European
Reference
Network
for rare or low prevalence
complex diseases

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

NEWS

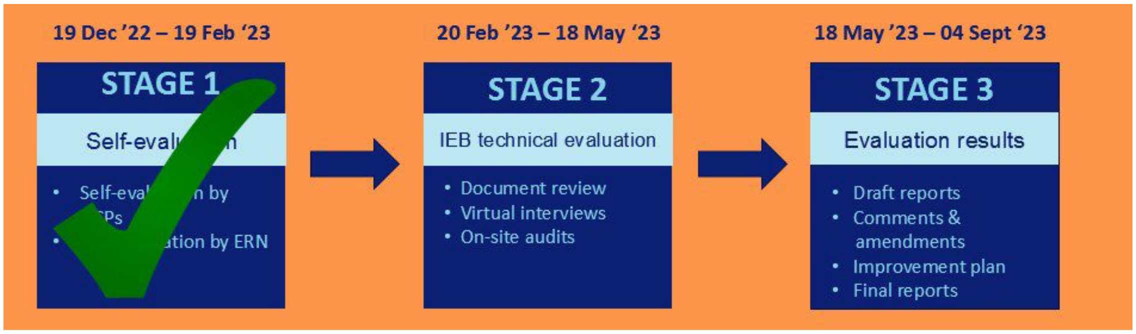
ERN CRANIO Newsletter

March 2023, Issue 14

5 Yr evaluation of the ERNs (Amequis)

The evaluation process for both ERN CRANIO and our Full Members is currently underway as part of the AMEQUIS framework. The aim of this framework is to continually assess, monitor, evaluate, and improve the performance of ERNs to ensure that ERNs remain responsive to the needs of the rare disease patient community, deliver on their objectives, and improve their performance over time.

In stage 1 of the evaluation process, which has recently been completed, all healthcare providers from ERN CRANIO, who have been part of the network since 2017, have filled out their self-evaluation form on the portal, and the ERN self-evaluation form has been submitted.



19 Dec ’22 – 19 Feb ‘23
STAGE 1
Self-eval n
- Self-eval n by Ps
- ation by ERN

20 Feb ’23 – 18 May ‘23
STAGE 2
IEB technical evaluation
- Document review
- Virtual interviews
- On-site audits

18 May ’23 – 04 Sept ‘23
STAGE 3
Evaluation results
- Draft reports
- Comments & amendments
- Improvement plan
- Final reports

Currently, we are in the midst of stage 2, where five ERN CRANIO centers have been selected to undergo an onsite audit. The evaluation body will conduct several virtual

After this period, from May 18th, 2023, to September 4th, 2023, an evaluation report will be provided.

If you require further information about the evaluation or have any questions, please contact the project manager, Ikram L'Khssim, via email at **i.lkhssim@erasmusmc.nl**

Launch of new ERN CRANIO videos!

Over the last months, several working groups with members from different ERN CRANIO member hospitals have joined efforts to create informative videos. The target audience for these videos are healthcare professionals outside of the ERN CRANIO network. The aim is to inform them on different rare diseases, how to recognize them and how to diagnose them.

ENT – difficult airway video series

A total of 4 videos were created by the working group, which has an overall topic of 'difficult airway management'. A special thanks goes out to:·

- Bas Pullens, otorhinolaryngologist & ENT workstream lead, Erasmus MC
- Lonneke Staals, anesthesiologist, Erasmus MC
- Briac Thierry, otorhinolaryngologist, Hôpital Universitaire Necker Enfants-Malades
- Nicolas Le Boulanger, otorhinolaryngologist, Hôpital Universitaire Necker Enfants-Malades
- Romain Luscan, otorhinolaryngologist, Hôpital Universitaire Necker Enfants-Malades

The video series consists of a 3D video on nasal intubation, fiberoptic intubation, intubation through the laryngeal mask and difficult airway management. Click on the link below to visit our YouTube channel and watch the video series!

