Communicating diagnosis and how to deal with results :
Addressing the needs of patients, families, and healthcare professionals

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ITHACA

ERN BOND
EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES

European Reference Networks | ERN CRANIO

European Reference Network | Neuromuscular Diseases (ERN EURO-NMD)

SJD Sant Joan de Déu Barcelona · Hospital

Vall d'Hebron Hospital | Vall d'Hebron | Vall d'Hebron Institut de Recerca

European Reference Networks

Disclosures

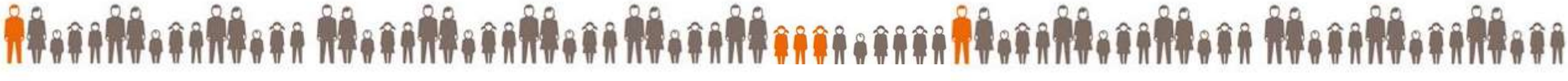
- Grant support to conduct clinical trials on SMA from Ionis/Biogen
- Serves as a consultant to AveXis, Novartis, Biogen, Biologix, Cytokinetics, Roche
- Serves as a scientific/medical advisor for non profit organizations such as FAME Chile, Famiglie SMA Italy, Familias SMA Argentina, ASEM Federation, FUNDAME, SMA Europe, TREAT-NMD, EURORDIS, IFSBH

Rare diseases

- Low prevalence
- Rare diseases affect around **6-8%** of the worldwide population
- Over 300 million people worldwide / over 3 million in Spain
- 7,000 conditions
- **80%** genetic origin





HOW IS GENETICS IN CLINICAL PRACTICE?

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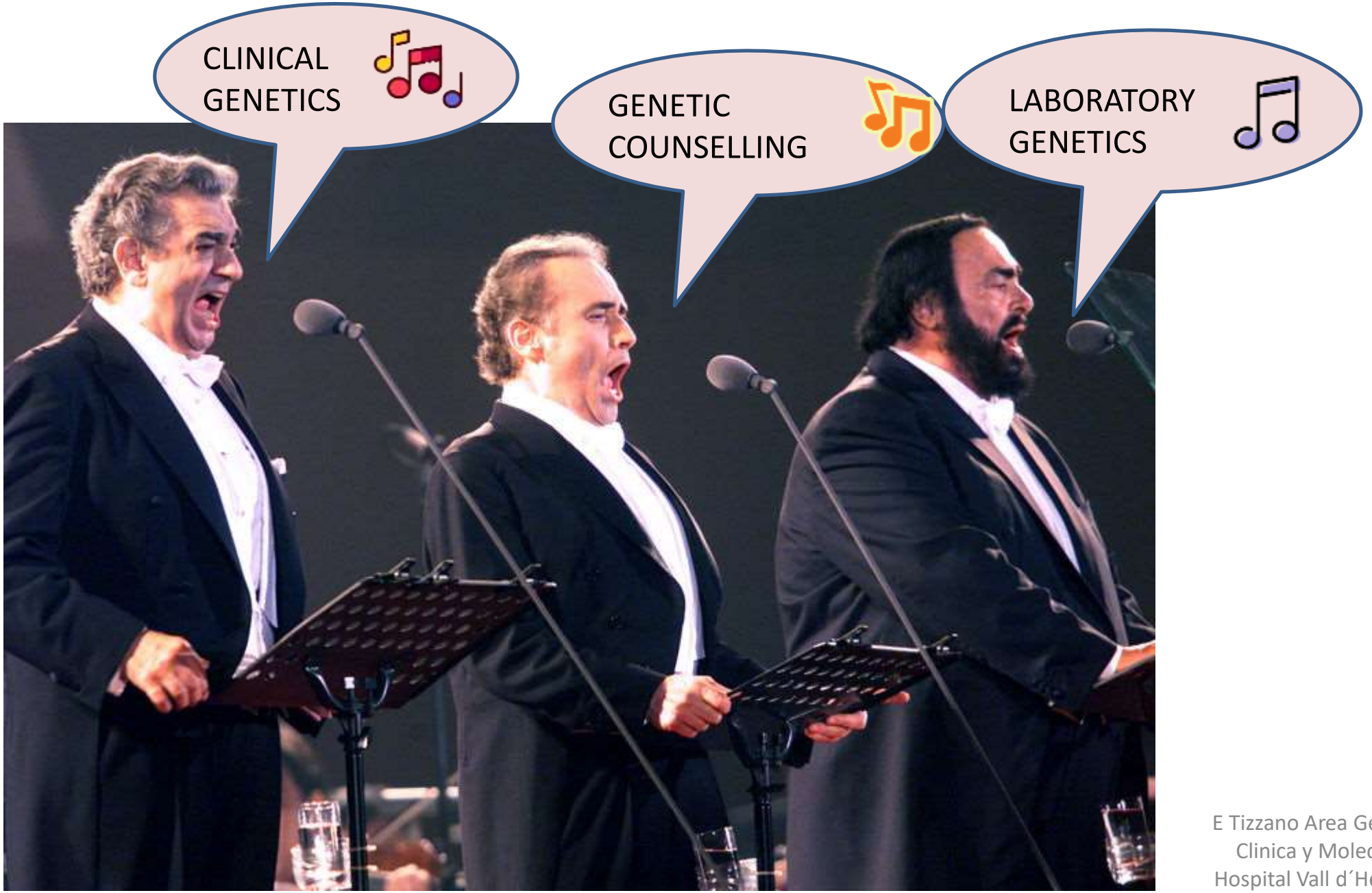


