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

Issue 18 - March 2024

ERN CRANIO NEWS

Scientific Committee Update

The ERN CRANIO Scientific Committee is thrilled to announce the addition of four distinguished experts joining our scientific committee, each bringing invaluable perspectives and expertise to our discussions.

Dawid Larysz works with craniofacial anomalies for the past 20 years. He holds an M.D. from the Medical University of Silesia, a Ph.D. in glioma gene expression, and an M.Sc. in clinical psychology. He's a consultant neurosurgeon and professor, with diverse experience spanning pediatric neurosurgery, radiation oncology, and craniofacial surgery. He is a president of Polish Craniofacial Society and is the head of the only Excellence Center for Craniofacial Surgery in Poland.

Kristin Billaud Feragen is a psychologist from the Oslo Cleft Lip and Palate team, with more than 70 publications.
Her doctoral and post-doctoral research focuses on investigating appearance satisfaction and psychological well-being in individuals with cleft lip and palate conditions.
Since 2015 Kristin holds a position of Research Coordinator at the Centre for Rare Disorders of the Oslo University Hospital, where she oversees a diverse portfolio of research projects.

Professor of Experimental Biology at the Catholic University in Rome, specializing in medical genetics. Her research focuses on mesenchymal stromal cells for regenerative medicine and craniofacial anomalies like craniosynostosis. With over 70 publications and funding from institutions like the NIH and European Commission, she leads a research group and teaches at the Catholic University.

University Hospital specializing in hearing loss. With over 50 research papers and involvement in national and international projects, Davide brings the ENT perspective to the Scientific Committee. He is a work group leader for the Radiology cross-workstream group. His work includes educating on rare craniofacial disorders and dysphagia in craniofacial patients.

Yearly gathering of monitoring data

The yearly monitoring exercise has come to an end in March. We want to thank everyone for contributing to the monitoring exercise, we are very happy to say that all of our members and affiliated partners have completed the exercise!

MONOBLOC WORKSHOP - NECKER 2024

Organized by the French Reference Center for Craniosynostoses and Craniofacial Malformations (CRANIOST), in collaboration with an international expert committee, this workshop will allow experienced craniofacial surgeons to learn how to manage faciocraniosynostoses by frontofacial monobloc advancement and posterior distraction.

Lectures

Treatment plan for faciocraniosynostoses

Posterior expansion : surgical technique

Frontofacial monobloc advancement: surgical technique

Post-operative management and complications

Results and research perspectives

Hands on training

Training on realistic soft- and hard-tissue 3D-printed models, 1 model for 2 participants

1 of the following senior supervisor per team :

- **Pr Nivaldo Alonso** (São Paulo, Brazil)
- **Dr Eric Arnaud** (Paris, France)
- **Dr Syril James** (Paris, France)
- **Pr Roman-Hossein Khonsari** (Paris, France)
- **Pr Boris Laure** (Tours, France)
- **Pr Irene Mathijssen** (Rotterdam, Netherlands)
- **Dr Giovanna Paternoster** (Paris, France)
- **Dr Roberto Requena** (Santiago, Chile)
- **Dr Yoshiaki Sakamoto** (Tokyo, Japan)

Face Me update

The next phase of the Face Me project has started from Dublin onwards. **We are very happy to announce that the next host for the Face Me project are Hopital Necker - Enfants Malades and patient organisation Les P'tits Courageux!** The project will go through a new phase of innovation and brainstorm, to create something different from the 'pilot' phase. The project still focuses on the patient-healthcare provider relationship and what it means for healthcare providers in different countries to become a better guide for their patients.

Moreover, we are currently creating a **toolkit to perform workshops in your hospital or patient organisation**. The workshops focus creating a dialogue between healthcare providers and patients or parents, to understand what the other needs and what the other needs to trust. Are you interested in performing a workshop in your hospital or patient organisation? Click on the button below to send us an e-mail!

Stay tuned via our social media channels and website page!

Contact us!

ERN CRANIO - ORPHANET update

ORPHA_1520 Craniofrontonasal Dysplasia has been updated by our ERN CRANIO coordinator Irene Mathijssen and clinical geneticist Prof. dr. Andrew Wilkie from Oxford University Hospital.

Did you know that the ORPHANET website is a repository of information on all rare diseases across the world? In the monitoring exercise of this year, the European Commission asked all member centres if they use ORPHANET codes in their hospital, to make an inventory of the readiness of Europe to implement the orphacodes in the hospitals. In the coming years, we will focus on updating the information of the ERN CRANIO orphacodes on the ORPHANET website. Please contact us if you are willing to update this information or click on the button below to access the ORPHANET website.

