European Reference Network
for rare or low prevalence complex diseases

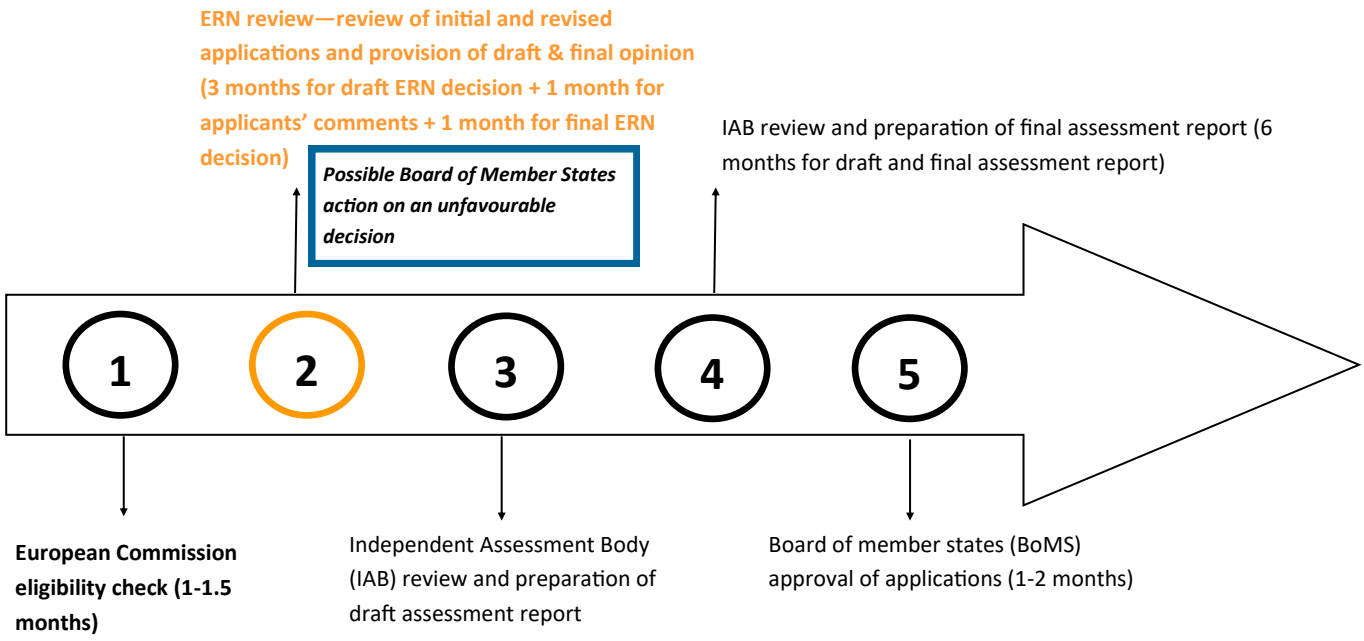
Network
Craniofacial anomalies and ear, nose and throat disorders (ERN CRANIO)

NEWSLETTER

MAY 2020
ISSUE 5: EXTENDED ISSUE

ASSESSMENT OF NEW MEMBER APPLICANTS:

WHERE ARE WE IN THE PROCESS?



The first step of the assessment process (European Commission eligibility check) is now complete. 23 applicants passed the eligibility check and are now being internally reviewed by the ERN CRANIO assessment committee. The deadline for ERN review was originally set at 30 April 2020 but it has now been postponed until 30 September 2020 in light of the COVID-19 outbreak.

ERN CRANIO PATIENT REGISTRY - AN UPDATE

In 2019, ERN CRANIO responded to a call published under the European Commission Consumers, Health, Agriculture and Food Executive Agency (CHAFEA) annual work programme (2019) and submitted a proposal to receive financial support for the development of an ERN CRANIO patient registry (200 000 EUR).

5/24 ERNs had already received funding from the health programme (annual work programme 2016) and have/are currently developing their own rare disease registry. This call was open for the remaining 19 ERNs to apply for financial support.