E Tizzano Area Genetica Clinica y Molecular Hospital Vall d´Hevbron

Medical Genetics

Clinical Genetics

Detects malformations and dysmorphia
and its implications
Recognizes common genetic entities and specific syndromes
Interact with other specialists for diagnosis, follow-up, treatments
Collaborate in the development of advanced therapies

Genetic Counselling

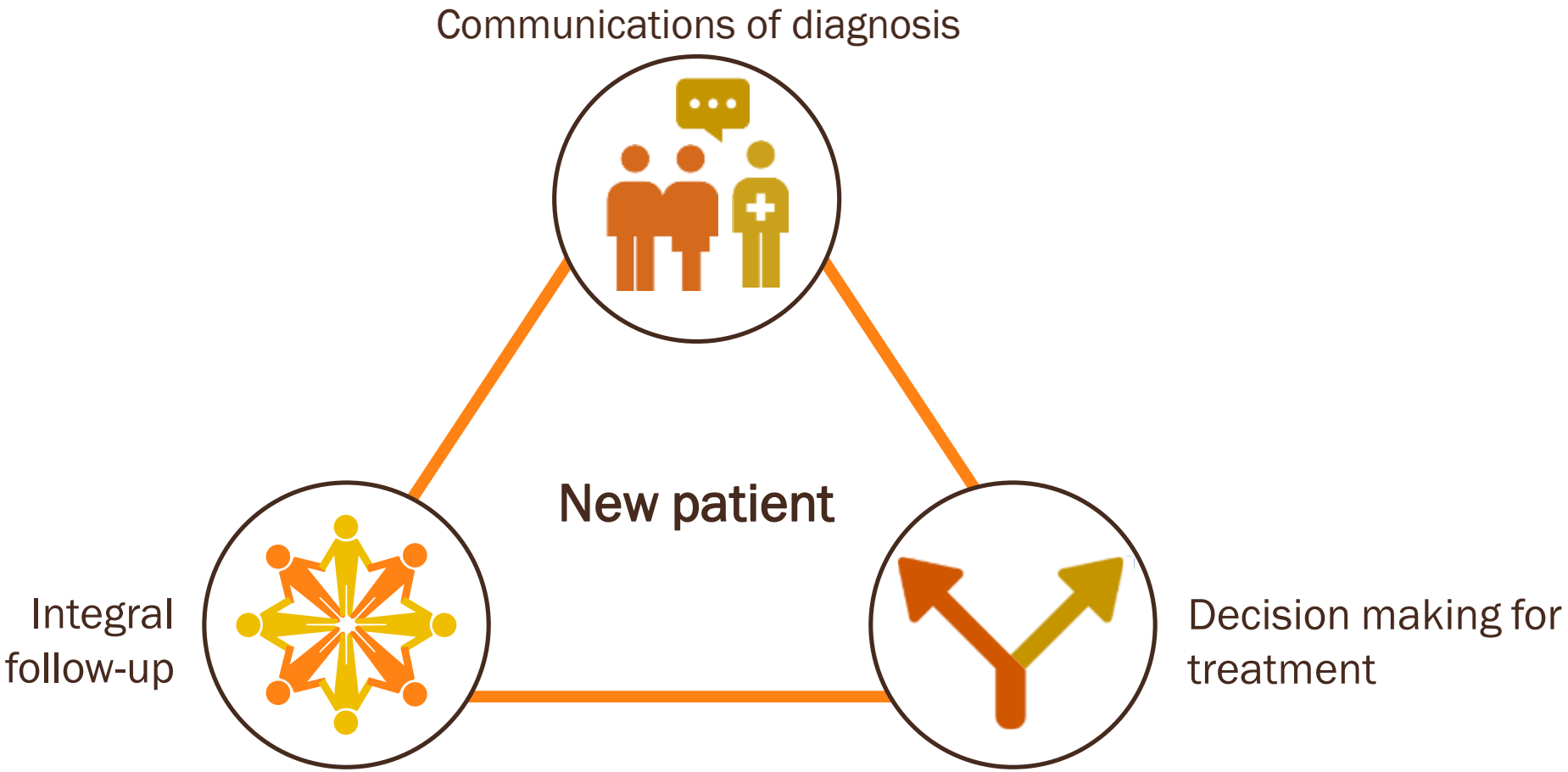
Know the types of inheritance and the concepts and processes of genetic counseling, including communication with the patient and adaptation to a certain condition in the family and
the importance of correctly reporting and giving advice regarding the results obtained in genetic tests

