ERNICA RESEARCH CATALOGUE

Updated 22 April 2024

Table of Content

Origin

Oesomics Anastomose Molecular Signatures of Esophageal Atresia

Description: This project aims to identify transcriptomic pathways which help to better understand mechanisms of EA and its different forms, as well as help to prenatal diagnosis of this rare disease.

Workstreams: Esophageal Diseases; Fetal Medicine

Keywords: esophageal atresia, epigenetic, protogenomic, metabolomics, amniotic fluid

Principal Investigator: Frédéric Gottrand, University of Lille

Contact: melanie.leroy@chu-lille.fr

Partners / centres involved: PRISM (INSERM Nord Ouest), Go@L (Université de Lille), Bilille (Université de Lille)

Status: In Progress

Origin

Thoracoscopic Stage Internal Traction Repair Reduces Time to Achieve Esophageal Continuity in Long Gap Esophageal Atresia

Description: This study aims at comparing the management of long gap esophageal atresia (LGOA) between two high-volume centers. Conclusions were that thoracoscopic internal traction technique reduces time to achieve esophageal continuity and the need for esophageal substitution while maintaining a similar early complication rate.

Workstreams: Esophageal Diseases

Keywords: long gap esophageal atresia, esophageal atresia, internal traction, minimally invasive surgery, delayed primary anastomosis

Principal Investigator: Dominika Borselle

Contact: dominika.borselle@umw.edu.pl

Partners / centres involved: Department of Paediatric Surgery and Urology, Wroclaw Medical University, Poland; Great Ormond Street Institute of Child Health, University College London, UK; Department of Specialist Neonatal and Paediatric Surgery, Great Ormond Street Hospital for Children, NIHR, London, UK

Status: Completed/published

Origin

Functional genetics first: evaluation of disturbed biological pathways underpinning defective esophageal development

Description: Evaluation of candidate pathways responsible for disturbed esophageal development using in-vitro and in-vivo models

Workstreams: Esophageal Diseases

Keywords: Esophageal Atresia, functional genetics, clinical genetics

Principal Investigator: Erwin Brosens

Contact: e.brosens@erasmusmc.nl

Partners / centres involved: Erasmus MC

Status: In progress

Origin

Transition into adulthood: elucidating the biological mechanisms of increased esophageal cancer risk in adult patients born with esophageal atresia

Description: Elucidating the biological mechanisms of increased esophageal cancer risk in adult patients born with esophageal atresia. Molecular genetix experiments, in-vitro experiments.

Workstreams: Esophageal Diseases; Transition

Keywords: Esophagus, Barrett, Esophageal Cancer

Principal Investigator: Erwin Brosens

Contact: e.brosens@erasmusmc.nl

Partners / centres involved: Manon Spaander, Erasmus MC

Status: In progress

Origin

Beyond coding: short and long read whole genome and methylation sequencing to explain missing heritability in isolated and complex EA and CDH

Description: Short and long read whole genome and methylation sequencing to explain missing heritability in isolated and complex esophageal atresia and congenital diaphragmatic hernia

Workstreams: Esophageal Diseases; Malformations of the Diaphragm

Keywords: whole genome sequencing, methylation sequencing

Principal Investigator: Erwin Brosens

Contact: e.brosens@erasmusmc.nl

Partners / centres involved: Erasmus MC

Status: Consortium building

Origin

Identifying the culprits: rna seq to find the genes responsible for esophageal atresia and Congenital Diaphragmatic Hernia in copy number variations

Description: Transcriptome studies in esophageal and diaphragm biopsies and fibroblasts, Copy Number Variation Profiling

Workstreams: Esophageal Diseases; Malformations of the Diaphragm

Keywords: RNA seq, CDH, EA/TEF Copy Number variation

Principal Investigator: Erwin Brosens

Contact: e.brosens@erasmusmc.nl

Partners / centres involved: Erasmus MC, Department of General, Visceral, Vascular and Thoracic Surgery, Unit of Pediatric Surgery, Universitätsklinikum Bonn, Bonn, Germany.

Status: In progress

Origin

The MICKEY MOUSE trial: MICrobiota, KEY between Mother and Offspring USsing SEquencing techniques

Description: Prospective 'multi-omics' trial including 150 preterm neonates at high risk for NEC as well as their mothers. The trial will analyse among others the fecal microbiome, metabolome, inflammasome and (epi)genome of the neonate as well their mothers to offer insight in the pathophysiology of NEC, thereby identifying possible biomarkers and putative tailored prevention strategies.