The ERN CRANIO proposal was submitted in October 2019, describing a plan to build a registry focused on the outcome of treatment using standardised diagnosis-specific outcome sets (starting with craniosynostosis and cleft lip/palate). It also proposed to include patient reported outcome measures and outcomes on patients' quality of life.

ERN CRANIO was **SUCCESSFUL** in obtaining this financial support. It is now the intention to start putting this proposal into practice.

WELCOMING NEW AFFILIATED PARTNERS TO ERN CRANIO!

In 2019, a call was open for member states to designate healthcare providers to become 'affiliated partners' of ERN CRANIO. There are two types of affiliated partner: ***Associated National Centres & National Coordination Hubs*** (Click here for more information).

ERN CRANIO received a total of 9 affiliated partnership applications from 8 different member states (x 7 Associated National Centres, x 2 National Coordination Hub applications). A bilateral agreement is signed by both the affiliated partner's chief executive officer and the ERN CRANIO coordinator. This defines areas for collaboration and is tailored to each affiliated partner's capacity and level of experience. An ERN CRANIO affiliated partners integration strategy has also been developed, which is currently awaiting approval from the board of member states.

Bilateral agreements have now been signed for the following centres and we are delighted to welcome them to the network:

- ***University Hospital Salzburg*** (Austria) - Associated National Centre
- ***Riga Stradins University, Institute of Stomatology, Cleft Lip and Palate Centre*** (Latvia) - Associated National Centre
- ***University Medical Centre Ljubljana*** (Slovenia) - Associated National Centre
- ***Vilnius University Hospital Santaros Klinikos*** (Lithuania) - Associated National Centre
- ***Prof. dr S Popowski Regional Specialised Children's Hospital, Olsztyn*** (Poland) - Associated National Centre

Welcome to ERN CRANIO!

The remaining bilateral agreements will be finalised and a videoconference call will be arranged with the affiliated partners to discuss *'Affiliated partnership in practice'.*

ERN CRANIO CLINICAL GUIDELINES

Development and publication of an ERN CRANIO clinical guideline on craniofacial microsomia

Craniofacial microsomia (CFM) is estimated to occur in 1:3000 to 1:5000 live births. Whilst CFM is the second most common congenital disorder of the face after cleft lip and palate, no (inter)national guideline existed to guide its clinical management. CFM is also a highly challenging disease to diagnose, treat and monitor due to its wide phenotypic spectrum.

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Over the past year, ERN CRANIO clinicians have systematically developed a European clinical guideline on CFM, which incorporates evidence-based recommendations from multiple disciplines to support the provision of optimal patient care. The guideline is founded upon real-life experiences of patients and their parents. A survey was completed by patients and their families to help identify the gaps in care provision and the difficulties and challenges they experienced. Responses guided the questions to be addressed in the clinical guideline.

The guideline is soon due to be published open-access in the ***Journal of Craniofacial Surgery*** to facilitate dissemination to healthcare professionals around the world involved in the care of patients with CFM. Other dissemination efforts will also be undertaken to ensure accessibility to patients and their families and a patient-version of the guideline will be developed.

Development of future clinical guidelines within ERN CRANIO

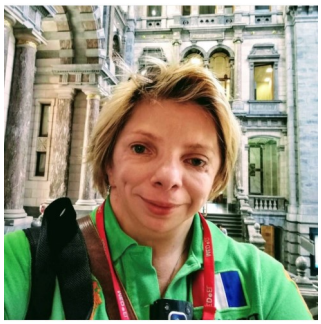
European Commission

At a meeting held in Brussels on 6 February 2020, the European Commission announced the provision of technical assistance to the ERNs for the development, appraisal and implementation of clinical guidelines and clinical decision support tools.

This assistance will be provided by a consortium led by Fundacion Progreso y Salud, who obtained the 4 – year contract following a European Commission tender.

The consortium proposes to support ERNs to adopt a common methodology and deliver training activities. They also propose to support each ERN with the production of two new clinical practice guidelines and review of five clinical decision support tools. Support provided by the consortium will be ERN-led and adapted to the diversity of the networks.