Laboratory Genetics

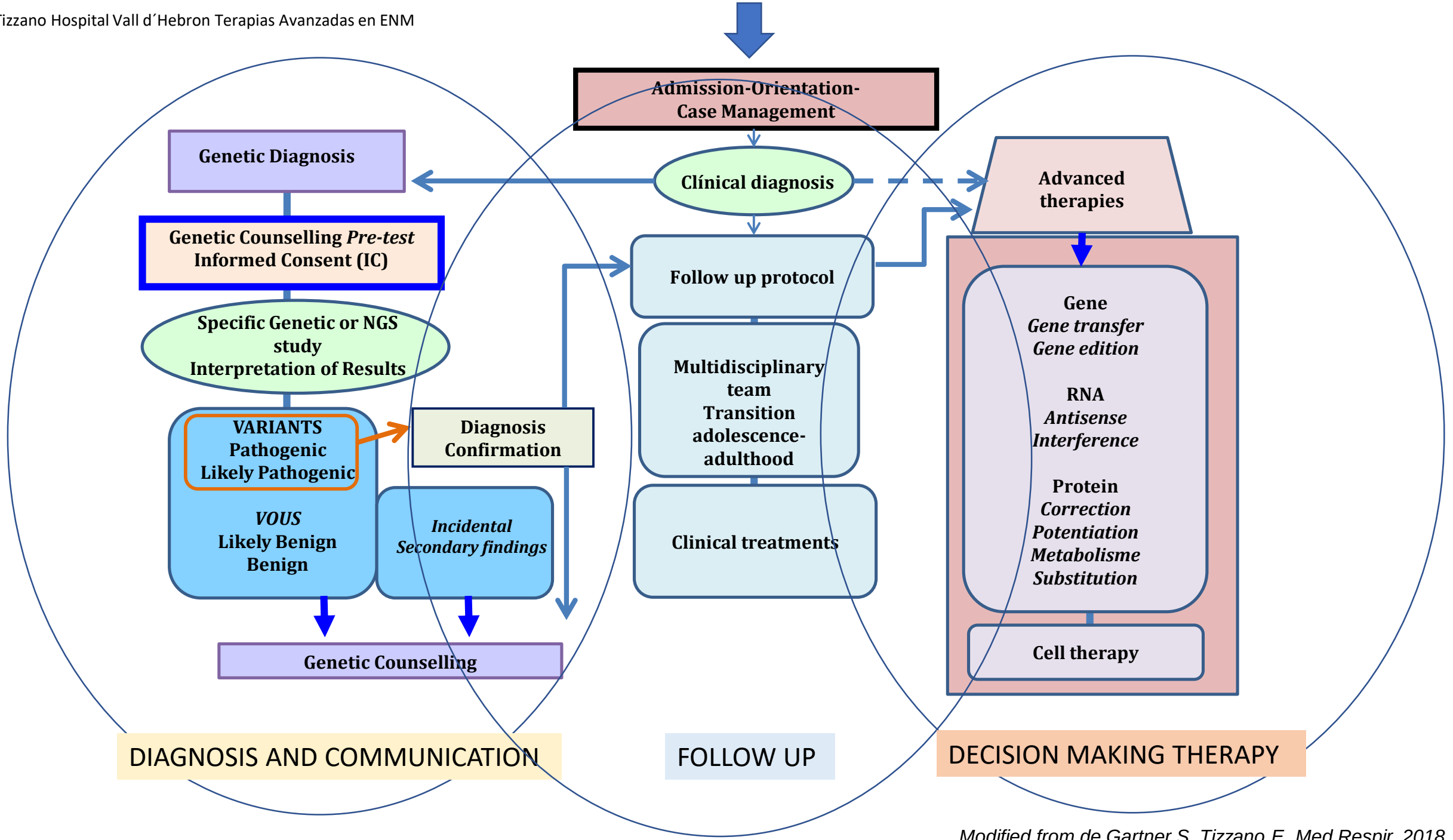
Perform genetic tests:
Chromosome studies
Array CGH
Number of copies
Sanger sequencing
Massive (NGS) sequencing
Interprets data from genetic and genomic studies