CPMS 2.0 update

CPMS 2.0 is currently in development, with ongoing efforts to enhance its features. During this period, we encourage users to continue utilizing the existing CPMS platform. Should any assistance be required, feel free to reach out to the ERN CRANIO coordination team. The transfer of all ERNs to the new CPMS 2.0 platform will start in April and should be finished **before September 2024**. A webinar and instructions for this new platform will follow in the coming months.

For comprehensive guidance on using the current CPMS, the ERN CRANIO team has created manuals and instructions for members. You can access information on logging into CPMS and instructions for uploading a panel on the ERN CRANIO website at CPMS | ERN CRANIO website (ern-cranio.eu).

Dublin educational session webinars now online!

The recordings from the educational session in Dublin on 17 November this year are now available to watch online! Head to our YouTube page by clicking the button below!

ERN CRANIO YouTube channel

ERN CRANIO WORKGROUPS UPDATE

Facial Nerve Palsy workgroup

A new disease-specific working group is being created for facial nerve palsy. We are searching for clinicians that treat patients with this disease, preferably plastic surgeons or maxillofacial surgeons, but others are welcome as well. Within this group we consider the following diseases: Congenital facial nerve palsy / Isolated hereditary congenital facial paralysis, Moebius Syndrome. We are contacting only the centres within our network with official acknowledgement for this disease within our ERN. We would greatly appreciate your centre’s participation in this group as well. Some of the proposed projects for this group will be: 1) development of an ERNCRANIO registry dataset for facial nerve palsy, based on ICHOM, and 2) consensus study on the classification methodology for facial nerve palsy, 3) research. Contact one of our project managers: j.steerneman@erasmusmc.nl

Prenatal screening workgroup

Calling specialists in prenatal screening! Be part of our upcoming ERN CRANIO prenatal screening group, focusing on detection of ERN CRANIO rare diseases prenatally. Join us in sharing knowledge, collaborating on research, developing standards of care, and participating in regular meetings to enhance knowledge exchange across Europe. Do you have an expert in your hospital or are you an expert in prenatal screening? Contact one of our project managers: j.steerneman@erasmusmc.nl

collaborative platform aimed at addressing sleep disturbed breathing, and training sessions and case discussions within our ERN, placing a particular emphasis on sleep measurements, as requested by our members.
We're happy to let you know that we are launching a short course dedicated to exploring sleep disturbed breathing. Scheduled for May (exact date will follow), this course will comprise three informative sessions, complemented by in-depth discussions centered around challenging cases. If you possess any cases that warrant discussion during our upcoming sessions, we welcome you to share them with us for inclusion in the upcoming course.
Should you be enthusiastic about participating in and following the course, or if you're interested in joining our sleep disturbed breathing group, please don't hesitate to reach out to us via email at: i.bos@erasmusmc.nl.

OR Nurses Cross-ERN working-group

The OR Nurses group has recently expanded to include OR nurses from ERNICA and eUROGEN. The inaugural Cross-ERN OR Nurses group meeting was held at the end of January.
If you're an OR nurse interested in joining this group, please express your interest by sending an email to: i.bos@erasmusmc.nl.

Anesthesiologists group

The first Anesthesiologists working group meeting will take place on the 21st of March at 16:00. If you're eager to join the group but haven't yet registered, please don't hesitate to reach out to us via email at: i.bos@erasmusmc.nl.

ERN CRANIO REGISTRY UPDATE

The ERN CRANIO team, together with Molgenis, has been diligently finalizing the remaining details of the registry in preparation for data upload. We're excited to share that in February, we successfully conducted a pilot and integrated real data into the registry.

At present, our focus lies in completing a comprehensive manual and developing instructional videos to facilitate the launch of the onboarding process, set to

On February 29th, we’ve hosted a webinar in collaboration with the ERDRI team, aimed at informing our center representatives about the SPIDER pseudonymization tool and the necessary preparatory steps required for its implementation.
In April, we'll be conducting a second webinar, in conjunction with the ERDI team, specifically tailored for data managers from HCPs that have already signed the data sharing agreement with us. This session will provide detailed guidance on utilizing the SPIDER tool for data pseudonymization.

For all HCPs that have entered into a data sharing agreement with us, we are currently scheduling onboarding meetings, starting in April 2024.
If your institution has signed a data sharing agreement with ERN CRANIO, kindly provide us with the contact details of the designated data managers and representatives who will be granted access to your data, by sending an email to i.bos@erasmusmc.nl.