Orodental – amelogenesis imperfecta video series

A total of 4 videos were created by the working group, which has an overall topic of ‘amelogenesis imperfecta’ in orodental anomalies. A special thanks goes out to:

- Marie-Cécile Manière - pediatric dentist, Hopitaux universitaires de Strasbourg
- Agnès Bloch-Zupan - oral biologist, Hopitaux universitaires de Strasbourg
- Isaac Maximiliano Bugueno Valdebenito - dentist, Hopitaux universitaires de Strasbourg
- Olivier Etienne - dentist, Hopitaux universitaires de Strasbourg
- Serena Lopez - dentist, Hopitaux universitaires de Strasbourg
- Willem Fennis - dentist, UMC Utrecht
- Jamilla Ross - dentist, UMC Utrecht

This video consists of different topics when considering amelogenesis imperfecta, including the diagnosis, differential diagnosis, treatment & management and genetics of this rare orodental disease. Click on the link below to visit our YouTube channel and watch the video series!

Craniosynostosis – recognizing craniosynostosis video series

A total of 2 videos were created for this video series, both focussing on how to recognize craniosynostosis. The first video focusses on informing healthcare professionals (such as general practitioners) on how to recognize babies with a craniosynostosis. A special thanks goes out to:

- Ulrich-Wilhelm Thomale - plastic surgeon, Charité Hospital
- Dylan Murray – plastic surgeon, Children's Health Centre Dublin
- Noele O'Mahoney – project manager, Children's Health Centre Dublin
- Mieke Pleumeekers – plastic surgeon, Erasmus MC

The second video of this video series was developed to inform midwife and sonographers on the prenatal detection of craniosynostosis in unborn babies. A special thanks goes out to Nina Peters (fetal medicine & gynaecologist, Erasmus MC) for her effort on making this video!

Educational session on Radiology Berlin annual meeting

The educational session on radiology from the annual meeting in 2022 has been uploaded to our YouTube channel. Please click on the link below if you are interested in the radiology session from the annual meeting in Berlin, different topics are included such as MRI and CT evaluations on craniosynostosis, black-bone sequencing and surgical and clinical aspects.

ERN CRANIO meetings

ERN CRANIO cleft speech meeting

On the 27th of January, the ERN CRANIO cleft speech meeting was held at the university medical center Utrecht. Over 30 speech and language experts from all over Europe (!) gathered to discuss the speech outcome measures to be used in the ERN CRANIO cleft lip and palate registry. With many inspiring presentations and plenary discussions, the day proved fruitful, and a consensus was reached on what outcome measures are to be included in the registry!

We want to thank all participants for joining in our collective effort in improving care in Europe and we look forward to the next cleft speech meeting.

ZeldSamen symposium

On February 16th ERN CRANIO attended the ZeldSamen symposium. A very interesting event where parents and professionals were able to have discussions about the needs of parents of children with a rare genetic condition, improving care for these children/adults, and involving patients and their parents in research. All in all, it was a very successful day!

February 2023

On the 24th and 25th of February, the very first all-ePAG in-person meeting was hosted in Rotterdam, the Netherlands. 7 of the 8 patient representatives of ERN CRANIO were present for the strategy meeting focused on the future of our European Patient Advocacy Group (ePAGs). Our ePAGs consists of the following patient representatives (names from left to right & bottom):

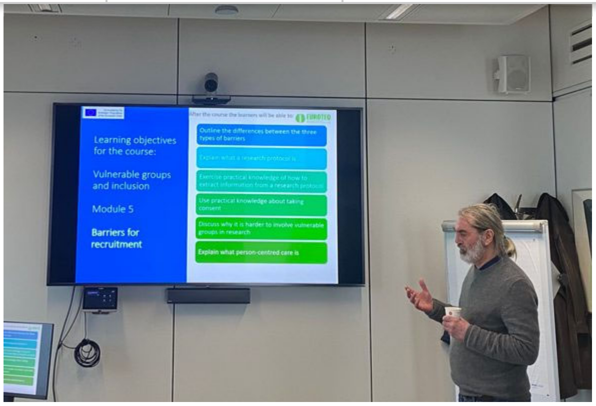
- Anne-Sophie Mercey-Jarosz, representing Association mille et une têtes
- Gareth Davies, representing European Cleft Organisation
- Mariët Faasse, representing LAPOSA
- Karen Wilkinson-Bell, representing Headlines UK
- Sandra Mösche, representing Elterninitiative Apert-Syndrome
- Ivana Marinac, representing Rare Diseases Croatia
- Philippe Pakter, representing Pierre Robin Europe
- Michel Francois, representing ALPC

It was meaningful to get this group together and think about the future. Interested in the patient organizations they represent or getting into touch with our ePAGs for a project? Please contact project manager Jana Steerneman (j.steerneman@erasmusmc.nl) for more information!

ePAG + NEUcrest meeting!