A complete approach to the genetic problem of the patient and the family

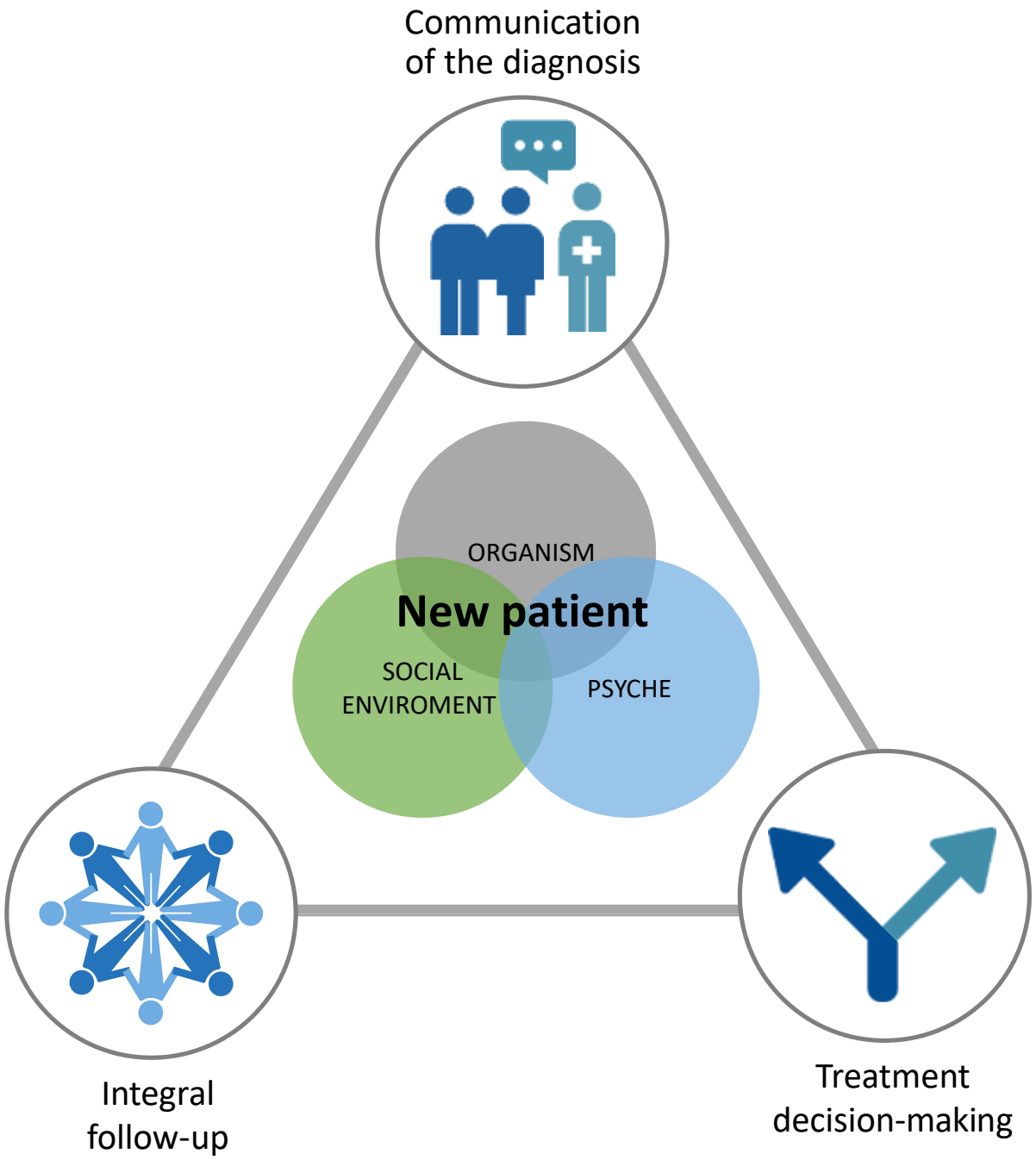
Main tasks when facing a new person with a genetic disorder



Rovira et al. 2023



Modified from de Gartner S, Tizzano E, Med Respir, 2018



Consider three main aspects of an individual

The three main tasks include communication of diagnosis, integral follow-up and decision making of treatment that as a whole should consider a balance in the three main aspects of an individual:
organism,
psyque
and social environment

The first task

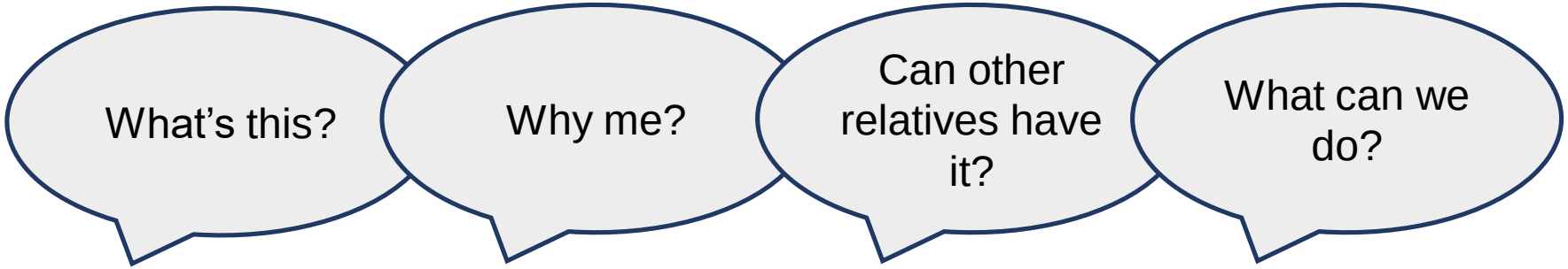
Communicating the diagnosis

Give information gradually, a small team with a pre- and post-diagnostic test interview[1]

- Need time to assimilate the diagnosis[2]
- Will not retain all the information provided[2]
- Denies of the disease is usually the first reaction[3]

1. Tizzano E, unpublished data and speaker's opinion (in preparation) . 2. Havermans T, et al. J Cyst Fibros. 2015;14:540-6. 3. Ferrario SR, et al. Neuropsychiatr Dis Treat. 2017;13:909-16.

