Health, habilitation and rehabilitation in the CRPD

Perspectives of people living with a rare disease & disabilities

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Work Forum on the Implementation of the UN Convention on the Rights of Persons with Disabilities in the EU and the Member States
Brussels, 29 May 2017

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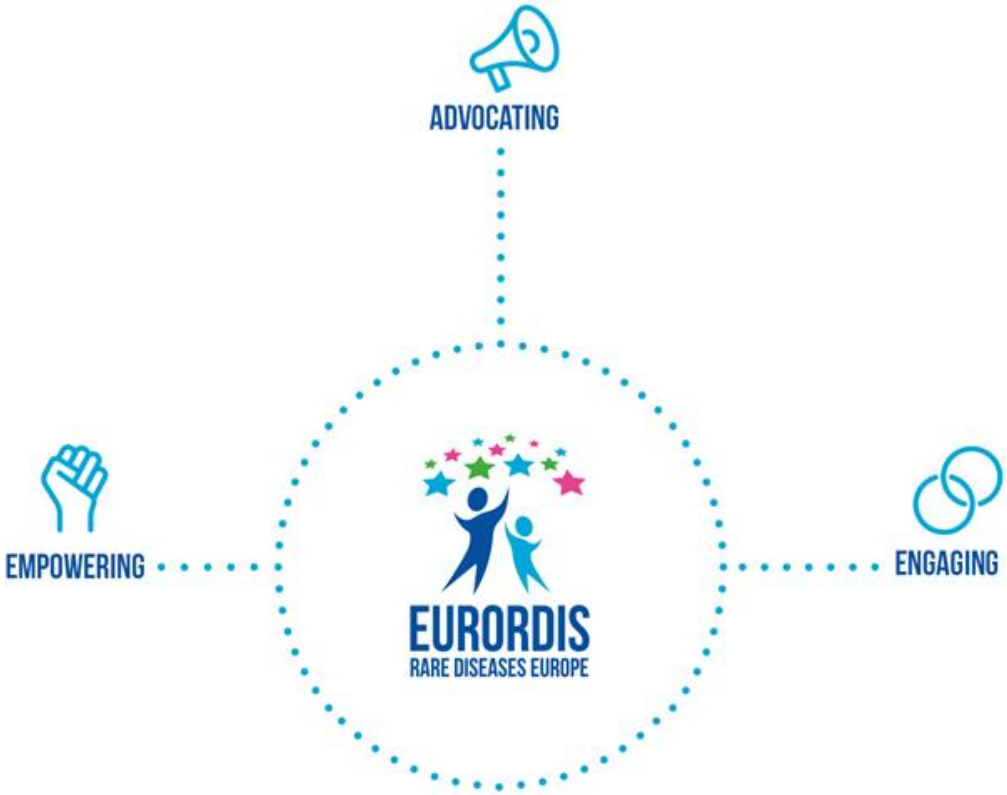
EURORDIS-Rare Diseases Europe

Mission: *to work across borders and diseases to improve the lives of people living with a rare disease*

- ✓ **800+** member patient organisations
- ✓ **69 countries** (28 EU countries)



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What is a rare disease?

OVER
6000
distinct rare diseases

Each one affects fewer than
1 IN
2000
PEOPLE

All together, an estimated
30
MILLION PEOPLE
are living with a rare disease in Europe

Expertise, knowledge, information on diseases and their consequences are **scarce and difficult to access**

Rare, complex, chronic, disabling, progressive, degenerative, often life-threatening

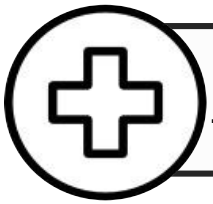
NO
CURE
for the vast majority of diseases and few treatments available

They are **geographically scattered** and often **isolated**

Few experts, geographically **scattered**

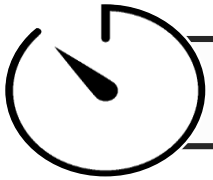
Research is fragmented

Challenges faced by people living with a rare disease & their carers



Challenges accessing health care: undiagnosed, misdiagnosed, wait years for diagnosis; rejection by HC professionals; difficulties accessing treatment

Significant impact on everyday life activities: impairments, activity limitations, participation restrictions

