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# ERN CRANIO

## 8th annual meeting

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**16 & 17 November 2023**  
**Dublin, Ireland**



**European  
Reference  
Network**

for rare or low prevalence  
complex diseases

 **Network**

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



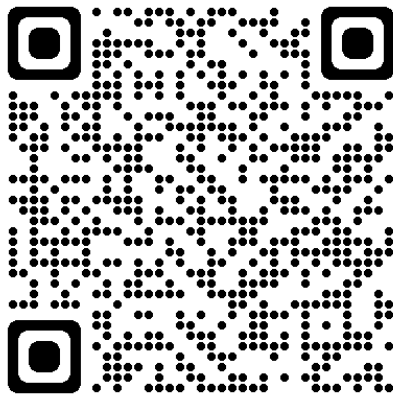
European  
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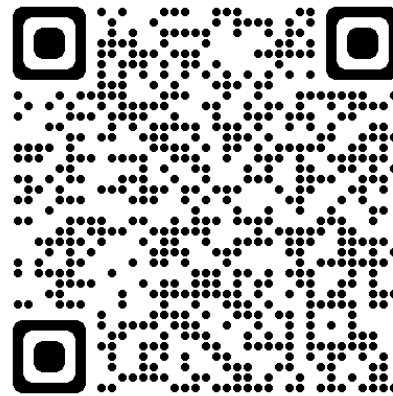
 Network

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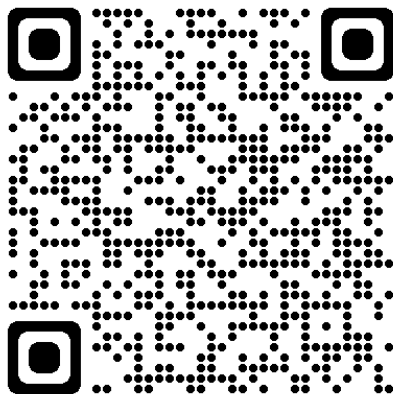
# FOLLOW US ON SOCIAL MEDIA!



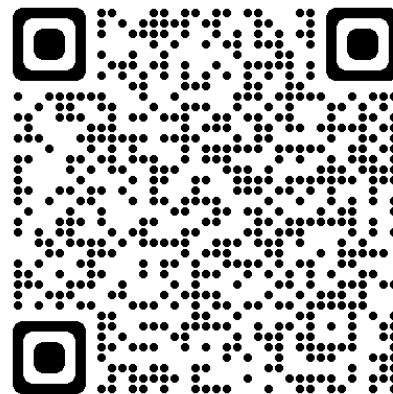
LINK TO INSTAGRAM



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**Network**

Craniofacial anomalies  
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p. 1

p. 3

p. 4

p. 6

p. 6

p. 7

p. 8

p. 9

p. 10

# CONTENT

- ◆ GENERAL PROGRAMME
- ◆ PRACTICAL INFORMATION
- ◆ EDUCATIONAL SESSION SPEAKERS
- ◆ WORKGROUP AGENDAS
  - ◆ CRANIOSYNOSTOSIS
  - ◆ CRANIOFACIAL MICROSOMIA
  - ◆ CLEFT LIP/PALATE
  - ◆ ORODENTAL ANOMALIES
  - ◆ ENT DISORDERS
- ◆ MEETING NOTES



# GENERAL PROGRAMME

## THURSDAY 16 NOVEMBER

### 14.00 - 17.00 GMT SCIENTIFIC COMMITTEE MEETING

Location: Camden Court Hotel, Dublin - YEATS ROOM

Scientific committee members only - by invitation

### 14.00 - 17.00 GMT NURSE SPECIALIST MEETING

Location: Camden Court Hotel, Dublin - BECKETT & LIBERTY ROOM

Nurse specialist members only - by invitation

### 14.00 - 17.00 GMT 3D TECHNICIAN MEETING

Location: Camden Court Hotel, Dublin - PAVILION & TIVOLI ROOM

3D technician members only - by invitation

### 14.00 - 17.00 GMT 'FACE ME' WORKSHOP

Location: Camden Court Hotel, Dublin - ABBEY ROOM

By invitation only

### 17.00 - 18.30 GMT ERN CRANIO BOARD MEETING

Location: Camden Court Hotel, Dublin - BECKETT & LIBERTY ROOM

Board members only - by invitation

### 19.00 - 20.00 GMT TOUR THROUGH DUBLIN

Location: Gather at Camden Court Hotel, Dublin

A historical tour of the Royal College of Surgeons in Ireland University of Medicine  
and Health Sciences