On the 24th of February, Mariët Faasse, Gareth Davies, and Karen Wilkinson-Bell hosted a workshop for NEUcrest PhD candidates!

The NEUcrest project is another European initiative on neural crest cells, for more information please click here. The workshop was focused on patient-friendly writing for researchers and how to engage and involve patients in research. A full day was spent in the workshop and closed with informal drinks afterwards to strengthen the connection between our two European projects (ERN + NEUcrest).



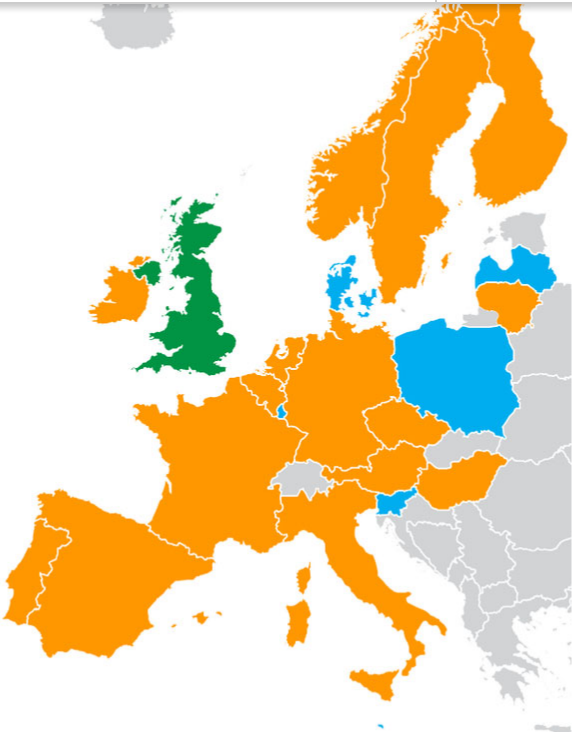
experience of living with Treacher Collins syndrome.

ERN CRANIO updates

Supporting partners

We are excited to announce that in addition to our ERN Full Members and Affiliated Partners, ERN CRANIO has expanded to include Supporting Partners! These Supporting Partners are healthcare providers, medical societies, and other individuals or entities who contribute to the work of the network in various ways. It's important to note that Supporting Partners do not have a commercial relationship with ERNs or their Full Members, Affiliated Partners, or the European Commission.

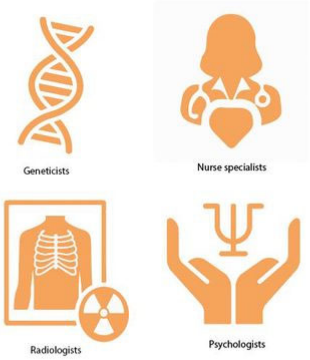
We are grateful to welcome our new ERN CRANIO Supporting Partners, including Great Ormond Street Hospital Children, Oxford Craniofacial Unit, and Alder Hey Children's Hospital.



FaceMe update

During the annual meeting of ERN CRANIO (November 2022 in Berlin) we officially launched the project 'Face Me'. During the journey of this project, we will shed light on the mutual understanding between surgeons and patients in an innovative and artistic way, by working with different European artists.

Irene Mathijssen: "I think that as doctors and caretakers we can be even better guides for our patients when we see every aspect of their lives, not just the physical part. And show more of



Cross-workstream working group update

At the end of 2022, a call was published for geneticists, psychologists, nurse specialists, and radiologists to join different cross-workstream working groups.