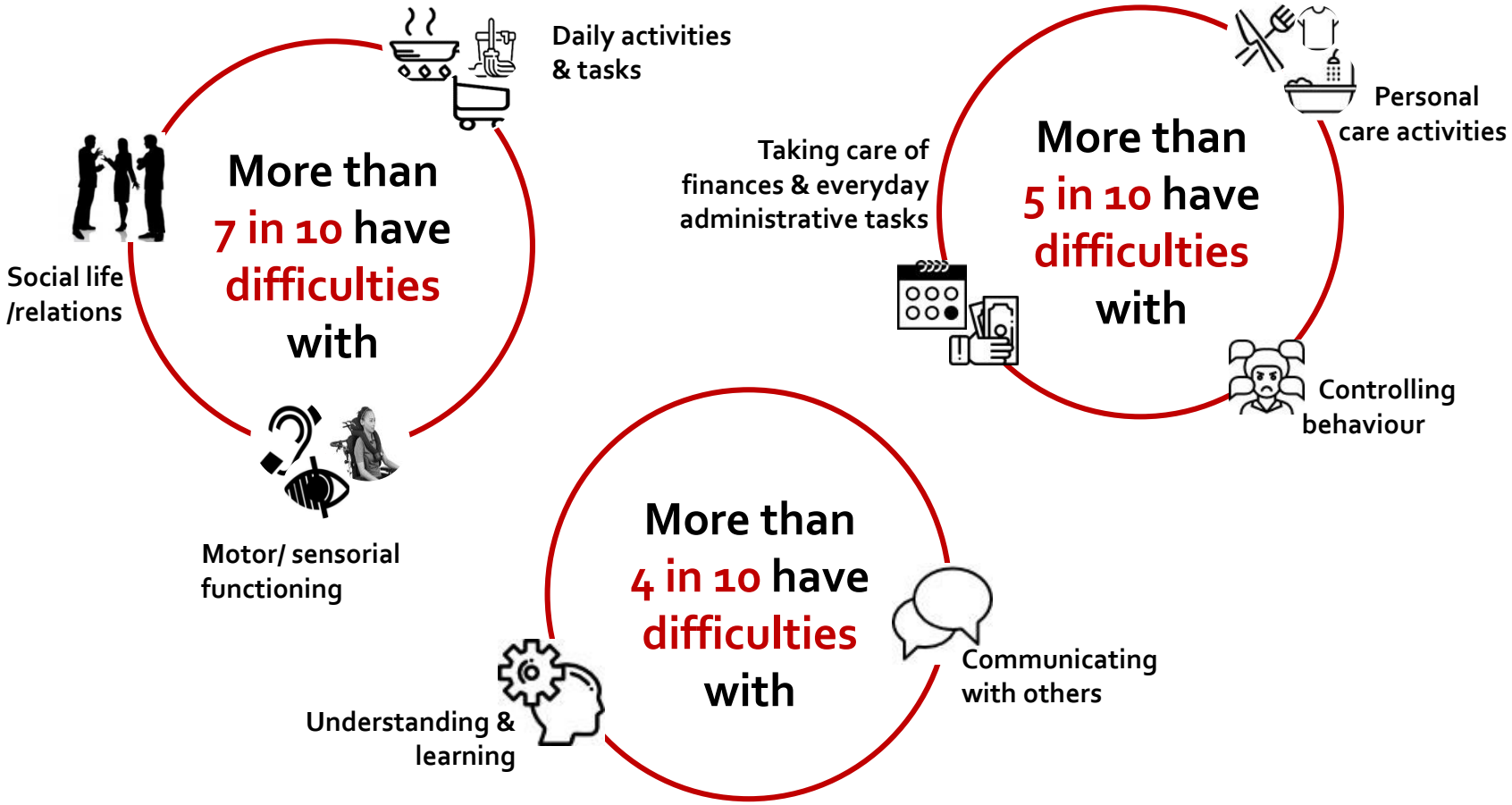


Significant time and care burden, which falls heavily on women (often main the main carer)

Strong impact on work-life: absence from work, hampered professional activity, economic burden

Impact on mental health and well-being

People living with a rare disease have difficulties with several Activities of Daily Living