Workstreams: Intestinal Diseases

Keywords: necrotising enterocolitis, prospective trial, multi-omics

Principal Investigator: J.B.F. Hulscher and E. Kooi

Contact: j.b.f.hulscher@umcg.nl

Partners / centres involved: University Medical Center Groningen

Status: In Progress

Origin

The use of Artificial Intelligence in prediction and clinical decision making in NEC

Description: Behavioral AI technology will be used to assess those factors that are important in the decision to proceed to surgery versus palliative care in critically ill neonates with NEC, with the aim of constructing a decision aid for clinicians faced with this dilemma.
Simultaneously, Q methodology will be used to investigate parental factors and develop a decision aid for parents, to improve true shared decision making.
Finally the use of AI will be investigated to predict NEC in high risk neonates, aiming to develop an algorithm that can identify neonates who develop NEC before they actually do.

Workstreams: Intestinal Diseases

Keywords: necrotising enterocolitis, artificial intelligence, shared decision making, prevention

Principal Investigator: J.B.F. Hulscher

Contact: Rosa Verhoeven (PhD student, r.verhoeven@umcg.nl)

Partners / centres involved: UMCG, members from all ERNICA centers have been approached to complete the choice experiments

Status: In Progress

Origin

Fecal microbiota transplantation (FMT) for the prevention and treatment of NEC in a mouse model

Description: This study will investigate the influence of the bacterial composition of the intestine on the incidence of NEC and its severity in the NEC mouse model by manipulating the intestinal microbiota using fecal microbiota transplantation.

Workstream: Intestinal Diseases

Keywords: Fecal microbiota transplantation, necrotizing enterocolitis

Principal Investigators: Prof. Dr. Martin Lacher, PD Dr. Steffi Mayer

Contact: Martin.Lacher@medizin.uni-leipzig.de, Steffi.Mayer@medizin.uni-leipzig.de

Partners / centres involved: Helmholtz Centre for environmental research, Leipzig, Germany

Status: In progress

Origin

Necrotizing Enterocolitis Core Indicator Set (NECCIS) project

Description: The objective is to achieve a set of quality indicators for NEC that can be used to measure and compare quality of care between different centers. The selection of the indicators set follows a methodological design so that everybody recognizes this tool in quality analysis. The resulting set will be implemented in a European Clinical Audit for necrotizing enterocolitis (EPSA|ERNICA Registry).

Workstreams: Intestinal Diseases

Keywords: Necrotising enterocolitis, core indicator set

Principal Investigator: J.B.F. Hulscher

Contact: Otis C. van Varsseveld (o.c.van.varsseveld@umcg.nl)

Partners / centres involved: NECCIS steering committee consisting of Marijn J. Vermeulen (Care4Neo), Marc Miserez, Marie Spruce (NEC UK Charity), Miriam Duci, Rony Sfeir, Carmen Mesas Burgos, Simon Eaton, Lucas Matthyssens, Mohamed R. Boukhris, Martin Lacher, Elena Palleri, Lorenzo Norsa, Elisabeth M.W. Kooi, Johannes W. Duess and Daniel Rossi (ERNICA), Marlinde van der Kamp (ERNICA), Jan B.F. Hulscher and Otis C. van Varsseveld (University of Groningen), Irene de Haro Jorge and Jordi Prat Ortells (Hospital Sant Joan de Déu Barcelona)

Status: In Progress

Origin

The Premier Core Set Of Quality Indicators For Hirschsprung's Disease Care

Description: This study aimed to establish a consensus-driven core set of quality indicators to benchmark HSCR care across hospitals, regions, or countries within a European clinical audit framework.

Workstreams: Intestinal Diseases; Gastroenterological Diseases

Keywords: Hirschsprung's disease, indicators, quality of care

Principal Investigator: Daniel Rossi

Contact: d.rossi@dica.nl

Partners / centres involved: Erasmus MC, Karolinska institutet

Status: In progress

Origin

Unveiling a decade of the European Pediatric Surgery Audit: comprehensive insights into Hirschsprung's Disease in the Netherlands

EPSA
EUROPEAN PEDIATRIC SURGICAL AUDIT

Description: This study aims to provide a detailed overview of Hirschsprung's Disease (HSCR) in the Dutch population using data from the European Pediatric Surgery Audit (EPSA). The study, a multicenter retrospective cohort study, intends to align national practices with the ERNICA guidelines, enhance medical care standards, and contribute to a more comprehensive understanding of HSCR.