### 20.15 - 23.00 GMT WELCOME DINNER

Location: Brasserie Sixty6, Dublin, Ireland

Address: 66-67 South Great George's Street, Dublin, D02 YD61, Ireland



## FRIDAY 17 NOVEMBER

Location: Camden Court Hotel, Dublin - BECKETT, LIBERTY, PIVLION & TIVOLI

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08.00 - 08.15 GMT	REGISTRATION
08.30 - 09.00 GMT	WELCOME & ERN CRANIO 2023 - 2027 - <b>hybrid session</b>
09.00 - 09.30 GMT	FACE ME - <b>hybrid session</b>
09.30 - 11.15 GMT	EDUCATIONAL SESSION ON INNOVATION - <b>hybrid session</b>

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Moderated by Dylan Murray, centre representative Children's Health Ireland

- Innovation in surgery Ronan Cahill
- AI tool for speech analysis Sarah Versnel & Victor van Roey
- Automatic analysis of MRI scans for children with Craniosynostosis Henri Vrooman & Linda Gaillard
- Stem cell implication in rare craniofacial diseases: from cellular physiopathology to advanced cell-based therapies Wanda Lattanzi
- AI and the conception of a virtual expert to facilitate prediagnosis of rare diseases with Oro dental anomalies. Agnès Bloch-Zupan
- Development and surgical validation of a 3D-printed tracheostomy simulator with realistic biomechanics. Briac Thierry
- Clinical and research applications of photogrammetry in craniofacial surgery Maxime Taverne

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11.15 - 11.30 GMT CLOSING PART 1 & OPENING PART 2

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11.30- 11.45 GMT GROUP PHOTO

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11.45 - 12.30 GMT LUNCH

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12.30 - 15.30 GMT WORKGROUP SESSIONS

- Craniosynostosis working group
- Craniofacial microsomia working group
- Cleft lip/palate working group
- Oro dental working group
- Ear, Nose, and Throat (ENT) working group

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15.30 - 16.30 GMT 22q11 WORKGROUP

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# PRACTICAL INFORMATION

## HOTEL INFORMATION

**Location:** Camden Court Hotel

**Address:** Camden Street Lower, Saint Kevin's, Dublin, D02 W086, Ireland

- Check-in: 16:00u GMT
- Check-out: 12:00u GMT
- Free Wi-Fi included
- Breakfast from 07:00u - 10:30u

### From airport to hotel

- **Public transport:** busline 16 or airport Shuttle bus. Please see local transport websites for more information or use Google Maps.
  - **Travel time:** 40 - 60 mins
- **Taxi/town car:** taxi's are available at the airport
  - **Travel time:** 30 mins

## TOUR THROUGH DUBLIN INFORMATION

**Location:** Lobby, Camden Court Hotel

**Address:** Camden Street Lower, Saint Kevin's, Dublin, D02 W086, Ireland

Gather at Hotel Lobby at 19:00u.

## RESTAURANT BRASSERIE SIXTY6 INFORMATION

**Location:** Brasserie Sixty6, Dublin

**Address:** 66-67 South Great George's Street, Dublin, D02 YD61, Ireland

If you will not join the tour through Dublin, please find the travel directions from the hotel to the restaurant below:

- **Public transport:** tramline 83, 9, 16, or 122 goes from the hotel to the restaurant. Please see local transport websites for more information or use Google Maps.
  - **Travel time:** +/- 10 mins
- **Walking:** it is a 15 minute walk from the hotel to the restaurant. Please use Google Maps for more information.



# EDUCATIONAL SESSION SPEAKER INFORMATION

## INNOVATION IN SURGERY

**Speaker: Ronan Cahill, Children's Health Ireland**

This presentation will discuss innovation in surgery by Professor Ronan Cahill, Director UCD Centre for Precision Surgery & Digital Surgery Unit, Mater Misericordiae University Hospital, Dublin, Ireland.

## AI TOOL FOR SPEECH ANALYSIS

**Speakers: Sarah Versnel & Victor van Roey, Erasmus MC**

This presentation will discuss a project plan to investigate the use of AI for the assessment of speech in cleft patients across Europe. This AI tool can be used as an innovative tool to assist speech & language therapists in speech assessments.