The nurse-specialist working group is currently led by Elin Weissbach (nurse specialist, Erasmus MC) and has officially kicked-off in February.

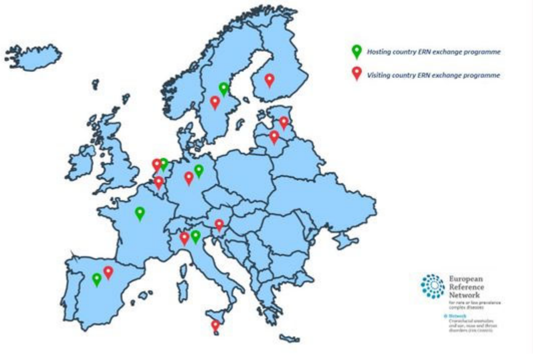
The launch of Face Me in Berlin was a great success and we would like to thank everyone who offered to participate in this project. As of this moment, the involved artists in this project are in the artistic process of gathering all information from the ERN CRANIO network.

This projects is a collaboration between ERN CRANIO, True Blue - Creative Healthcare Communications, Art Partner, and Sylvie Zijlmans & Hewald Jongenelis.

The co-leads of this working group are Marizela Kljajic (psychologist, Sahlgrenska University Hospital) and Jolanda Okkerse (psychologist, Erasmus MC).

The geneticsts working group was officially kick-off on the 6th of March. The co-leads for this working group are Wanda Lattanzi (clinical geneticist, Fondazione Policlinico Universitario A. Gemelli) and Alexandra Topa (clinical geneticist, Uppsala University Hospital.

Are you interested in joining one of these working groups? Please contact Jana Steerneman (j.steerneman@erasmusmc.nl)



Clinical exchange programme

We are pleased to announce that the new clinical exchange programme has been launched as of February 2023, following the remarkable success of our last programme. Interested in participating in this programme?

Contact the coordination team for more information via:
ern-cranio@erasmusmc.nl

Collecting annual monitoring data

The process of collecting the annual monitoring data has commenced, and a majority of the ERN CRANIO centers have already submitted their data. For those who have not yet done so, please make sure to still send your center's data a.s.a.p. to i.lkhssim@erasmusmc.nl

Call for cross-workstream working group

We are planning to launch a new cross-workstream workgroup for OR assistants in the near future. The primary objective of this workgroup is to combine expertise and knowledge in the respective workstreams, work collaboratively, and hold regular meetings to enhance knowledge exchange across Europe.

Would you like to join or know someone within your team who would be interested in participating in this cross-workstream working group? Please get in touch with the project manager, Ikram L'Khssim, by emailing **i.lkhssim@erasmusmc.nl**.

ERN-wide News

EJP RD

EJP RD - ERN

WORKSHOP

GENETICS AND PRECISION MEDICINE IN RARE DISEASES

Organizer: Carolina Neves, Associação Protectora dos Diabéticos de Portugal (APDP)

18-19 MAY 2023
Lisbon, Portugal

ERN WORKSHOPS
THE EJP RD "ERN RESEARCH TRAINING WORKSHOPS CALL"

EUROPEAN JOINT PROGRAMME RARE DISEASES

European Reference Network

The European Joint Programme on Rare Diseases is an initiative that has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement N°825575

EJP RD ERN Workshop

📢Notice to medical doctors, geneticists & molecular researchers: a new #EJPRD - ERN workshop intended for you is being organised!

EURORDIS Membership Meeting

EURORDIS is offering patient fellowships for up to 40 patient advocates to attend the EURORDIS Membership Meeting 2023 Stockholm taking place 25-27 May in Stockholm, Sweden. These fellowships aim at empowering patient advocates by offering a platform for networking opportunities, access to information and sharing experiences.

The programme covers:

- Registration (fee waivers)
- Travel (return trip economy fare flight or train)

🚨Registration deadline: 23 March 2023

📅Date of the event: 18-19 May 2023

📍 Location: Lisbon, Portugal (event in English)

Learn More

Fellows will be selected based on their advocacy skills and objectives, by an ad hoc committee and notified by 20th March 2023.

Application deadline: 15 March 2023

Learn More

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