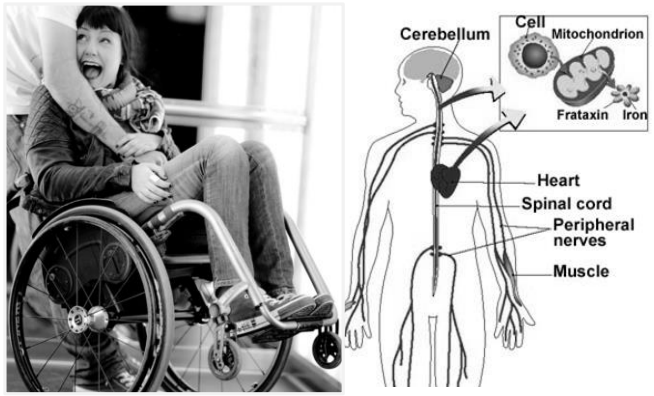
"Disabilities is an umbrella term, covering impairments, activity limitations, and participation restrictions" WHO

Symptoms vary & can be invisible in many ways

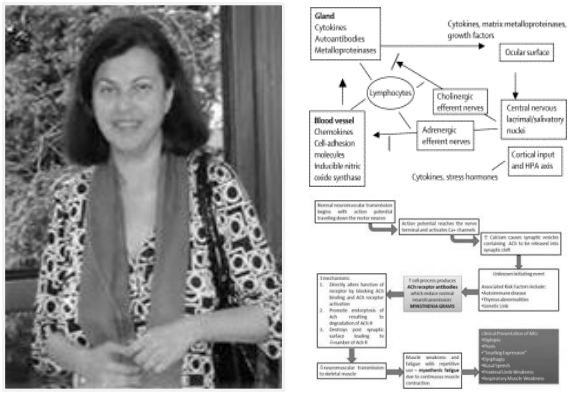
“**One day you appear completely healthy, the next day you are sick**, and two days later you appear completely normal again” Female, Spain

“**I don't look ill but am very ill** with a condition which no one understands or has heard of” Female, United Kingdom

Katerina is in a wheel chair…
She has a complex neurodegenerative disease *(Friedreich's ataxia)*



N.R. looks healthy…
She has two autoimmune diseases…
(Myasthenia Gravis & Sjogren Syndrome)



Access to healthcare and habilitation for people living with a rare disease: complex and hard to manage

