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

July 2023, Issue 16

ERN CRANIO 8th annual meeting

16 & 17 November 2023

Save the Date for the 8th ERN CRANIO Annual Meeting!

We are excited to announce that the 8th ERN CRANIO annual meeting has been scheduled to take place in Dublin, Ireland, from Thursday, November 16th, to Friday, November 17th, 2023. We are eagerly looking forward to welcoming you to Dublin for this event.

More detailed information regarding the meeting will be provided at a later date. However, we kindly request you to mark this event in your agenda and save the date.

Registry pilot- cleft lip/palate and/or craniosynostosis

Friendly Reminder: Submit Your Data for the ERN CRANIO Registry Pilot Project

Don't miss out on the opportunity to participate in the pilot project for the ERN CRANIO registry focusing on cleft lip/palate and craniosynostosis level 1 datasets.

The purpose of this pilot is to assess the engagement/feasibility of centers in providing the specified data. Please be assured that the data collected will only be used for the purpose of this pilot and will not be utilized for any other purposes.

To join the pilot project, kindly fill out the Excel form that was sent to your email and submit it to ern-cranioregistry@erasmusmc.nl by July 7th.

Data Sharing Agreements

full members. These agreements are a crucial requirement to ensure that access to our Registry is granted.

We are delighted to inform you that we have already received signed Data Sharing Agreements from **3** of our full member centers, and we are committed to finalizing agreements with all our members. Furthermore, the ERN CRANIO Scientific Committee is in the final stages of developing the Data Access Policy, which will be distributed after receiving approval from the committee. Simultaneously, we are actively engaged with our Data Privacy Officer in developing a comprehensive Data Protection Impact Assessment, which will be promptly shared with your center representatives once it has been approved by our legal department.

Please feel free to reach out to us if you have any questions or require further information via the following e-mail address: ern-cranioregistry@erasmusmc.nl

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New regulation on Medical Devices

The implementation of the new European regulation (2017/745) reflects a genuine commitment to ensuring optimal patient safety. However, it has also triggered a wave of re-certification panic, resulting in the removal of important medical devices from the market and leaving a significant number of patients in distressing therapeutic dead-ends.

One area heavily impacted by this regulation is the operation of in-house 3D platforms used to produce simple, harmless devices of low complexity, such as splints for fractures or dental wafers. This complexity issue is particularly relevant in the context of rare diseases, where these devices are utilized in limited quantities.

Regrettably, the new regulation has already resulted in a clear decline in the quality of care for patients with rare diseases. The forced externalization of device production, previously available in-house, has introduced logistical challenges. Moreover, the unavailability of certain devices due to market removal necessitates the gradual degradation of existing supplies, further compromising patient well-being.

the enforcement of this regulation. Alongside the statement, we will be distributing a survey aimed at capturing and mapping the full extent of these issues.

In a collaborative effort, ERICA will distribute the statement and survey to all ERNs, gathering responses from various stakeholders involved. By analyzing these responses, we hope to identify potential solutions and advocate for necessary changes that can mitigate the adverse consequences brought about by this regulation.

Together, we can work towards finding a balance between patient safety and ensuring access to essential medical devices, particularly for those afflicted with rare diseases. Our shared commitment to addressing these issues will lead to improved care and better outcomes for patients in need.

Successful Clinical exchanges

Over the last 2 months, we have had several successful clinical exchanges again! We are very happy to hear that the clinical exchanges are received so well among our members. ***Do you still want to go on exchange during the summer? Please contact one of our project managers by clicking here.***

Learn more about our clinical exchanges!

Last May, Marizela Kljajic hosted a clinical exchange programme at Sahlgrenska University Hospital for Katerina Papadopoulou and Manja Rančigaj Gajšek with a focus on exchanging knowledge on the psychological care of patients with craniofacial anomalies.

Santa Maria visited the pediatric neurosurgery team, led by Prof. Dr. Lars Kolby, in the Sahlgrenska University Hospital. The exchange focused on craniosynostoses and the techniques used to operate children at the Sahlgrenska University Hospital. The exchange proved very fruitful!

ERN 5 Yr evaluation (AMEQUIS)

19 Dec '22 – 19 Feb '23

STAGE 1

Self-eval h

- Self-eval n by
  Ps
- tion by ERN

20 Feb '23 – 18 May '23

STAGE 2

IEB technical e tion

- Document w
- Vi ual in ews
- s

18 May '23 – 04 Sept '23

STAGE 3

Evaluation results

- Draft reports
- Comments & amendments
- Improvement plan
- Final reports

As part of the AMEQUIS framework, the third phase of the evaluation process for ERN CRANIO and its Full Members is currently in progress.