Genetic information: peculiarities



It is a very hard emotional situation, with family implications and many new concepts and information to assimilate

What is genetic counseling?

*Genetic counseling is a **communicative process** that addresses the human problems associated with the **risk** of occurrence or recurrence of a genetic disease in a family.*

Evolution of genetics

Health perception

Genetic information: peculiarities

Characteristics of genetic information

↓

Potentially stressful and distressing

↓

Direct implications for **family** members

Lifetime genetic condition

Pre-symptomatic diagnoses or predictions of susceptibility to pathologies

Complexity of information (inheritance, risk, penetrance)

The stress and anxiety caused by the possibility of a genetic alteration in the family makes understanding even more difficult

What is genetic counseling in the context of communication of diagnosis ?

- Comprehension of **medical concepts**.
- Interpretation of family and medical histories to assess the chance of disease **occurrence** or **recurrence**.
- **Education** about inheritance, testing, management, prevention, resources and research.
- Choose **the most appropriate action** based on risk, available options and ethical and/or religious principles.
- In case of undergoing a **genetic study**: counseling prior to the study (implications, advantages and limitations), post-test counseling.

Facilitate decision making and act in accordance with the made decision.

Genetic counseling process

Obtain medical histories to determine genetic risks

Inform about the disease (features, natural history, genetic and environmental factors…)

Assess and communicate risks

Explain clinical implications (management, reproductive options…)

Genetic testing

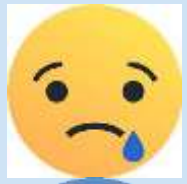
Provide support throughout the whole process, support in decision making

PATIENT / FAMILY

Negation

Discomfort

Decision



Expectation

Sadness

Acceptation

Hope

Patient and parents' emotional coping abilities and cognitive capacities are challenged.

PROFESSIONAL HEALTHCARE

Fluid and transparent communication

Skilled team

Emotional support

Promote opportunity recognition

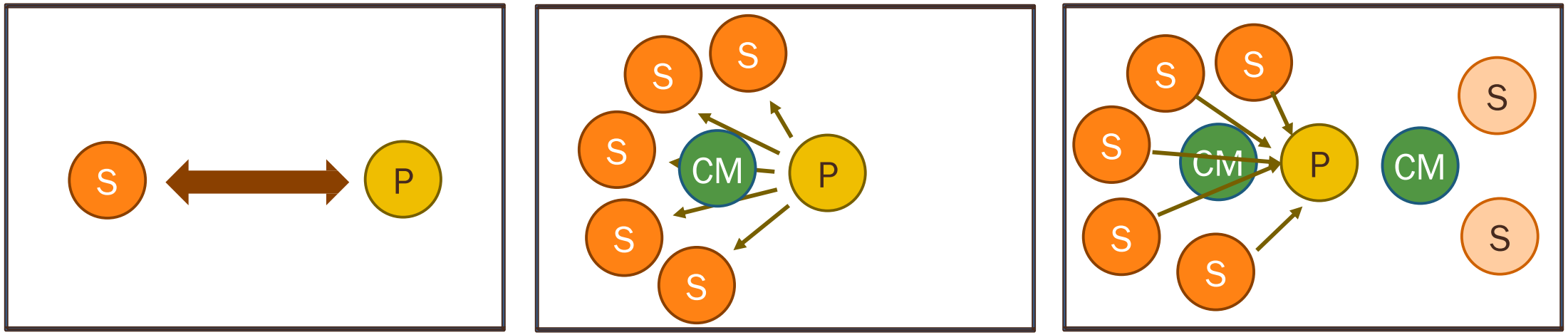
Efforts to minimize psychological stress and help to improve personal control.

Issues and attitudes of the patient/family when communicating diagnosis and alternatives to be conducted/approached by the healthcare professional responsible for the process of communication

The second task

Integral follow-up

Moving from traditional monographic consultation towards implementation of integrated and multidisciplinary clinics with the collaboration of case Managers



