is it to see which side is blank versus which side is...

- » a not applicable
- » b not difficult
- » c somewhat difficult
- » d very difficult
- » e impossible to see or do visually

2. How difficult is it for you to see a sunny spot in the garden to plant a tree or flower?

- » a not applicable
- » b not difficult
- » c somewhat difficult
- » d very difficult
- » e impossible to see or do visually

3. How difficult is it for you to recognize a family member or friend based on their size or build standing 5 ft away?

- » a not applicable
- » b not difficult
- » c somewhat difficult
- » d very difficult
- » e impossible to see or do visually



European
Reference
Network
for rare or low prevalence
complex diseases

Network
Eye Diseases (ERN-EYE)

ERN-EYE Holistic views

- ✓ Curation Orphanet /HPO done at the Saint Odile meeting



- ✓ **Launch of surveys (including TWG5)**
 - **Data repository of service** users/patients that access low vision services in each HCP and MS
 - Global **mapping of all low vision centers** across all Members states affiliated with ERN
- ✓ **Identify the best « ideal » pathway for care (age)**
- ✓ **Preparation of first guidelines/best practices :**
 - low vision evaluation
 - How to investigate a visually impaired child in the EU functional impairment,
 - Progression or non-progression of the disease....
- ✓ How to involve actively patient in research and PROMS

Holistics ERN's & ERN-EYE



- Disparities in the EU/MS on how a visually impaired/blind patient has **access to support** (allowances, reimbursement optic aids, specific training course, ...) => **regional level & national level !**
- Disparities on the **professionals availability** and organisation in the HCPs and organisation in each MS => **regional level & national level !**
- Commonalities in the **care pathways** (ie genetic testing but also specificities : **independance** is the key word
- => ERN-EYE Communication strategy (information on low vision and blindness) on going (but expensive)
- => ERN-EYE ready to contribute to harmonization with other ERNs and INNOVcare : example develop in the EU case manager's, ...



European
Reference
Network

for rare or low prevalence
complex diseases

Network
Eye Diseases (ERN-EYE)





Thank
you