Workstreams: Intestinal Diseases; Gastroenterological Diseases

Keywords: Hirschsprung's disease, outcomes, genetic factors, imaging

Principal Investigator: Daniel Rossi

Contact: d.rossi@dica.nl

Partners / centres involved: UMCG, EMC, MUMC, UMCU, Radboud UMC, AMC

Status: Preparation phase/writing protocol

Origin

Examining definitions and diagnostic criteria of Hirschsprung's related Enterocolitis and Outlet obstruction among ERNICA centers

Description: This study aims to investigate the variability in definitions and diagnostic criteria of Hirschsprung-associated enterocolitis (HAEC) and outlet obstruction, involving a survey distributed to pediatric surgeons and gastroenterologists at ERNICA centers.
The study aims to understand differing diagnostic approaches and contribute to standardizing care for HSCR patients, and emphasizes the importance of standardizing HAEC and outlet obstruction diagnosis and treatment across expert centers in Europe.

Workstreams: Intestinal Diseases; Gastroenterological Diseases

Keywords: Hirschsprung's disease, outcomes, genetic factors, imaging

Principal Investigator: Daniel Rossi

Contact: d.rossi@dica.nl

Partners / centres involved: UMCG, EMC, MUMC, UMCU, Radboud UMC, AMC

Status: Preparation phase/writing protocol

Origin

How to assess, maintain and sustain data quality: an overview of completeness and accuracy of four years of registering in the Dutch Pediatric Surgical Audit

EPSA
EUROPEAN PEDIATRIC SURGICAL AUDIT

Description: The study evaluates data completeness and accuracy, assessing its utility for quality of care, benchmarking, and research.

Workstreams: Esophageal Diseases, Intestinal Diseases, Gastroenterological Diseases, Malformations of the Diaphragm and AWD

Keywords: Data quality, verification, audit

Principal Investigator: Nadine Teunissen

Contact: Daniel Rossi, PhD student, d.rossi@dica.nl

Partners / centres involved: UMCG, EMC, MUMC, UMCU, Radboud UMC, AMC

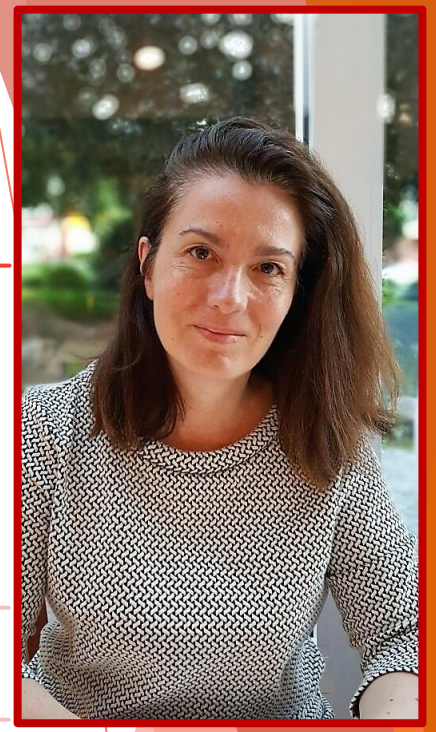
Status: In progress

Origin

XanthoVision

ClinicalTrials.gov

APHP website



Description: Role of xanthophylla in visual function: a randomised study. The aim of this study is to compare visual function before and after XANTH supplementation in patients with exclusive artificial nutrition and XANTH deficiency, in order to prove their role in visual function.

Workstreams: Nutrition;

Keywords: artificial nutrition, enteral nutrition, visual function

Principal Investigator: Francisca Joly

Contact: francisca.joly@aphp.fr

Partners / centres involved: French multicentric study

Status: In progress

Origin

Intestinal Organoids: A Model to Study Necrotizing Enterocolitis (NEC) and The Influence of modulating agents on the intestinal ephitelial injury.

Description: This study aims to create an ex vivo model of NEC tissue damage mechanism, starting from tissues of healthy neonates, to examine in depth cellular responses (cellular damage and intestinal epithelial regeneration after injury). Secondarily, this study aims to depict the effects of modulating agents on damage mechanism into the NEC model.

