



European Reference Network

for rare or low prevalence complex diseases

Network

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

NEWSLETTER

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ERN CRANIO RARE DISEASE REGISTRY PROPOSAL - SUBMITTED!



In May 2019 the Consumers, Health, Agriculture and Food Executive Agency (CHAFEA) published the annual work programme 2019 for grants under the third programme for the union's action in the field of health (2014-2020). This includes an ERN call for rare disease registries.

5/24 ERNs already received funding from the health programme (annual work programme 2016) and have/are currently developing their own rare disease registry. This call published under the annual work programme 2019 was open for the remaining 19 ERNs, such as ERN CRANIO, to apply for financial support (200 000 EUR per ERN).

We propose to build a registry focused on the outcome of treatment using standardised diagnosis-specific outcome sets (starting with craniosynostosis and cleft lip/palate). We propose to include patient reported outcome measures and outcomes on patients' quality of life.

Now we await the results of this application...

EXPANSION OF THE ERN CRANIO NETWORK

The second call for new healthcare centres to join the ERNs as full members was launched on 30th September 2019. It will remain open until 30 November 2019. Applications are to be completed online using a new application tool. All information on how to apply can be found via this dedicated webpage:

https://ec.europa.eu/health/ern/consultations/2019_call_membership_en

This launch marked the deadline for **affiliated partner** applications.

Affiliated partners are not members of an ERN. Affiliated partners are expected to provide a link to the ERN for those European countries without a healthcare provider involved.

Affiliated partners will have the opportunity to use the Clinical Patient Management System. We can also offer our support in regards to strengthening and developing their clinical expertise. Affiliated partners help to increase the profile of ERNs across Europe and provide more ERN entry points for patients. We can also benefit from their knowledge and gain an insight into the different healthcare systems and infrastructures across Europe.



There are currently 29 European hospitals involved in ERN CRANIO as full members, from 11 member states. In total, we received 9 affiliated partner applications from 8 additional member states. These applications are currently being processed.

ERNs seek to support the provision of high quality care to patients from across the European Union, regardless of where they are located. Affiliated partners can help us on this mission!

INTRODUCING MICHEL FRANCOIS! ERN CRANIO patient representative for genetic hearing loss

Michel is a member of ALPC (<https://alpc.asso.fr/>), a French association that brings together individuals with deafness, their families and professionals on the issue of French language acquisition.

Can you tell us a bit about yourself and your experiences with genetic hearing loss?

Well... I am the father of two deaf young adults. Twenty-two years ago, hearing loss was not easy to diagnose in young children and late diagnosis was common. In a few months' time, we, parents, experienced the shock of discovering both our children were deaf (our son was 4 and our daughter 1 year old).

Suddenly, we had to overload our weekly professional obligations with a series of appointments with a speech therapist, a hearing aid dispenser, a psychomotor therapist, a psychologist, and an ENT doctor. In that period of turmoil and exhaustion, we needed to restore our confidence in our parental skills. The professionals we met, helped us, but also encouraged us to meet other parents.

We had a lot of questions in mind: "Will our children be able to speak?" usually, the first question that occurs to most hearing parents (90 per cent of deaf children are born to hearing parents). But speech is just like the tip of the iceberg. We rapidly became aware of the complexity of language acquisition and the impact of deafness. We did not want our children to be stuck at a basic level of communication only dealing with daily life topics. We wanted them to have access to symbolic thinking, abstraction, literacy through full mastery of a language and we wanted to remain the main language providers for them, as any other parents.

How did you find out about the network?

To fulfil our educational goals, we joined the ALPC association and rapidly learnt French Cued Speech (LfPC): a gestural technique that makes our home language totally accessible to our deaf children ("complete spoken language through vision"). As most of hearing loss cases have a genetic origin, ALPC association has developed a partnership with the **Reference Centre for Genetic Deafness** (Necker Hospital, Paris, France), created in 2000 by doctor Sandrine Marlin. The **RCGD** is affiliated to ERN CRANIO.

What are your main motivations for wanting to be involved in ERN CRANIO?

"Share. Care. Cure" is the European Reference Network motto. As a parent of deaf children and member of the ALPC association, I think I can contribute to the concept of sharing: sharing experiences, finding common points for better care. For instance, universal neonatal hearing screening has now been implemented in most European countries implying fewer late diagnoses, but I think the shock felt by parents at the time of diagnosis and strategies of language acquisition are still current issues.

We look forward to introducing Michel to the ENT working group and to our upcoming annual meeting in Rome.

ANNUAL MEETING 2019, ROME! 29-30 NOVEMBER

Our 2019 annual meeting will take place in Rome this November.

We have a full programme planned and look forward to welcoming those of you who have registered!

The meeting will feature an educational session on difficult airway in craniofacial disorders. There will also be the opportunity to meet in smaller diagnostic-specific groups.



OTHER RELEVANT MEETINGS AND EVENTS

NOVEMBER

13th: ERN workshop for research infrastructures, Brussels

14th-15th: ERN coordinators meeting, Brussels

29th-30th: 2019 ERN CRANIO annual meeting, Rome

ERN CRANIO healthcare professionals and patient representatives: Don't forget to share any ERN CRANIO-related dissemination activities and events with us!