## AUTOMATIC ANALYSIS OF MRI SCANS FOR CHILDREN WITH CRANIOSYNOSTOSIS

**Speakers: Henri Vrooman & Linda Gaillard, Erasmus MC**

This presentation will discuss automatic analysis of MRI scans for children with craniosynostosis with a particular focus on metopic synostosis. Tissue segmentation for determining global brain volumes and lobe-specific volumes, ventricle segmentation and tractography will be discussed.

## STEM CELL'S IMPLICATION IN RARE CRANIOFACIAL DISEASES: FROM CELLULAR PHYSIOPATHOLOGY TO ADVANCED CELL-BASED THERAPIES

**Speaker: Wanda Lattanzi, Fondazione Policlinico Universitario A. Gemelli**

Stem cells are key regulators for tissue homeostasis and repair in adult tissues, where they reside in dedicated microenvironments known as "stem cell niches". Defects and/or malfunctions in craniofacial stem cell niches can disrupt craniofacial development and morphogenesis and are implicated in the pathophysiology of rare diseases. Targeting stem cells in craniofacial rare diseases hence represents a suitable strategy in the design and development of experimental treatments and precision medicine approaches. I will present as paradigm example, data from craniosynostosis research performed in my lab.



## AI AND THE CONCEPTION OF A VIRTUAL EXPERT TO FACILITATE PREDIAGNOSIS OF RARE DISEASES WITH ORODENTAL ANOMALIES

**Speaker: Agnès Bloch-Zupan, Hopitaux universitaires de Strasbourg**

The D-IA-GNO-DENT project, hosted by CHRU de Strasbourg - Hôpitaux Universitaires de Strasbourg, Reference Center for Rare Oral and Dental Diseases, CRM R O-Rares, Université de Strasbourg, IGBMC, CERBM-gie, the Fondation Maladies Rares proposes to develop AI solutions for early detection of rare oral diseases. These conditions often impact multiple organ systems and can be identified through the recognition of teeth developmental abnormalities signatures. The objective is to promote access to prediagnosis of these conditions for both patients and health professionals and to orientate them to expert centers and networks ultimately reducing the diagnostic odyssey.

## DEVELOPMENT AND SURGICAL VALIDATION OF A 3D-PRINTED TRACHEOSTOMY STIMULATOR WITH REALISTIC BIOMECHANICS

**Speaker: Briac Thierry, Hôpital Universitaire Necker Enfants-Malades**

The objectives of the development and surgical validation of the 3D-printed tracheostomy stimulator with realistic biomechanics were to raise students' awareness of the dimensions of the neonatal pediatric model and the particularities of pediatric tracheostomy.

## CLINICAL AND RESEARCH APPLICATIONS OF PHOTOGRAMMETRY IN CRANIOFACIAL SURGERY

**Speaker: Maxime Taverne, Hôpital Universitaire Necker Enfants-Malades**

Photogrammetry is the field of optics and geometry that enables the three-dimensional digitization of an object based on the combination of multiple photographic images. In clinics, it is a very useful technique which contributes to non-invasively obtaining 3D morphometric data on patients. In the context of promoting massive, fast and non-invasive phenotyping within the ERN Cranio, we here present a range of examples of clinical questions that photogrammetry may be able to address.





# WORKGROUP AGENDAS

## CRANIOSYNOSTOSIS

Location: Beckett & Liberty room, Camden Court Hotel

12.30 - 12.50 GMT UPDATE ON ERN CRANIO REGISTRY Linda Gaillard

13.00 - 13.45 GMT UPDATE 3D WORKING GROUP Maxime Taverne, Guido de  
Jong, Tareq Abdel Alim