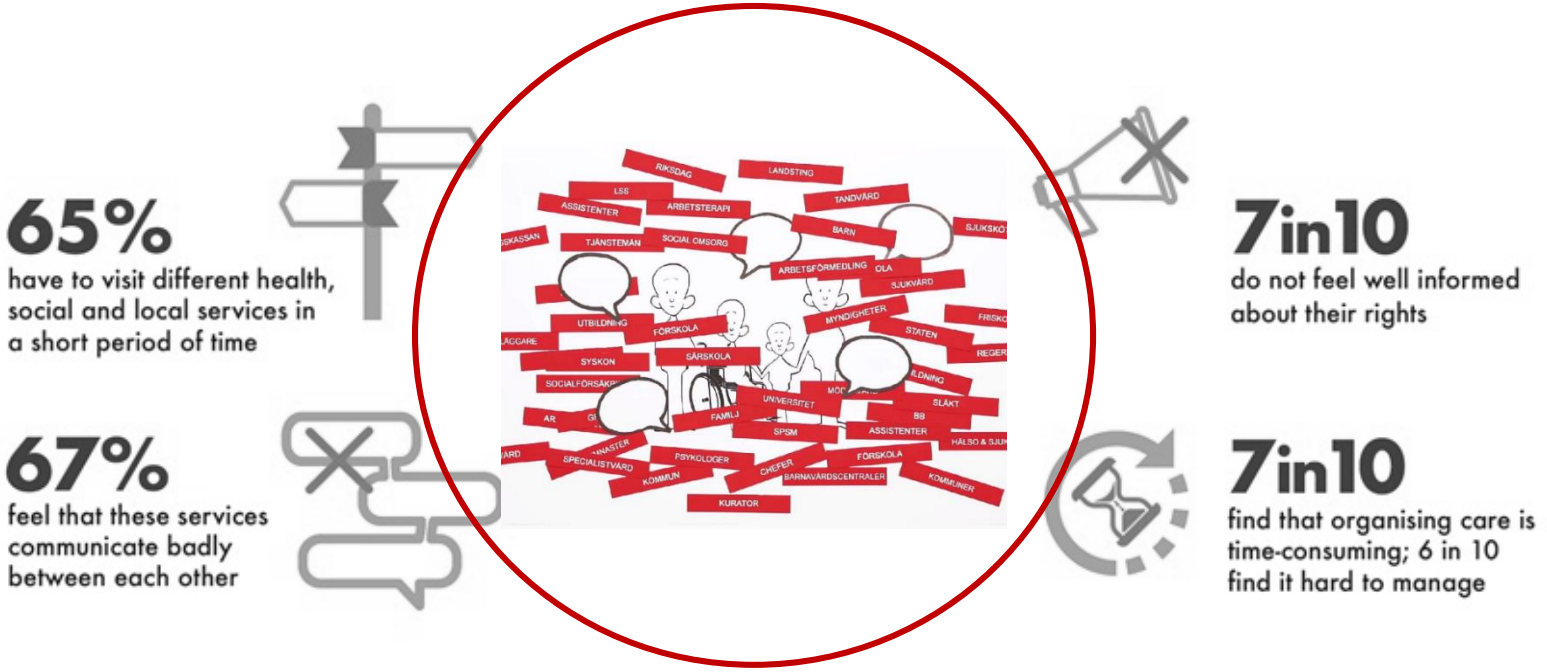


Image used for illustration purposes. Source: [Nationella Funktionen Sällsynta Diagnoser](), Sweden

People living with a rare disease have uncovered health and rehabilitation needs

48%

Have **uncovered needs** **regarding** **access to rehabilitation and therapies** *(21% have access but find they are not enough to cover their needs; 27% do not have access but find that they would need it)*

28%

Have **uncovered needs** **regarding** **access to health services** *(14% have access but find that they are not enough to cover their needs; 14% do not have access but find that they would need it)*

1 in 5

Experienced **rejection by healthcare professionals,** often due to the complexity of the disease

People living with a rare disease have uncovered needs regarding disability benefits

34% of respondents submitted to disability assessment **find that the % of disability assigned to them too low**

50% have **uncovered needs regarding access to disability benefits** *(28% have access but find that these are not enough to cover their needs; 22% do not have access but find that they would need it)*

Examples of good practices to support access to health, rehabilitation and habilitation

Initiatives supporting, empowering and engaging people living with rare diseases and their carers

European policy

- Commission Communication (2008): Rare Diseases: Europe's Challenges
- Council Recommendation on an action in the field of Rare Diseases (2009)
- EUCERD and Commission Expert Group on Rare Diseases

https://ec.europa.eu/health/rare_diseases/overview_en

Policy: Commission Expert Group Recommendations to Support Integration of RD into Social Policy

Unanimously adopted by representatives of all EU Member States (April 2016)