WELCOMING IVANA! ERN CRANIO patient representative for Treacher Collins Syndrome



We would like to introduce you to Ivana Marinac, ERN CRANIO's new patient representative for Treacher Collins Syndrome (and associated conductive hearing loss). Ivana is from Zagreb, Croatia and is an otorhinolaryngologist by profession. Within ERN CRANIO her role will focus on representing the voice of patients with Treacher Collins Syndrome, acting as a bridge between the patient community and professionals. Ivana has Treacher Collins Syndrome herself so has first hand experience of living with the rare disease.

"Being part of both worlds gives me the opportunity to understand efforts and taken direction by the professionals but also patients´ fears, hopes and experiences"

Ivana is part of both The Croatian Association for Rare Disease and the NGO Cochlea Implant Croatia.

We asked Ivana, "what does it mean to you to be involved within ERN CRANIO as a patient representative?"

"Medicine is a science of uncertainty and an art of probability" – said William Osler. These words beautifully describe a complexity of patient – medical professional relationship. I was once called a prodigy on the crossroad of few medical conditions (and challenges) by a friend, so I have asked myself how I can serve the best for both communities I belong to - how to take best from both worlds. I have had numerous experiences as a patient where I felt that professionals did not understand me clearly so I do understand other patients, especially those in my shoes, but also colleagues professionals when a treatment period does not take a rectilinear path and a situation become mutual misunderstanding and a situation of possible dispute by not having a clear path of handling their conditions, a clear trajectory line of choosing what is important or achievable for them. Today, not having an urgent condition for medical handling gives the part of patients with craniofacial anomalies a big opportunity of choosing a medical procedure which tackles their specific pursuit according to the best evidence-based medicine protocols. From the patient perspective, I would underline and bold the imbalance of attainable goal as opposed to existing but inaccessible medical procedures for certain, mostly small or newer, EU countries. Individual national healthcare legislation complexes a problem even more by having specifically incomparable peculiarities in order that certain medical procedure is not accessible to all EU citizens. Having in mind all I wanted to sum up, there is a serious work ahead of us - to have a fruitful discussion by having a thorough debate of both sides".

THE EUROPEAN CONFERENCE ON RARE DISEASES (ECRD 2020)

As a result of the COVID-19 pandemic, the 10th European Conference on Rare Diseases & Orphan Products (ECRD 2020) due to take place in Stockholm will now take place **ONLINE** on 14 - 15 May 2020. Click here to find out more and follow the LinkedIn page! Registration remains open until 10th May (for payments via credit card).

We are really happy to announce that a poster on the ERN CRANIO patient journeys will be presented at this online event titled:

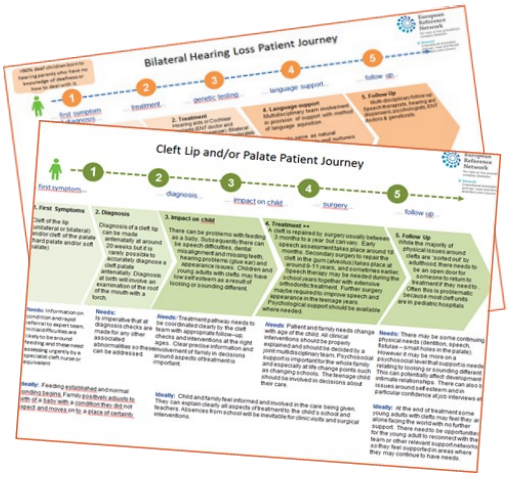
'Patient Journeys: Personal experiences shaping clinical priorities'

Patient Journeys are visual documents which map disease-specific patient/ family 'needs' and 'ideal support scenarios' at key clinical stages. Patient Journeys have been developed / are in development for:

Syndromic Craniosynostosis, Bilateral Hearing Loss, Pierre Robin Sequence, Craniofacial Microsomia, Cleft Lip/Palate and Treacher Collins Syndrome.

Patient journeys can be used to shape the network's clinical priorities at disease-level. Conducting an assessment of the common needs/ideals may also help to shape clinical priorities on a broader level.

The plan to conduct an assessment of common needs is set out on this poster. It also notes the plan to integrate or present the patient journeys (based on real-life experiences) alongside expert recommendations.



We look forward to sharing details of this project and next steps with other members of the rare disease community!

Sending you all our well wishes at this challenging time.