13.45 - 14.05 GMT REDO SCAPHOCEPHALY Marie-Lise van Veelen-Vincent

### BREAK

14.20 - 14.40 GMT THERAPEUTIC PATIENT EDUCATION Giovanna Paternoster

14.40 - 15.00 GMT PATIENT-PARTNERSHIP Patient representatives

15.00 - 15.20 GMT QUALITY OF LIFE ASSESSMENT Marizela Kljajic

15.20 - 15.30 GMT CLOSING OF THE MEETING Marie-Lise van Veelen-Vincent



## CRANIOFACIAL MICROSOMIA

Location: Yeats Room, Camden Court Hotel

12.30 - 12.35 GMT	WELCOME	Eppo Wolvius
12.35 - 12.55 GMT	CFM REGISTRY DATASET	Victor van Roey
12.55 - 13.25 GMT	UPDATE FACIAL DYSOSTOSIS CONSENSUS STATEMENT	Victor van Roey
13.30 - 14.00 GMT	IMPROVING PATIENT COMMUNICATION AND EXPECTATION MANAGEMENT	Ivana Marinac
<b>BREAK</b>		
14.15 - 15.00 GMT	RESEARCH PROJECTS IN CFM	
● 14.15 - 14.30 GMT	3D planning of surgery	Eppo Wolvius
● 14.30 - 14.45 GMT	How to use 3D imaging in research	Eppo Wolvius
● 14.45 - 15.15 GMT	Treatment of children with HMF and the use of microsurgery	Krzysztof Dowgierd
15.15 - 15.30 GMT	WRAP-UP AND ANY OTHER BUSINESS	Eppo Wolvius



## CLEFT LIP/PALATE

Location: Pavilion & Tivoli Room, Camden Court Hotel

12.30 - 12.35 GMT	WELCOME	Aebele Mink van der Molen
12.35 - 12.45 GMT	ERN CRANIO NEWS & UPDATES	Aebele Mink van der Molen
12.45 - 13.10 GMT	ERN CRANIO CLEFT LIP/PALATE REGISTRY	
	 DATASET CONSENSUS	Philip van der Goes
	 REGISTRY UPDATE & DASHBOARD UPDATE	Philip van der Goes
13.10 - 13.20 GMT	ERN CRANIO UPDATE SCIENTIFIC COMMITTEE	Aebele Mink van der Molen
13.20 - 14.00 GMT	RESEARCH REGULATIONS & PROPOSALS	Aebele Mink van der Molen
<b>BREAK</b>		
14.15 - 14.50 GMT	FUTURE INNOVATIONS	Aebele Mink van der Molen
14.50 - 15.05 GMT	ERN CRANIO 2023 - 2027 PROJECTS	Aebele Mink van der Molen
15.05 - 15.15 GMT	UPDATES PATIENT REPRESENTATIVES	Jana Angelova
15.20 - 15.30 GMT	WRAP-UP AND AOB	Aebele Mink van der Molen



## ORODONTAL ANOMALIES

Location: Gallery Room, Camden Court Hotel

12.30 - 12.35 GMT WELCOME Agnès Bloch-Zupan

12.35 - 13.35 GMT ERN CRANIO REGISTRY Agnès Bloch-Zupan

◆ General update + dataset development

13.35- 14.05 GMT ERN CRANIO UPDATE SCIENTIFIC COMMITTEE

Agnès Bloch-Zupan  
& Ikram L'Khssim

◆ Research opportunities

### BREAK

14.20 - 14.50 GMT ERN CRANIO NEWS AND UPDATES

Agnès Bloch-Zupan  
& Ikram L'Khssim

◆ ORPHANET

14.50 - 15.25 GMT ERN CRANIO 2023 - 2027 PROJECTS

Agnès Bloch-Zupan  
& Ikram L'Khssim

15.25 - 15.30 GMT WRAP-UP AND AOB

Agnès Bloch-Zupan



## ENT DISORDERS

Location: Abbey room, Camden Court Hotel

12.30 - 12.35 GMT      WELCOME      Bas Pullens

12.35 - 13.25 GMT      ERN CRANIO 2023 - 2027      Bas Pullens & Ikram L'Khssim

- Development of educational videos
- Webinar series
- CPMS 2.0
- Guideline development

13.25 - 13.55 GMT      ERN CRANIO UPDATE SCIENTIFIC COMMITTEE      Bas Pullens

- Research opportunities

### BREAK

14.10 - 15.15 GMT      ERN CRANIO REGISTRY      Philip van der Goes

- Update on ERN CRANIO registry
- Update and discussion rare congenital deafness dataset

15.15 - 15.30 GMT      WRAP-UP AND AOB      Bas Pullens





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Sláinte Leanaí Éireann



Children's Health Ireland



The 8th annual meeting of ERN CRANIO was organised in collaboration with the Children's Health Ireland Hospital in Dublin, Ireland.

A special thanks goes out to the Dublin team for helping to organise this event!