European Reference Network
for rare or low prevalence complex diseases

Network
Inherited and Congenital Anomalies (ERNICA)

NEWSLETTER

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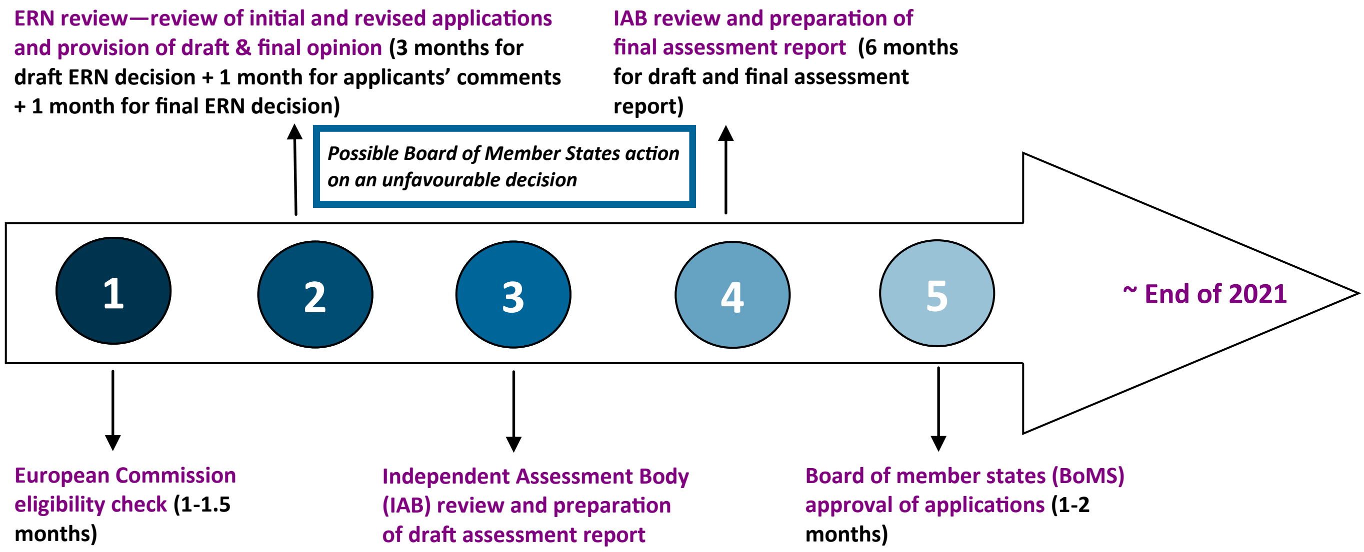
THE 2ND CALL FOR FULL ERN MEMBERSHIP— CLOSED!

The second call for new healthcare centres to join the ERNs as full members officially closed on **30th November 2019.** Across all 24 ERNs, a total of 841 applications were received. ERNICA received **44 applications**.

What happens now?



ERN review—review of initial and revised applications and provision of draft & final opinion (3 months for draft ERN decision + 1 month for applicants' comments + 1 month for final ERN decision)

IAB review and preparation of final assessment report (6 months for draft and final assessment report)

Possible Board of Member States action on an unfavourable decision

European Commission eligibility check (1-1.5 months)

Independent Assessment Body (IAB) review and preparation of draft assessment report

Board of member states (BoMS) approval of applications (1-2 months)

For more detailed information on the assessment process click here.

The call for affiliated partners is also now closed. These partnerships are now being formalised and an integration strategy is being prepared (European Commission deadline: end of January 2020).

ERNICA GUIDELINE FOR GASTROSCHISIS: INITIAL DISCUSSIONS

On 2nd December 2019, 23 ERNICA representatives from 10 member states met at Erasmus MC, Rotterdam to discuss the development of a guideline for gastroschisis. The group included obstetricians, fetal and pediatric surgeons, neonatologists and pediatricians and the meeting was led by Carmen Mesas Burgos (Karolinska University Hospital) and Lucas Wessel (University Hospital Mannheim).

Aims of the meeting: **1)** to summarise the available evidence on the management of gastroschisis (background to obtaining international expert consensus within ERNICA). **2)** To discuss which kind of guideline should be established (and in what form)

Discussion points: The starting point was the 2014 Danish gastroschisis guideline. Participants from Denmark provided valuable information on GRADE methodology and the need to formulate PICO questions to initiate the development of an ERNICA guideline. The development of a guideline rather than a consensus statement was agreed upon.