In the initial two stages, healthcare providers from ERN CRANIO completed self-evaluation forms, and some centers underwent onsite audits. Virtual interviews were conducted, and all relevant documents from the earlier stage were thoroughly reviewed.
A draft evaluation report has been shared with all stakeholders involved. Based on the preliminary findings, the ERN CRANIO network achieved a satisfactory result, with a score exceeding 85%, indicating that an improvement plan is not required. However, among the 21 evaluated centers within the network, 6 centers will need to develop an improvement plan to address specific areas for enhancement. Other centers are encouraged to provide their feedback on the draft report to further improve their performance.

15, 2023, for healthcare providers who have undergone an audit.

Learn More

ERN CRANIO updates

We want your feedback on our website!

📢 Your opinion is important to us!

We would like to improve the design, feel and content of our website and need your opinion to understand the needs within our network! The questionnaire consists of a total of 71 questions, providing answers will take 15 - 30 minutes (depending on how elaborate you are and if you skip certain sections).

📲 Please click here to access the survey!

Launch Patient-clinician group!

We are exited to announce that we can launch another group where experts and patient representatives work together on ERN CRANIO goals in partnership.

This group will consist of several patient representatives from our network and clinicians from any specialty (e.g. surgeon, geneticist, psychologist, nurse specialist, or any other specialism) within our network. The lead of the working

CPMS - user experience labs

The European Commission invites you to volunteer and improve the User Experience of the New CPMS!
They are looking for individuals to join the volunteer pool for User Experience (UX) Labs, where testing sessions are conducted by end users.

They specifically seek clinicians and non-clinicians, both current and new system users, who are interested in sharing their insights to shape the development of the New CPMS.
The main objective of the labs is to validate initial assumptions related to screens, navigation, labeling, and other aspects of the New CPMS.

These sessions will be scheduled monthly or bi-monthly and will require approximately 15-20 minutes of your time.

The European Guideline Robin Sequence

The members of the working group have been working together to develop a European Guideline on the Pierre Robin Sequence. This outstanding achievement has resulted in a professional guideline on this condition and will be published in the Journal of Craniofacial Surgery!

Congratulations to all workgroup members who have worked very hard on this achievement! Keep an eye out on our social media channels, once the guideline is published we will post on this!

Follow us on social media! Click on the icons below to go to our pages!

completing the following survey:
EUSurvey - Survey (europa.eu)

3rd ERICA General Assembly 6-7th July 2023 in Madrid, Spain

ERN CRANIO will be present at the 3rd ERICA General Assembly! The assembly is hosted by Servicio Madrileno de Salud- Hospital Universitario La Paz (SERMAS-HULP). Coordination office of ERN-TransplantChild. The aim of the General Assembly (GA) is to inform all the beneficiaries about the progress of the planned and completed ERICA project activities in more detail and to raise more awareness about the Rare Disease Research perspectives and cooperation within the ERN’s and other relevant partners.

Will you also be there? Let us know and we hope we can meet up in Madrid! Please send an e-mail to i.bos@erasmusmc.nl

ERN Wide News

3 ERN workshops opened for registration!

📢 Calling all researchers, clinicians, and healthcare specialistsinterested in cutting-edge advancements! The registration for three upcomingERN workshops is now open:

1 : "Advances in RegenerativeMedicine and Tissue Engineering for Rare Musculo-Skeletal Diseases": Discover the latestbreakthroughs in regenerative medicine and tissue engineering specificallytailored for rare musculo-skeletal diseases. Join us for this incredibleopportunity to learn from renowned experts in the field.