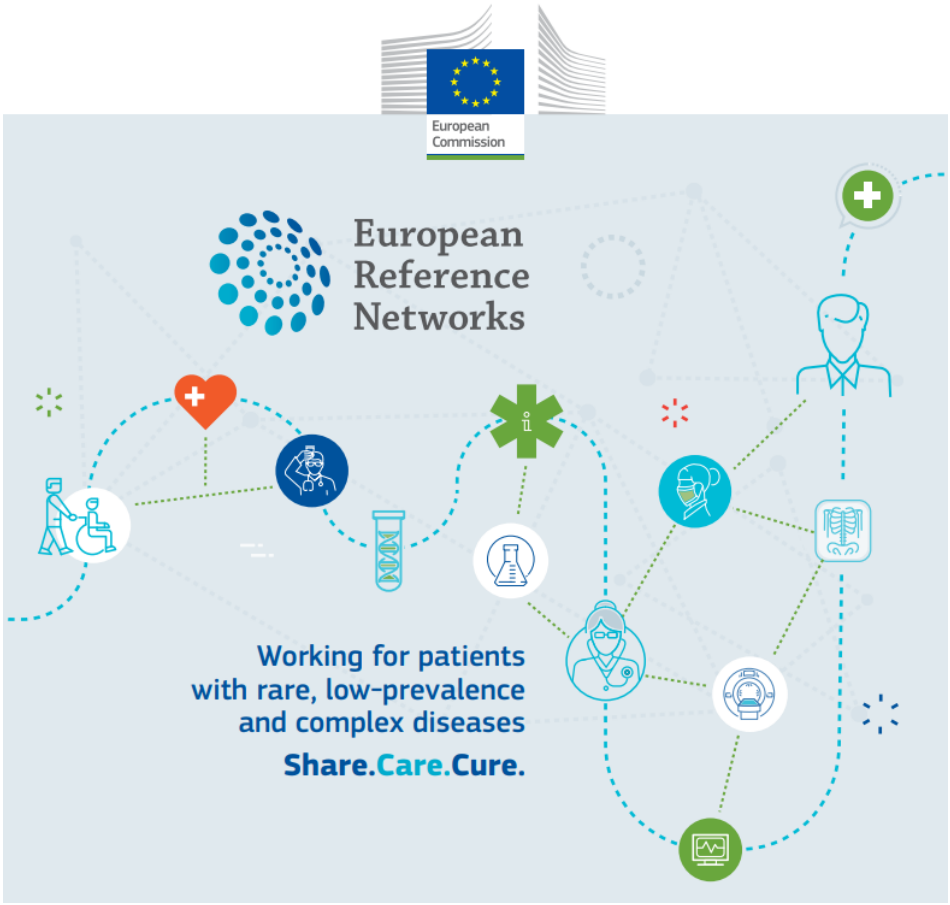
4. Member States (MS) should promote measures that facilitate **multidisciplinary, holistic, continuous, person-centred and participative care provision** to people living with rare diseases, supporting them in the **full realisation of their fundamental human rights**

8. Rare Disease specificities should be integrated into national systems assessing a person's level of functioning, in line with the United Nations Convention on the Rights of Persons with Disabilities

https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/recommendations_socialservices_policies_en.pdf

Healthcare: European Reference Networks (ERNs)

Networks harness the collective knowledge and experience of experts, focusing on a common goal to drive **improve access to diagnosis and treatment** by providing **high-quality healthcare**



- **Virtual healthcare**: specialist advice
- **Knowledge generation**: sharing experience and expertise, research and innovation
- **Knowledge dissemination**: clinical guidelines, healthcare pathways, education and training

Habilitation: European Network of Resource Centres for Rare Diseases (RareResourceNet)

Advancing holistic high quality care and services for people living with rare diseases across Europe

- **One-stop shop style services**
- **Holistic care and support**
- **Complementary to health care and social services**
- **Create a bridge between people living with a rare disease/families and stakeholders providing health care, social care and social support**

Examples of resource centres for rare diseases:
Ågrenska, Sweden
Frambu, Norway
NoRo, Romania
List of resource centres in Europe

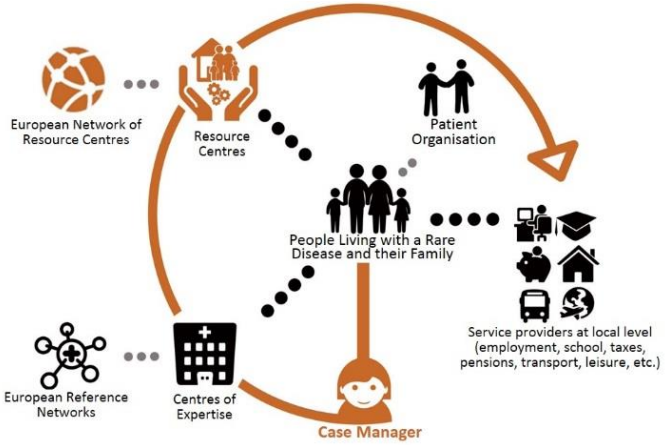
European projects: supporting integrated-holistic care

These projects co-organised multistakeholder workshop on holistic care for people living with a rare disease (April 2018): presentations and outcomes available here

INNOVCare

2015-2018

- Co-funded by EC-DGEMP
- Focus: integrated and holistic care
- Pilot of **case management for rare diseases** and other complex conditions