Topics to be considered are: **pre-natal management** (diagnosis and surveillance: do patients benefit?), **delivery** (when, where and how?), **post-natal management** (Operation immediately? Sedation or general anaesthesia?), **type of closure** (silo, what about when facial closure is not possible?) **surgery on complicated gastroschisis**, **other areas of post-natal management** (such as antibiotics, feeding) and **long-term follow-up.**

WHAT'S GOING ON IN THE ESOPHAGEAL DISEASES WORKING GROUP?

2nd ERNICA consensus conference: "Long-gap Esophageal Atresia: Perioperative, Surgical and Long-term Management" : Berlin, 13 – 14 November 2019

The 2nd ERNICA consensus conference on the *Perioperative, Surgical and Long-term Management of Patients with Long-gap Esophageal Atresia* took place in Berlin on the 13th and 14th November 2019. After an extensive literature search and preparation, relevant aspects regarding the management of patients with long-gap esophageal atresia were discussed. Consensus was reached on 90 out of 99 items (91%) after an anonymous online voting process.

24 ERNICA representatives attended, including pediatric surgeons, pediatric gastroenterologists, a neonatologist, a pulmonologist, a methodologist and patient representatives.

The consensus statements will be published in the European Journal of Pediatric Surgery. They will facilitate dissemination of evidence, standardisation and improvement of patient care, and will contribute to reducing morbidity in patients with long-gap esophageal atresia.

Esophageal atresia patient journey workshop: Berlin, 15 November 2019

Further progress was made on the esophageal atresia 'Patient Journey' project at a workshop held in Berlin on November 15. This project has a multidisciplinary team with its membership drawn from surgery, nursing and patient representatives.

The project has built on the work presented at the 2019 annual meeting in Padua and has identified eleven major stages in the patient journey from pre-natal to retirement. The key issues at each stage in the Patient Journey are being identified from both the clinical and parent/patient perspectives and in addition to collating relevant details (such as the follow-up checks at particular stages) the description of the journey is enhanced by the use of illustrations (which bring 'life' to the journey) and 'patient friendly' summaries which aim to simplify the associated clinical detail.

Two particular aspects of this project are (i) to build on existing good practices (e.g. the after-care and follow-up programmes developed by (or in conjunction with) patient groups in France, Germany and The Netherlands) and (ii) to identify gaps in good practice and/or information which should be addressed. A further workshop is planned for the New Year when it is intended to involve additional medical disciplines and the team also hopes to make further progress at the next ERNICA annual meeting in Lille, where it will also present an update on the progress made.

First patient-led survey in CDH

CDH UK is a registered charity that seeks to provide support to patients with congenital diaphragmatic hernia (CDH) and their families. This is provided in the form of emotional and practical support and by encouraging and funding relevant research in the field. CDH UK is a patient organisation affiliated with the ERNICA network.

ERNICA's CDH UK representative, Beverley Power, has recently published a paper in collaboration with ERNICA representatives from Great Ormond Street Hospital, London titled ***Long term feeding issue and its impact on the daily life of congenital diaphragmatic hernia survivors: Results of the first patient-led survey*** *(Pediatric Surgery International, 2019).* **Click on the link to read!**

OTHER RELEVANT MEETINGS, EVENTS & DEADLINES FOR YOUR INFORMATION:

JANUARY 2020

20th: Deadline for ERNICA centres to submit their 2019 monitoring data

FEBRUARY 2020

18th: European Joint Programme on Rare Diseases (EJP RD) Joint Transnational Call - pre-proposal submission deadline

MARCH 2020

6th: 2020 ERNICA intestinal failure expert meeting (details to be confirmed)
11-14th: The International Congress of Research on Rare and Orphan Diseases, (Re[act] congress and IRDiRC conference), Berlin
15th: EJP RD Internal call: Clinical trials methodology demonstration projects—initial deadline.

APRIL 2020

22nd: ERNICA research meeting, Lille (details to be confirmed)
23rd-24th: ERNICA annual meeting, Lille

Save the date!!

MAY 2020

15-16th: The 10th European Conference on Rare Diseases and Orphan Products, Stockholm