Workstreams: Intestinal Diseases;

Keywords: NEC, Organoids, Human specimens, Extracellular Vesicles

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: Padua University Hospital

Status: In progress

Origin

Enhancing diagnosis of Hirschsrpung disease using Artificial intelligence based method

Description: This study introduces an AI-Based method aimed at automating the identification of ganglionic cells and hypertropic nerves in the specimens of patients with Hirschsprung's disease.

Workstreams: Intestinal Diseases;

Keywords: Artificial intelligence, Diagnosis, Hirschsprung disease, Histology.

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: Study performed in collaboration with engineers from Padua University

Status: In progress

Origin

Pathological processes of abnormal lung development in congenital diaphragmatic hernia

Description: The aims of this project are to better understand the pathogenesis of lung hypoplasia or pulmonary hypertension and to develop new and innovative therapeutic approaches based on the analysis of molecular target proteins.

Workstream: Malformations of the Diaphragm

Keywords: Congenital diaphragmatic hernia, lung hypoplasia, pulmonary hypertension, inflammation

Principal Investigators: PD Dr. Richard Wagner

Contact: Richard.Wagner@medizin.uni-leipzig.de

Partners / centres involved: Harvard Medical School, Universtiy of Manitoba, Faculty of Medicine Paris Sud

Status: In progress

Origin

Epidemiology, pathophysiology and outcome of autoimmune pancreatitis in children

Description: Autoimmune pancreatitis is a very rare disease in children. We recently have summarized the litterature and came up with diagnostic and management recommendations.

The aim of this follow-up study is to determine the spectrum of the disease, segragate patients depending on immune/inflammatory etiology and evaluate the impact of treatment on long term outcome.

Workstream: Gastroenterological Diseases

Keywords: AIP, autoimmune pancreatitis, inflammatory pancreatitis, inflammatory bowel disease

Principal Investigators: Isabelle Scheers

Contact: isabelle.scheers@saintluc.uclouvain.be

Partners / centres involved: ESPGHAN

Status: Pending ethical approval

Origin

A reverse translational approach for the study of Tricho-Hepato-Enteric Syndrome: a paradigm disease for inflammatory bowel disorders

Description: The main aim of the study is to investigate the pathogenic mechanisms leading to enteropathy in TTC37 deficiency, and to identify novel potential therapies for THES through a bedside to bench reverse translational research approach.

Workstream: Gastroenterological Diseases

Keywords: TTC37, congenital enteropathy, rare IBD, intestinal failure, tricho-hepato-enteric syndrome

Principal Investigators: Giorgio Perilongo, Padua University Hospital

Contact: Mara Cananzi (mara.cananzi@aopd.veneto.it)

Partners / centres involved: Fondazione Istituto di Ricerca Pediatrica Città della Speranza, Padua, Italy

Status: In progress

Origin

Development and therapeutic application of an extracorporeal normothermic perfusion system in intestinal transplantation: the end of the ice age

Description: Development and therapeutic application of an extracorporeal normothermic perfusion system in intestinal transplantation.

Workstream: Intestinal Diseases; Intestinal Failure

Keywords: Normotermic perfussion, Intestinal Transplantation, Children

Principal Investigators: Ane Miren Andrés

Contact: ane.andres@salud.madrid.org

Partners / centres involved: Children Hospital La Paz

Status: In progress

Origin

Prevention of rejection by adoptive transfer of regulatory T lymphocytes in an experimental model of intestinal transplantation

Description: Prevention of rejection by adoptive transfer of regulatory T lymphocytes in an experimental model of intestinal transplantation.

Workstream: Intestinal Diseases; Intestinal Failure

Keywords: Intestinal Transplantation, Rejection

Principal Investigators: Francisco Hernandez Oliveros

Contact: fhernandezo@salud.madrid.org

Partners / centres involved: Children Hospital La Paz

Status: In progress

Origin

Multiomics of congenital disease including structured multimodal follow-up

Description: All children with congenital malformation are characterized on various levels including genetic and quality of life in our lifelong structured and multimodal follow-up.