Monographic consultation

Interdisciplinary clinics

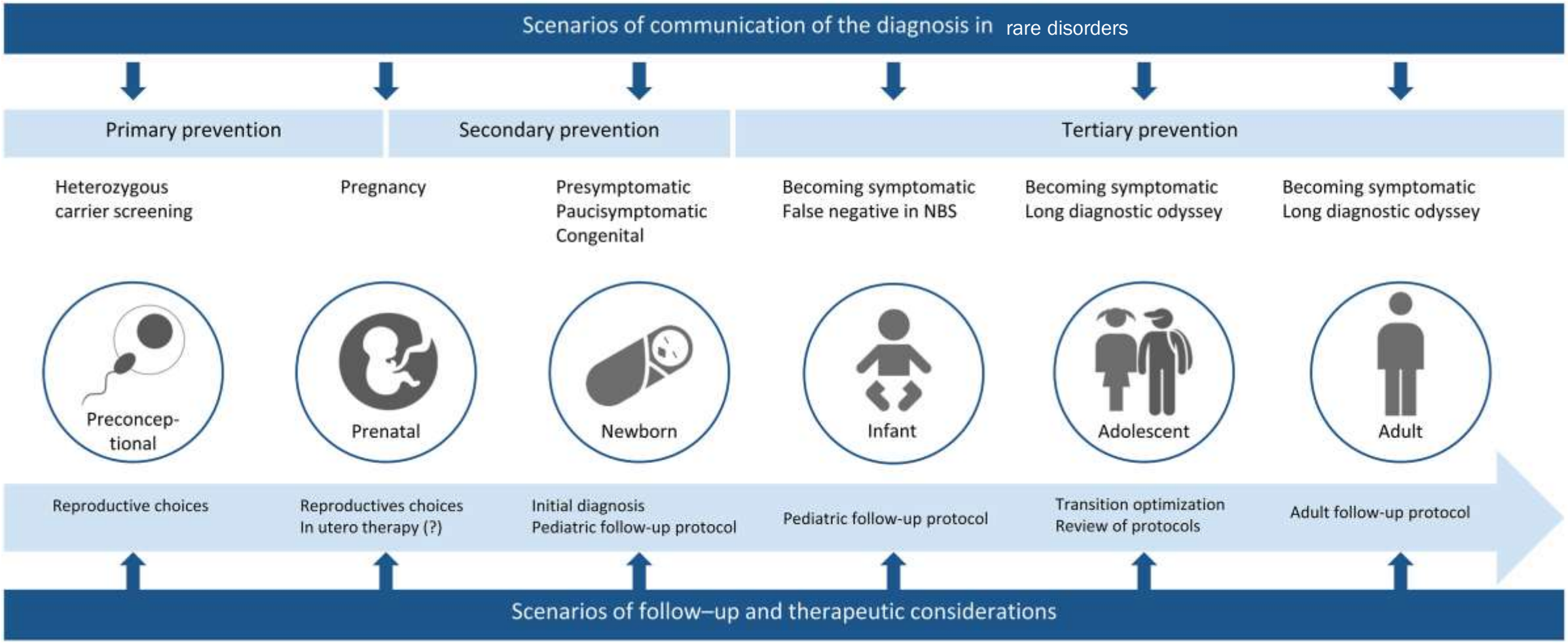
Multidisciplinary clinics

S S Specialist

P Patient or caregiver

CM Case Manager

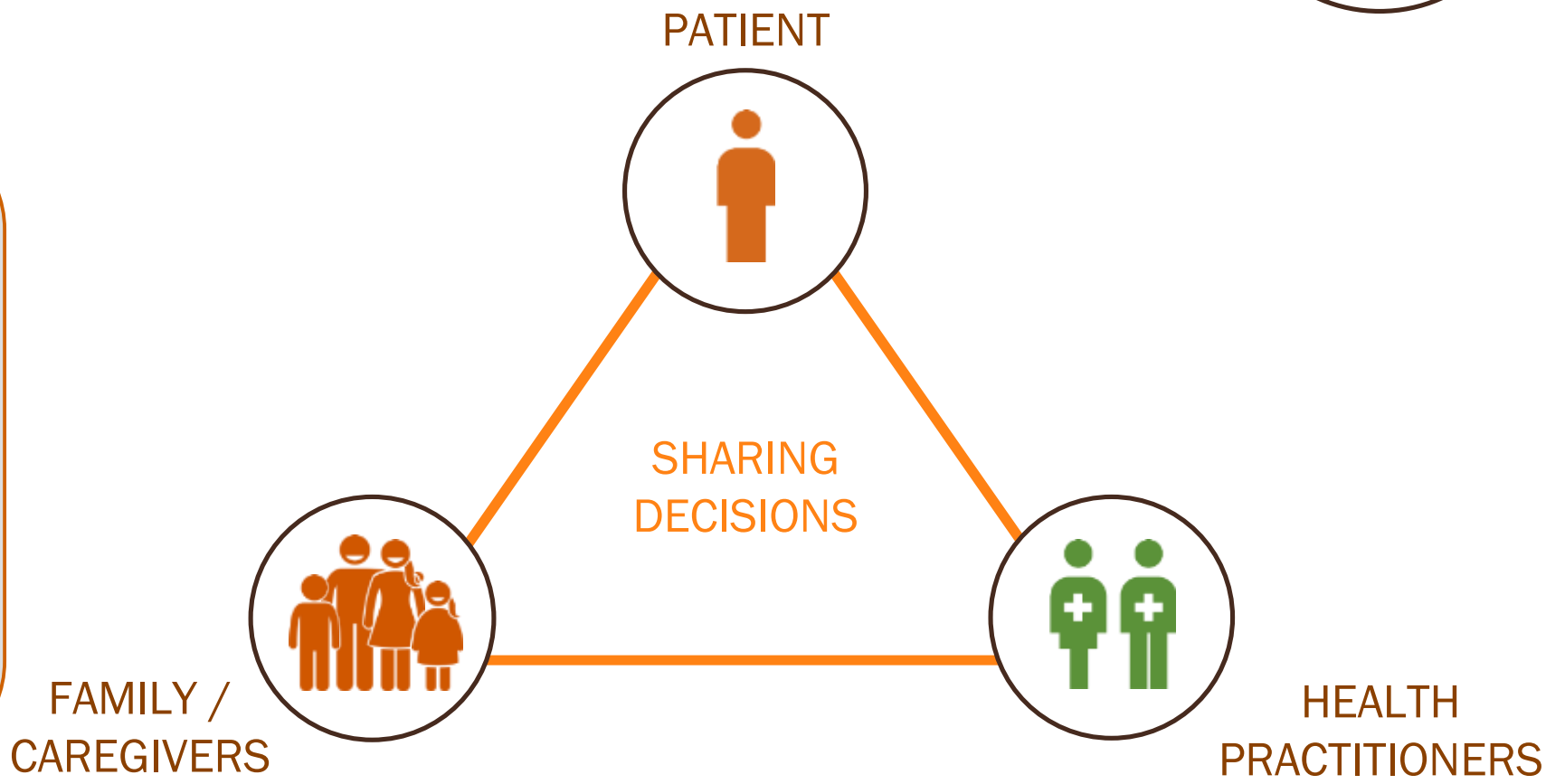
Tizzano E. unpublished data and speaker's opinion (in preparation).