ERN CRANIO RESEARCH UPDATE

Journal of craniofacial surgery opportunity!

There is a special opportunity for ERN CRANIO members to submit their articles to the **Journal of craniofacial Surgery for a special supplement for the 40th year anniversary.** The editor of the journal has given our ERN the special possibility for our state of the art research and network. The deadline for submission of the articles will be October 2024.

There will be limited space in the journal, only 12 issues will be published in the supplement. Therefore, we would like to make an inventory of the research in the network and which align best with the Journal’s vision. Please click on the button below to fill out our survey to apply for publishing your research in this journal.

Apply for publishing your research here!

JOURNAL OF CRANIOFACIAL SURGERY

A European multicenter outcome study on the different perioperative airway management policies following midface surgery in syndromic craniosynostosis: a proposal for a Standard Operating Procedure.

Perioperative airway management following midface advancement in children with Apert and Crouzon/Pfeiffer syndromes can be challenging, and protocols often vary between hospitals. Little is known about the safety of immediate extubation and potential respiratory complications following midface advancements. Therefore, through this ERN collaboration, our aim was to investigate perioperative airway management in midface advancements and assess postoperative respiratory complications. Ultimately, seven ERN centres participated in this study, and we were able toenrol 275 patients. The study revealed that immediate extubation following midface surgery is a safe alternative to delayed extubation in most cases, as it is associated with a minimal need for additional airway support and few complications.

Read the full article here

Syngnathia: identifying genetic causes & timely and appropriate surgical treatment

Syngnathia is a very rare craniofacial malformation with challenges in phenotypic and genotypic diagnosis and the surgical/non-surgical management. Recently, the paper below from our Paris expertise center sheds new light on the genetic background of this condition.

A research group has been established, which aims to collect more syngnathia cases from our ERN-CRANIO network. By grouping together these rare cases we will better understand the genetics of this condition. Second, the timing and choice of surgical techniques for correcting syngnathia remains controversial and sharing expertise would be of value to identify the optimal treatment.

Do you have any syngnathia cases in your hospital? Please contact our project manager to join this research group: j.steerneman@erasmusmc.nl

Read the abovementioned article here

RARE DISEASE NEWS

Rare Disease Day - 29 February 2024

RAREDISEASEDAY.ORG

Rare Disease Day is the global awareness-raising campaign for the 300 million people living with a rare disease worldwide and their families, taking place on the last day of February each year. The campaign has the goal of increasing equity for people living with a rare disease, improving their lives with equitable access to diagnosis, treatment, care and social opportunity. Rare Disease Day is open to everyone to get involved and seeks to build an engaged grassroots community of patients and families, whilst raising awareness amongst healthcare professionals, policy makers, institutions and the general public.

JARDIN

Joint Action on Integration of ERNs into National Healthcare Systems - JARDIN

6 - 8 March 2024, Brussels Belgium

More than 6,000 rare diseases (RD) are known to date. They are defined as diseases affecting no more than 5 in 10,000 people in Europe. Expertise and resources for these conditions are scarce. It is estimated that about 30 million people are living with a RD in Europe, making effective networking and cooperation measures for diagnosis and treatment essential.

In 2017, the EU therefore established the European Reference Networks (ERNs) which are a voluntary, coordinated cooperation between all member states in the field of highly specialised health services. These networks will generate immense added value for European citizens living with a RD or complex conditions. There are currently 24 networks connecting more than 1,600 clinical centres across the Union.

To improve the accessibility of the ERNs for patients in member states, the EU is now funding a pioneering 3-year project involving all member states plus Norway and Ukraine, the Joint Action on Integration of ERNs into National Healthcare Systems ('JARDIN'). JARDIN will be coordinated by Professor Till Voigtländer from the Medical University of Vienna in Austria and will produce recommendations, and implementation pilots in the main fields of action, such as patient pathways, national reference networks, and data management for rare diseases. These pilots constitute a promising way to provide EU member states with directly implementable solutions for their healthcare systems.

JARDIN's kick-off meeting took place in Brussels on the 6th to 8th March 2024. It marked the beginning of a collaborative effort to shape the future of the ERNs.