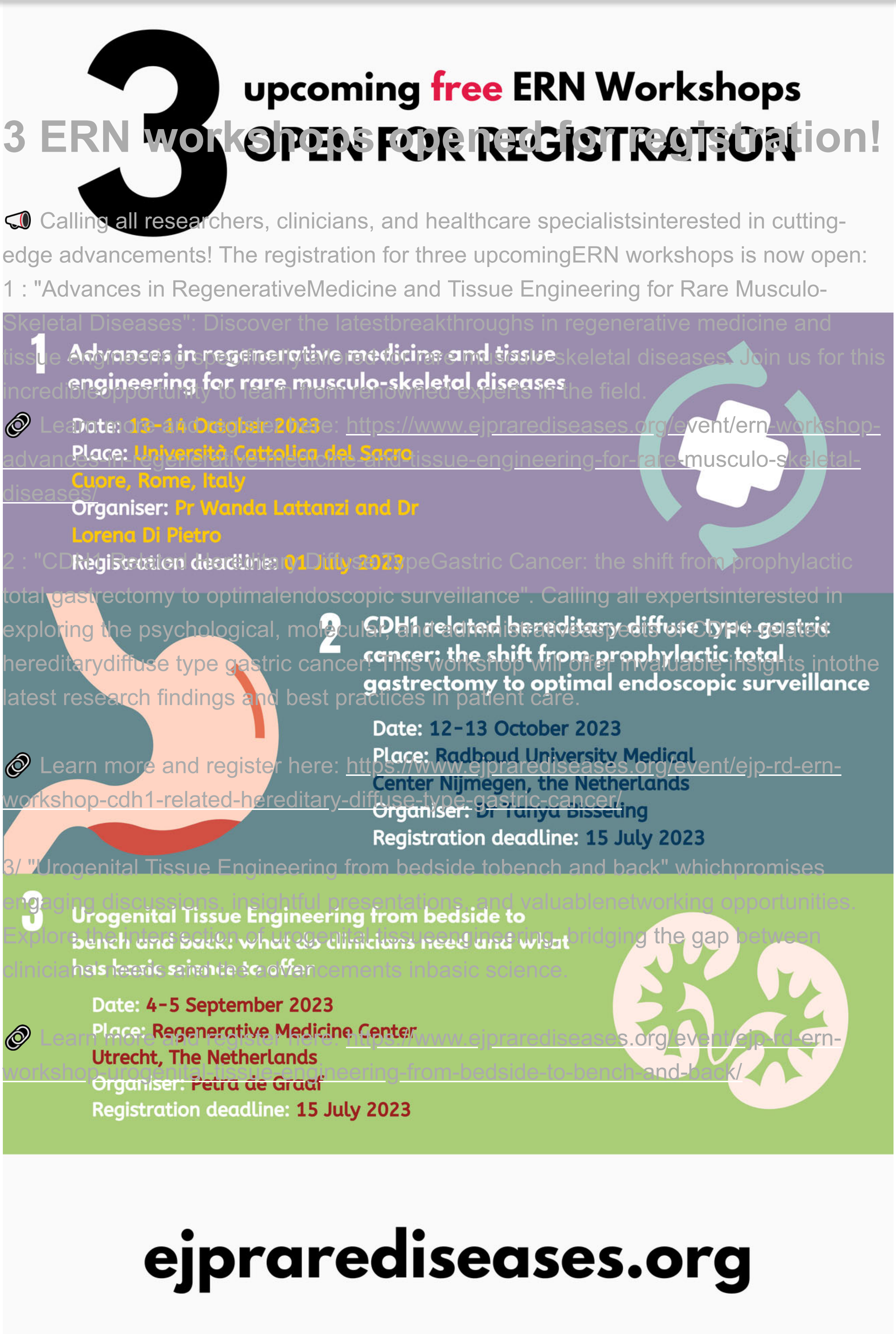
🔗 Learn more and register here: https://www.ejprarediseases.org/event/ern-workshop-advances-in-regenerative-medicine-and-tissue-engineering-for-rare-musculo-skeletal-diseases/

2 : "CDH1-Related Hereditary Diffuse TypeGastric Cancer: the shift from prophylactic total gastrectomy to optimalendoscopic surveillance". Calling all expertsinterested in exploring the psychological, molecular, and administrativeaspects of CDH1-related hereditarydiffuse type gastric cancer! This workshop will offer invaluable insights intothe latest research findings and best practices in patient care.

🔗 Learn more and register here: https://www.ejprarediseases.org/event/ejp-rd-ern-workshop-cdh1-related-hereditary-diffuse-type-gastric-cancer/

3/ "Urogenital Tissue Engineering from bedside tobench and back" whichpromises engaging discussions, insightful presentations, and valuablenetworking opportunities. Explore the intersection of urogenital tissueengineering, bridging the gap between clinicians' needs and the advancements inbasic science.