Scenarios of communication of the diagnosis in rare disorders
Primary prevention
Secondary prevention
Tertiary prevention
Heterozygous carrier screening
Pregnancy
Presymptomatic Paucisymptomatic Congenital
Becoming symptomatic False negative in NBS
Becoming symptomatic Long diagnostic odyssey
Becoming symptomatic Long diagnostic odyssey
Preconcep-tional
Prenatal
Newborn
Infant
Adolescent
Adult
Reproductive choices
Reproductives choices In utero therapy (?)
Initial diagnosis Pediatric follow-up protocol
Pediatric follow-up protocol
Transition optimization Review of protocols
Adult follow-up protocol
Scenarios of follow–up and therapeutic considerations

The third task

Decision making for treatment

- Consensus for follow-up and treatment[1]
- Managing expectations of the patients and the family / caregivers[1,2]



1. Tizzano E. Rev Med Clin Condes. 2018;29:512-20. 2. Serra-Juhe C, Tizzano EF. Eur J Hum Genet. 2019 [ePub ahead of print].

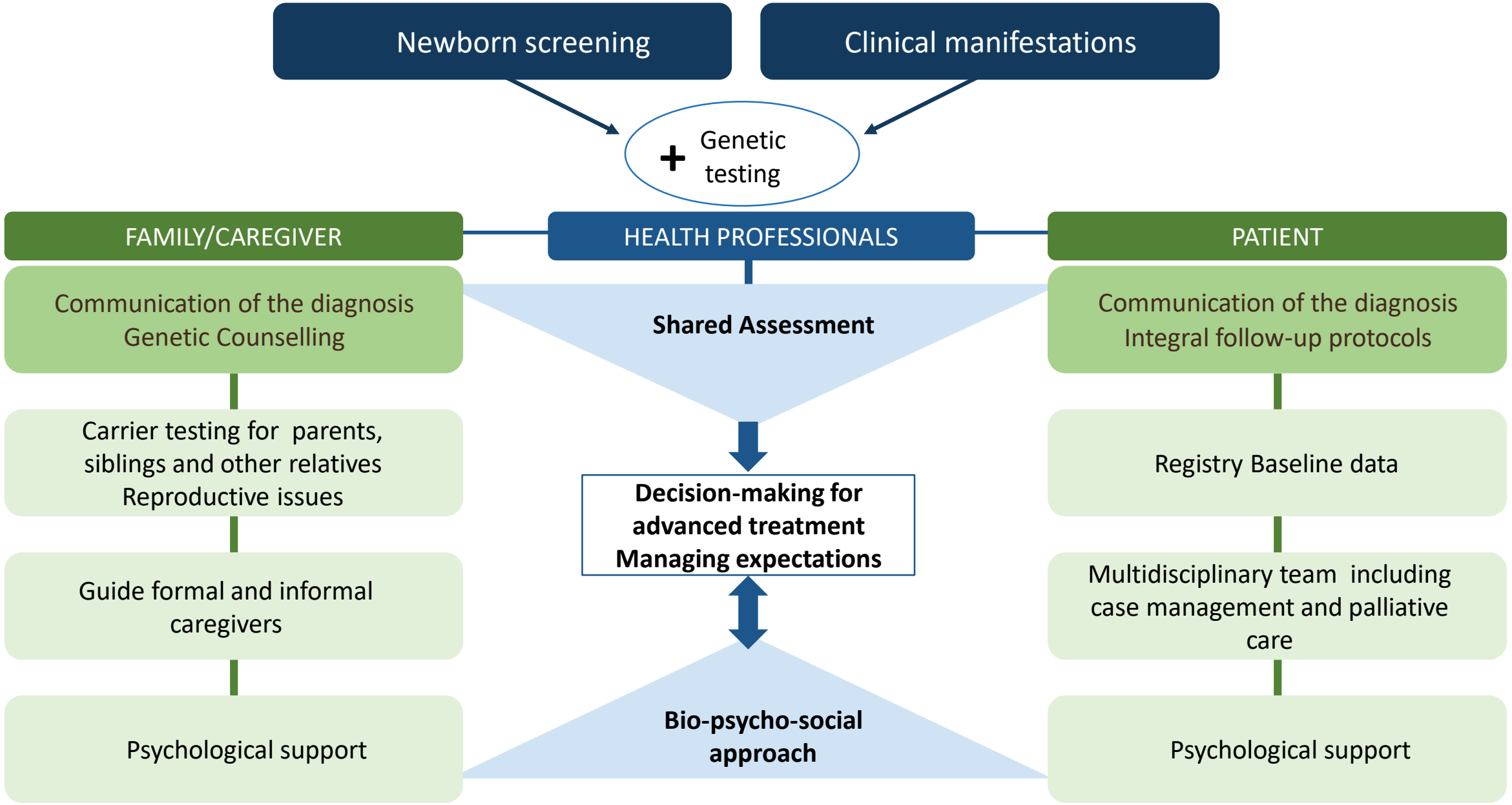


Figure 3 Legend. The processes to be accounted by health professionals facing a new patient. New patients may be detected at

MANAGING EXPECTATIONS WHEN THERE IS A DIAGNOSIS AND A POSSIBLE TREATMENT

Patients give importance to the emotional impact of the experience of receiving medical care.

Consider the different perspectives of patients/family/caregivers and physicians[2]

Report on data limitations in some cases and the importance of standards of care[1,3]

Achieving consensus on outcomes that are meaningful to the patient[4]

Create confidence in the multidisciplinary team[4,5]

CAUTION: Patients may encounter inaccurate information or information directed at patients in other countries where medical care may be different.

1. Tizzano EF, Finkel RS. Neuromuscul Disord. 2017;27:883-9. 2. Lorenzetti RC, et al. Am Fam Physician. 2013;87:419-25.
3. Wang CH, et al. J Child Neurol. 2007;22:1027-49. 4. Rovira et al. in preparation . 5. Nunlist MM, et al. Fam Pract Manag. 2016;23:21-4.

Levels of prevention in genetic disordsers

HEALTH COSTS

Tertiary prevention

Treat all patients with manifesting disease

Mainly influence in standard of care and evolving phenotypes

Secondary prevention

Treat all pre-symptomatic cases detected through newborn screening

Mainly influence in burden and development of the disease