European Conference on Rare Diseases

15 & 16 May 2024, Brussels Belgium

ACTION WITHIN REACH

PIONEERING SOLUTIONS FOR RARE DISEASES

HYBRID 15 & 16 MAY 2024

12th European Conference
on Rare Diseases and Orphan Products

Join us for ECRD 2024, the largest patient-led conference on rare diseases in Europe. This event plays a key role in shaping policies for individuals with rare conditions, focusing on practical solutions under the theme "Action Within Reach." ECRD provides a valuable opportunity to stay informed and contribute actively, particularly in areas like EU specialized services and cross-border healthcare access. The conference covers topics relevant to ERNs, making it a significant gathering for stakeholders including patient advocates, academics, healthcare professionals, industry representatives, and policymakers, both in-person and online. Organized by EURORDIS-Rare Diseases Europe, ECRD 2024 aims to foster collaboration and consensus on comprehensive rare disease policies, influencing the next EU legislative cycle.

European Cleft Palate Craniofacial Association (ECPCA) Congress

25 - 28 June 2024, Milan Italy

Dear Friends,

Milan warmly welcomes you to the ECPCA 2024 – The European Cleft Palate Craniofacial Association Congress founded in Gothenburg in 2015, from 24 to 28 June 2024.

Our conference venue, a stunningly beautiful 19th century palace, is located right in the city center. Milan is a beautiful city of contrasts, offering you fashion, shopping, and design, mingled with history and art, from the Duomo to the Castle, from the Scala Opera house to the masterpieces of Leonardo da Vinci. By train or car, in less than

Going back to science, such a stimulating setting is absolutely ideal for even the most heated scientific discussions during the day, which may easily be settled in the evening in front of a glass of relaxing wine and divine Italian food!!
We are looking forward to meeting you in Milan!

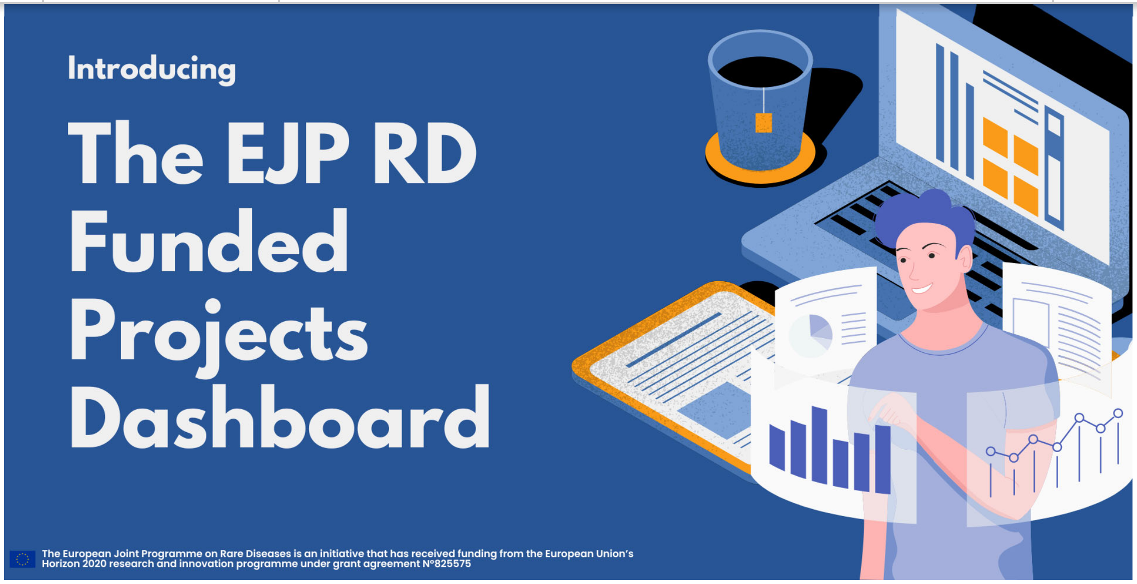
Maria Costanza Meazzini
ECPCA President

ERN CRANIO 9th annual meeting

6 - 8 November 2024, Gdansk Poland

The ERN CRANIO’s 9th annual meeting will take place on the 6,7, and 8th of November 2024 in Gdansk, Poland. Please reserve these dates in your calendars. The main topic of the meeting will be on **difficult airways in craniofacial anomalies and ENT disorders**. A call for ideas and speakers for the educational session will follow later in the year.

EJP RD Dashboard launched!



To mark Rare Disease Day, The European Joint Programme on Rare Diseases (EJP RD) the EJP RD FundedProject Dashboard! Dive into a comprehensive view of all funded projects (Joint Transnational Calls and Networking Support Scheme calls. Uncover essential project details, budgets, abstracts, and other fascinating figures. Explore the Dashboard by clicking on the button below!: https://funded-projects.ejprarediseases.org/

EJP RD Dashboard

Funded by
the European Union