Workstreams: Esophageal Diseases; Intestinal Diseases; Gastroenterological Diseases; Quality of Life; Malformations of the Diaphragm and AWD; Fetal Medicine

Keywords: multiomics CDH, esophageal atresia, ARM

Principal Investigator: Michael Boettcher

Contact: michael.boettcher@umm.de

Partners / centres involved: Mannheim, Rotterdam, Boston, Leipzig, Winnipeg

Status: In progress

Origin

Immunomodulation in ECMO

Description: Immunomodulation in ECMO: influencing innate immunity to improve alveogenesis.

Workstreams: Malformations of the Diaphragm and AWD;

Keywords: ECMO, CDH immunity

Principal Investigator: Michael Boettcher

Contact: michael.boettcher@umm.de

Partners / centres involved: Mannheim
Tobias Fuchs, Boston

Status: In progress

Intervention: Clinical Trials

STEPS-EA Trial

Publication

Explanatory video

Description: The STEPS-EA trial is an ERNICA multi-center randomized controlled trial looking at intralesional steroid injections to prevent refractory strictures in patients with esophageal atresia. The trial involves 1:1 randomization to injection with 10 mg/ml TAC (Kenacort-A 10) prior to balloon dilatation and balloon dilatation without any injection.

Workstream: Esophageal Diseases

Keywords: Esophageal atresia; intralesional steroid injections; anastomotic strictures

Principal Investigator: Dr. John Vlot, Erasmus MC

Contact person: Anne-Fleur van Hal, Erasmus MC

Contact: steps.ea@erasmusmc.nl

Partners / centres involved: Helsinki University Hospital, Centre Hospitalier Universitaire de Lille, Odense University Hospital, Rigshospitalet Copenhagen, Skåne University Hospital, Karolinska University Hospital, Erasmus MC - Sophia Childrens Hospital

Status: In progress

Intervention: Clinical Trials

A Randomized, Double-Blind, Parallel-Group, Placebo-Controlled Phase 3 Efficacy and Safety Study of Tezepelumab in Patients With Eosinophilic Esophagitis (CROSSING).

Description: A randomized, double-blind, placebo-controlled multicenter, phase 3 study to evaluate the efficacy and safety of tezepelumab administered subcutaneously (SC) using an accessorized pre-filled syringe (APFS) versus placebo in adult and adolescent patients with eosinophilic esophagitis (EoE).

Workstreams: Esophageal Diseases

Keywords: eosinophilic esophagitis, tezepelumab, Placebo

Principal Investigator: Antonio Gasbarrini

Contact: Irene Spinelli (irene.spinelli@guest.policlinicogemelli.it)

Partners / centres involved: See QR code

Status: In progress

Intervention

Study of the Effect of the Time of Diagnosis (Antenatal vs. Postnatal) on the Post-traumatic Reactions of Parents of Children Undergoing Oesophageal Atresia Surgery (DANAO)

Description: This is an observational, national multicentre (34 centres involved), cross-sectional study in psychology whose objective is to describe and compare the levels of post-traumatic reactions of mothers to the announcement of their child's EA diagnosis. An ancillary study will be carried out among the fathers present who agree to participate in the study.

Workstreams: Esophageal Diseases

Keywords: Esophageal Atresia, post-traumatic reactions, Diagnosis

Principal Investigator: Véronique Debarge

Contact: Katialine GROFF (clinical research assistant, katialine.groff@chu-lille.fr)

Partners / centres involved: CHU Amiens-Picardie, CHU Angers, CHU Besançon, CHU Bordeaux, CHU Brest, HFME Lyon, CHU Caen, CHU Clermont-Ferrand, CHI Créteil, CHU Dijon Bourgogne, CHU Grenoble, AP-HP Kremlin-Bicêtre, CH Le Mans, CHU Lille, CHU Limoges, AP-HM La Timone, CHU Montpellier, CHU Nantes, Fondation Lenval Nice, CHR Orléans, AP-HP Armand Trousseau, AP-HP Necker, AP-HP Roberts Debré, CHU Poitiers, CHU Reims, CHU Rennes, CHU Rouen, CHU St Etienne, CHU Strasbourg, CHU Toulouse, CHU Tours, CHU Nancy, CHU Félix Guyon, CHU Martinique

Status: In progress

Intervention

Pathophysiology of dumping syndrome in esophageal atresia

Description: The pathophysiology of dumping syndrome (DS) remains poorly elucidated and suggests the role of the sympathetic system and gastric emptying disorders. Glycemic Holter (HG), coupled with ECG holter - allowing calculation of RR space variability - and gastric emptying scintigraphy, will help to understand the pathophysiology of DS in EA. While the diagnosis of DS is usually based on orally-induced hyperglycemia (OIH), in this project we will be using the blood glucose holter (BGH). This technique, which is widely used to monitor diabetes in children and more recently in newborns and infants of diabetic mothers, continuously records variations in blood glucose levels during and after the child's usual meals for up to 7 days, as it is more physiological and better tolerated.