🔗 Learn more and register here: https://www.ejprarediseases.org/event/ejp-rd-ern-workshop-urogenital-tissue-engineering-from-bedside-to-bench-and-back/



3 upcoming free ERN Workshops
OPEN FOR REGISTRATION

1 Advances in regenerative medicine and tissue engineering for rare musculo-skeletal diseases

Date: 13-14 October 2023
Place: Università Cattolica del Sacro Cuore, Rome, Italy
Organiser: Pr Wanda Lattanzi and Dr Lorena Di Pietro
Registration deadline: 01 July 2023

2 CDH1 related hereditary diffuse type gastric cancer: the shift from prophylactic total gastrectomy to optimal endoscopic surveillance

Date: 12-13 October 2023
Place: Radboud University Medical Center Nijmegen, the Netherlands
Organiser: Dr Tanya Bisseling
Registration deadline: 15 July 2023

3 Urogenital Tissue Engineering from bedside to bench and back: what do clinicians need and what has basic science to offer

Date: 4-5 September 2023
Place: Regenerative Medicine Center Utrecht, The Netherlands
Organiser: Petra de Graaf
Registration deadline: 15 July 2023

ejprarediseases.org

COMPREHENSIVE
CLEFT CARE
WORKSHOP

6th International Comprehensive Cleft Care Workshop (CCCW)

The 6th International Comprehensive Cleft Care Workshop (CCCW) at the Hyatt Regency Tower in Barcelona, Spain, October 10-13, 2023 (www.cleftworkshop.org), a 3½ day meeting geared for advanced training for the cleft provider. The faculty includes 67 renowned speakers from 19 countries with a fully integrated multispecialty program.

Are you interested in this workshop? Click on the link below to find out more and to register!

Learn More

MOOC: Diagnosing Rare Diseases: from the Clinic to Research and back

Codeveloped by the Foundation for Rare Diseases, the European Reference Networks Genturis and Ithaca, and EURORDIS, this MOOC addresses the diagnostic research progress, types of genetic tests for rare diseases, and the impact of having or lacking a diagnosis on patients' lives.

The topics covered include:

- The diagnostic process and the types of genetic tests available for rare diseases
- The differences in rare genetic diseases patient pathways
- Technological advances for diagnostic research
- The role of collaborative studies and data sharing in rare diseases diagnosis
- The impact of having a diagnosis or lacking a diagnosis on patients' lives

Expert mentors will be readily available on the platform to address any questions that participants may have from May 1st to June 24th.

Learn More

CONFERENCE

Rare diseases and European reference networks:
How to ensure European solidarity for patients?

CONFERENCIA

Las enfermedades raras y las redes europeas
de referencia: **¿Cómo garantizar la solidaridad
europea a los pacientes?**

10-11/10/2023
BILBAO, BASQUE COUNTRY, SPAIN
BILBAO, PAÍS VASCO, ESPAÑA

European Economic
and Social Committee

EU23
SPANISH PRESIDENCY
COUNCIL OF THE EUROPEAN UNION

📢 Save the Date! 📅

and Social Committee (EESC) in collaboration with the Spanish Presidency of the EU Council and the Basque Country authorities.
📍 Bilbao, Euskalduna Conference Centre
📅 10-11 October 2023

Rare diseases affect a significant portion of the EU population, with around 7,000 identified conditions impacting up to 36 million people. These conditions often have a chronic, disabling, or life-threatening nature, severely impacting individuals' health and quality of life. Unfortunately, specific treatment options are limited, with up to 95% of rare diseases lacking targeted therapies or cures.
The 24 European Reference Networks (ERNs) play a crucial role in fostering collaboration among health systems at the EU level. They bring together top professionals, facilitating diagnosis, treatment, knowledge generation, and research for patients with rare diseases across Europe.

This conference aims to assess the current status and address key challenges in the EU policy framework for rare diseases and European Reference Networks. It responds to calls for action from the Czech Presidency of the Council, the previous political priority of the Trio, and active patient associations advocating for progress in this field. The event will provide an opportunity to share best practices, including those developed in the Basque Country, and work towards a comprehensive European approach that meets the needs of individuals living with rare diseases.

Excited to join the conference too? Let us know!
If you're interested in attending the conference, please reach out to the ERN CRANIO coordination team at ern-cranio@erasmusmc.nl and save the date in your calendar. Rest assured, we'll keep you updated with all the essential information as the event approaches.

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