RD ACTION
DATA AND POLICIES FOR RARE DISEASES

2015-2018

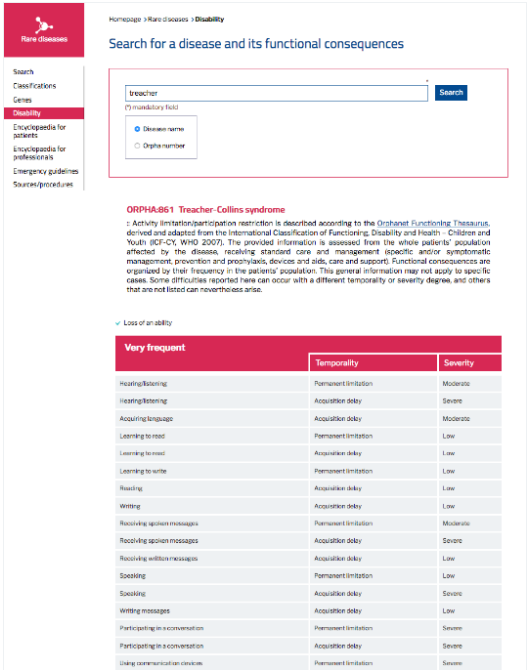
- Co-funded by EC-DGSANTE
- European Joint-Action for Rare Diseases
- Focus: supporting the development and implementation of European and national policies/recommendations on policy, information and data for rare diseases

www.innovcare.eu

www.rd-action.eu

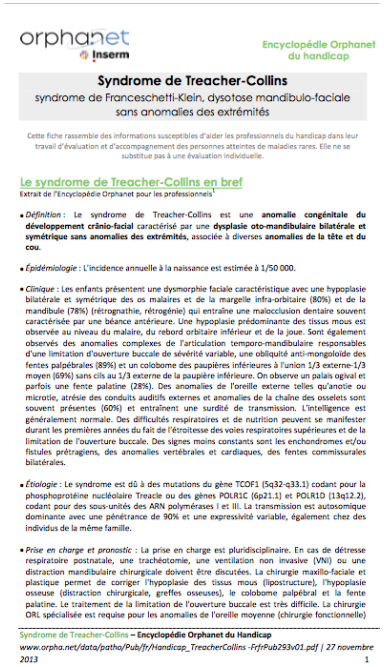
Orphanet Disability Project

- Provide information to health-social sector



Orphanet Functionning Thesaurus

- Based on ICF (WHO 2007)
- 139 items in 10 domains + environment items
- Allowing to describe: frequency, severity, temporality
- Annotations and texts made with contribution of: medical experts, medical-social sector professionals, patient organisations


orphanet
Inserm
Encyclopédie Orphanet du handicap

Syndrome de Treacher-Collins
syndrome de Franceschetti-Klein, dysotose mandibulo-faciale sans anomalies des extrémités

Cette fiche rassemble des informations susceptibles d'aider les professionnels du handicap dans leur travail d'évaluation et d'accompagnement des personnes atteintes de maladies rares. Elle ne se substitue pas à une évaluation individuelle.

Le syndrome de Treacher-Collins en bref
Extrait de l'Encyclopédie Orphanet pour les professionnels[1]