Primary prevention

Genetic counseling in all carriers detected through population screening

Mainly influence in the incidence and prevalence

SMA, spinal muscular atrophy.

1. Serra-Juhe C and Tizzano EF. Perspectives in genetic counseling for spinal muscular atrophy in the new therapeutic era: early pre-symptomatic intervention and test in minors. Eur J Hum Genet . 2019.

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REVIEW

The diagnosis communication process in spinal muscular atrophy: A cross-cutting view of the new challenges facing the therapeutic era

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WHEN LANGUAGE IS A BARRIER

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EDUCATIONAL INITIATIVE FOR PATIENTS WITH RARE DISORDERS AT HOSPITAL VALL HEBRON SINCE 2017 in SPANISH

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ORIGINAL ARTICLE

National Society of Genetic Counselors | WILEY

Beyond the disease itself: A cross-cutting educational initiative for patients and families with rare diseases

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Abstract

Rare diseases (RDs) as a whole affect a huge number of individuals although each specific condition comprises a low number of individuals. As a consequence, funds allocated to expand research to all conditions are often limited. Several initiatives have emerged to invest more resources for research in RDs, but patients express unmet needs regarding educational initiatives, awareness support, and psychosocial resources. We developed an educational training program in the format of weekly sessions covering basic medical scientific knowledge and psychosocial aspects of RDs. The aim of this initiative was to assess its overall impact regarding knowledge, psychological issues, and participant satisfaction. Items were evaluated through surveys before and after the sessions. Here, we report the experience and impact of two editions of this initiative with a total of 37 participants. Our results show improvements in knowledge and better management of the psychological impact. Moreover, participants were able to exchange experiences and concerns, most of which were shared even though the RDs were different. Overall, the program was evaluated by

THANK YOU FOR YOUR ATTENTION!

Questions???