Workstreams: Esophageal Diseases

Keywords: Esophageal atresia, Dumping syndrome, Glycemic Holter, orally-induced hyperglycemia

Principal Investigator: Madeleine Aumar

Contact: Katialine GROFF (clinical research assistant, katialine.groff@chu-lille.fr)

Partners / centres involved: CHU Lille

Status: In progress

Intervention

Primary pOsterioR TRacheopexy prevents collapse of the trachea in newborns with esophageal AtresIa and Tracheomalacia

Description: The aim of this trial is to evaluate if a primary posterior tracheopexy (PPT) can significantly decrease – or possibly prevent - the collapse of the trachea in newborns with Esophageal Atresia and Tracheomalacia. Additionally, the trial aims to determine whether the observed effect of PPT on tracheal stability is sustained over time.

Workstreams: Esophageal Diseases

Keywords: Primary Posterior Tracheopexy, Tracheomalacia, Esophageal Atresia

Principal Investigator: Maud Lindeboom

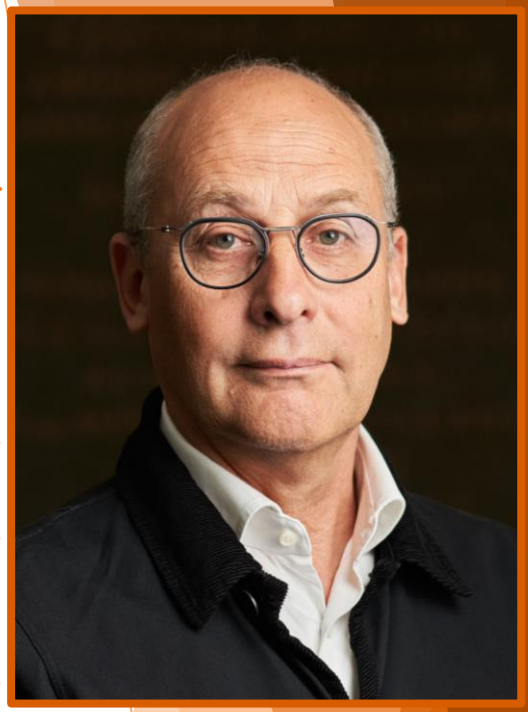
Contact: m.y.a.lindeboom@umcutrecht.nl

Partners / centres involved: UMC Utrecht, John Vlot (Erasmus Medisch Centrum), Colin Butler (Great Ormond Street Hospital), Jan Svensson (Karolinska Institutet)

Status: In progress

Intervention: Clinical Trials

RIC-NEC Trial: Remote ischemic conditioning in premature neonates with necrotizing enterocolitis



Description: International randomized multicenter trial led from Sick Kids in Toronto allocating preterm neonates with NEC to remote ischemic conditioning (RIC) or no RIC.

Workstream: Intestinal Diseases

Keywords: necrotizing enterocolitis, remote ischemic conditioning

Principal Investigator: Agostino Pierro (Toronto), Tomas Wester (local PI)

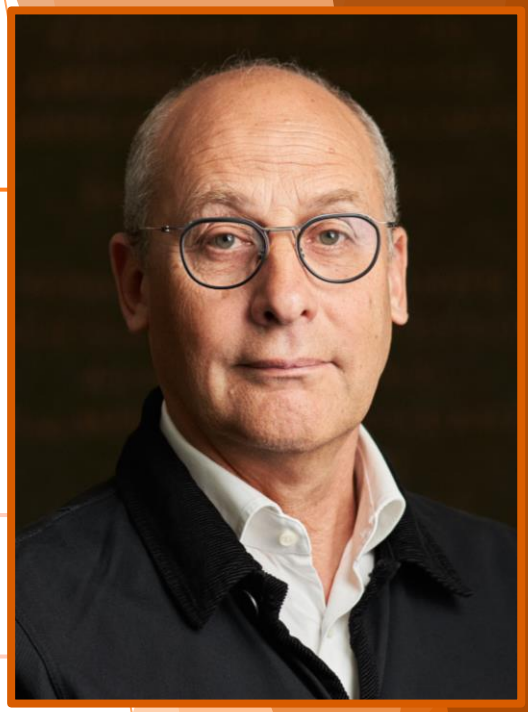
Contact: tomas.wester@regionstockholm.se