- *Définition :* Le syndrome de Treacher-Collins est une **anomalie congénitale du développement crânio-facial** caractérisé par une **dysplasie oto-mandibulaire bilatérale et symétrique sans anomalies des extrémités**, associée à diverses **anomalies de la tête et du cou.**
- *Épidémiologie :* L'incidence annuelle à la naissance est estimée à 1/50 000.
- *Clinique :* Les enfants présentent une dysmorphie faciale caractéristique avec une hypoplasie bilatérale et symétrique des os malaires et de la margelle infra-orbitaire (80%) et de la mandibule (78%) (rétrognathie, rétrogénie) qui entraîne une malocclusion dentaire souvent caractérisée par une béance antérieure. Une hypoplasie prédominante des tissus mous est observée au niveau du malaire, du rebord orbitaire inférieur et de la joue. Sont également observés des anomalies complexes de l'articulation temporo-mandibulaire responsables d'une limitation d'ouverture buccale de sévérité variable, une obliquité anti-mongoloïde des fentes palpébrales (89%) et un colobome des paupières inférieures à l'union 1/3 externe-1/3 moyen (69%) sans cils au 1/3 externe de la paupière inférieure. On observe un palais ogival et parfois une fente palatine (28%). Des anomalies de l'oreille externe telles qu'anotie ou microtie, atrésie des conduits auditifs externes et anomalies de la chaîne des osselets sont souvent présentes (60%) et entraînent une surdité de transmission. L'intelligence est généralement normale. Des difficultés respiratoires et de nutrition peuvent se manifester durant les premières années du fait de l'étroitesse des voies respiratoires supérieures et de la limitation de l'ouverture buccale. Des signes moins constants sont les enchondromes et/ou fistules prétragiens, des anomalies vertébrales et cardiaques, des fentes commissurales bilatérales.
- *Étiologie :* Le syndrome est dû à des mutations du gène TCOF1 (5q32-q33.1) codant pour la phosphoprotéine nucléolaire Treacle ou des gènes POLR1C (6p21.1) et POLR1D (13q12.2), codant pour des sous-unités des ARN polymérases I et III. La transmission est autosomique dominante avec une pénétrance de 90% et une expressivité variable, également chez des individus de la même famille.
- *Prise en charge et pronostic :* La prise en charge est pluridisciplinaire. En cas de détresse respiratoire postnatale, une trachéotomie, une ventilation non invasive (VNI) ou une distraction mandibulaire chirurgicale doivent être discutées. La chirurgie maxillo-faciale et plastique permet de corriger l'hypoplasie des tissus mous (lipostructure), l'hypoplasie osseuse (distraction chirurgicale, greffes osseuses), le colobome palpébral et la fente palatine. Le traitement de la limitation de l'ouverture buccale est très difficile. La chirurgie ORL spécialisée est requise pour les anomalies de l'oreille moyenne (chirurgie fonctionnelle)

Syndrome de Treacher-Collins – **Encyclopédie Orphanet du Handicap**
www.orpha.net/data/patho/Pub/fr/Handicap_TreacherCollins-FrFrPub293v01.pdf | 27 novembre 2013 1

Disability factsheets collection

- Description of disease
- Disability situations
- Living with a disability
- Aids to limit or prevent disabilities

Complementary information & Sources

Submission by the

NGO COMMITTEE FOR RARE DISEASES

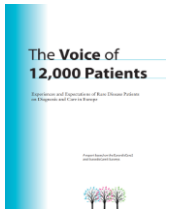
to the United Nations Special Rapporteur on the Rights of People with Disabilities for the study on "*the right of persons with disabilities to the highest attainable standard of health*" to be presented at the 73rd session of the General Assembly (October 2018).

- Access to accurate and timely diagnosis
- Proper and timely access to treatment
- Access to infraestructure
- Availability
- Marginalisation
- Examples of good practices from the rare disease community on access to health services & on engaging people with a rare disease in service design

You can [access the full contribution here]

Sources: challenges faced by people with a rare disease

EURORDIS-Rare Diseases Europe (2009). The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe. www.eurordis.org/publication/voice-12000-patients.



EURORDIS-Rare Diseases Europe (2017). Juggling care and daily life: The balancing act of the rare disease community. http://bit.ly/SurveyRD.

- Over 3000 respondents from 42 European countries; affected by 802 diseases
- Conducted through the EURORDIS survey initiative Rare Barometer Voices in the scope of the EU-funded project INNOVCare

EURORDIS-Rare Diseases Europe (2017). Access to treatment: Unequal care for European rare disease patients. http://bit.ly/SurveyAccessTreatment

- 1350 respondents from 21 European countries from 42 countries
- Conducted through the EURORDIS survey initiative Rare Barometer Voices

Rare Barometer Voices

A EURORDIS INITIATIVE

www.eurordis.org/voices

Thank you
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EURORDIS would like to thank all Rare Barometer partners for their support to co-fund the surveys conducted via EURORDIS survey initiative Rare Barometer Voices

Rare Barometer Voices

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eurordis.org/voices

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This project is co-funded by the European Union
EaSI PROGRESS, DG Employment, Social Affairs and Inclusion

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Co-funded by the Health Programme of the European Union

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