Partners / centres involved: Toronto and several other centers in North America and Europe

Status: In progress

Intervention

Impact of centralisation of pediatric surgery in Sweden



Description: The care of EA, CDH and HSCR was centralized to two centers in Sweden in 2018. The current study will compare short-term postoperative outcomes of EA, CDH and HSCR before centralization (2013-2018) and after centralization (2018-2023) in all four pediatric surgery centers in Sweden.

Workstream: Esophageal Diseases; Intestinal Diseases; Malformations of the Diaphragm and AWD;

Keywords: centralization, esophageal atresia, congenital diaphragmatic hernia, Hirschsprung's disease

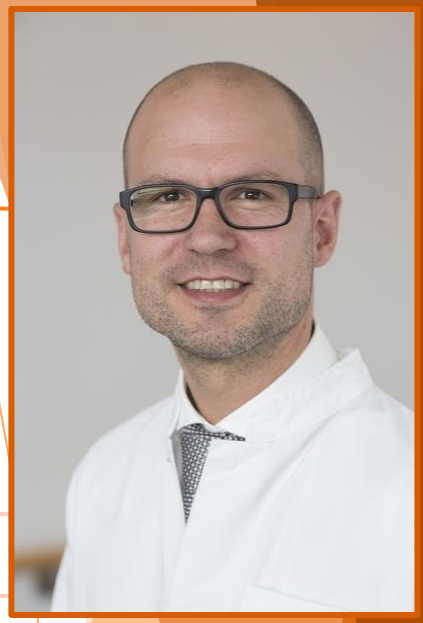
Principal Investigator: Tomas Wester

Contact: tomas.wester@regionstockholm.se

Partners / centres involved: Karolinska University Hospital, Stockholm, Skåne University Hospital, Lund, Queen Silvia Children's Hospital, Göteborg, University Children's Hospital, Uppsala

Status: In progress

Intervention: Clinical Trials



MUC-FIRE

Description: This randomised, multicentre, open-label, controlled study to determine the time between small bowel enterostomy repositioning and full enteral nutrition (NCT03469609) is investigating whether preoperative stool decanting can shorten the time to complete enteral nutrition after enterostomy repositioning.

Workstream: Intestinal Diseases; Nutrition

Keywords: Enterostomies, Necrotizing enterocoltits, focal intestinal perforation, mucous fistula refeeding

Principal Investigators: Prof. Dr. Martin Lacher, Dr. Omid Madadi-Sanjani

Contact: Martin.Lacher@medizin.uni-leipzig.de, madadi-sanjani.omid@mh-hannover.de

Partners / centres involved: Hannover Medical School, University of Vienna, Medical University of Graz, Munich Municipal Hospital, University Children´s Hospital Tuebingen, Universtiy Hospital Frankfurt, University Hospital Dresden, University Medical Center Augsburg, University Children´s Hospital Marburg, Marburg, Marien Hospital Witten, Children´s and Youth Hospital “Auf der Bult” Hannover

Status: In progress

Intervention: Clinical Trials

Study to Evaluate the Efficacy and Safety of Darvadstrocel in the Treatment of Complex Perianal Fistula in Pediatric Subjects with Crohn’s Disease

Description: A Phase 3, Open-Label, Multicenter Study to Evaluate the Efficacy and Safety of Darvadstrocel in the Treatment of Complex Perianal Fistula in Pediatric Subjects with Crohn’s Disease over a Period of 24 Weeks and an Extended Follow-up Period up to 52 Weeks

Workstream: Gastroenterological Diseases

Keywords: Clinical Trial; Crohn's disease; perianal fistula; Children; Darvadstrocel

Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: In progress

Intervention: Clinical Trials

Phase 3B Extension Study to Evaluate the Long-term Safety of Vedolizumab Intravenous in Pediatric Patients With Ulcerative Colitis or Crohn's Disease

Description: A Randomized, Double-Blind, Phase 3 Study to Evaluate the Safety and Efficacy of Vedolizumab Intravenous as Maintenance Therapy in Pediatric Subjects with

Workstream: Gastroenterological Diseases

Keywords: Randomized Clinical trial, Vedolizumab, Ulcerative Colitis, Pediatric

Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: In progress

Intervention: Clinical Trials

Open-label Induction And Maintenance Study Of Oral Tofacitinib In Children With Moderately To Severely Active Ulcerative Colitis

Description: Open-label induction and maintenance study of oral CP-690,550 (tofacitinib) in children with moderately to severely active ulcerative colitis

Workstream: Gastroenterological Diseases

Keywords: Ulcerative colitis; Children; Tofacitinib

Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: In progress

Intervention: Clinical Trials

Study to evaluate the efficacy, safety, pharmacokinetics and pharmacodynamics of oral ozanimod in pediatric subjects with moderately to severely active ulcerative colitis with an inadequate response to conventional therapy

Description: A phase 2/3, multicenter, randomized, double-blind study to evaluate the efficacy, safety, pharmacokinetics and pharmacodynamics of oral ozanimod (rpc1063) in pediatric subjects with moderately to severely active ulcerative colitis with an inadequate response to conventional therapy.

Workstream: Gastroenterological Diseases

Keywords: Phase 2/3 clinical trial; ozanimod; ulcerative colitis

Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: In progress

Intervention: Clinical Trials

Study to evaluate the efficacy, safety, pharmacokinetics and pharmacodynamics of oral ozanimod in pediatric subjects with moderately to severely active Crohn's disease with an inadequate response to conventional therapy

Description: A phase 2/3, multicenter, randomized, double-blind study to evaluate the efficacy, safety, pharmacokinetics and pharmacodynamics of oral ozanimod (RPC1063) in pediatric subjects with moderately to severely active Crohn's disease with an inadequate response to conventional therapy.

Workstream: Gastroenterological Diseases

Keywords: Randomized clinical trial; Crohn's disease; pediatric patients; ozanimod

Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: In progress

Intervention: Clinical Trials

Phase 3, Multicenter, Randomized, Platform Study of p19 Inhibition of the IL-23 Pathway to Establish Efficacy in Pediatric Crohn’s Disease (MACARONI-23)

Description: A Phase 3, Multicenter, Randomized, Platform Study of p19 Inhibition of the IL-23 Pathway to Establish Efficacy in Pediatric Crohn’s Disease (MACARONI-23)

Workstream: Gastroenterological Diseases

Keywords: Clinical Trial, Crohn's disease, p19inhibition

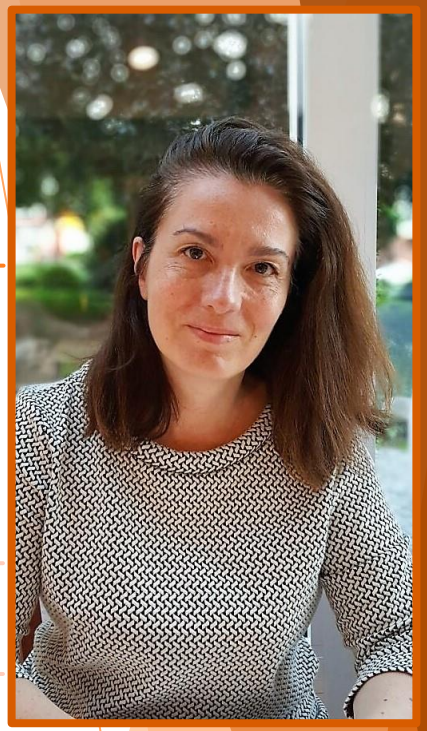
Principal Investigator: Javier Martín de Carpi

Contact: javier.martinc@sjd.es

Partners / centres involved: SJD Barcelona Children's Hospital

Status: Preparation phase/writing protocol

Intervention: Clinical Trials



DOLPHINS-2

Clinical Trials Register

MaRDi website

Description: A Multicenter, Proof-of-concept, Phase 2 Study to Evaluate the Safety, Tolerability, Pharmacokinetics, and Pharmacodynamics of HM15912 in Adult Subjects with Short Bowel Syndrome-associated Intestinal Failure

Workstreams: Intestinal Failure; Nutrition;

Keywords: short bowel syndrome, intestinal failure, parenteral support

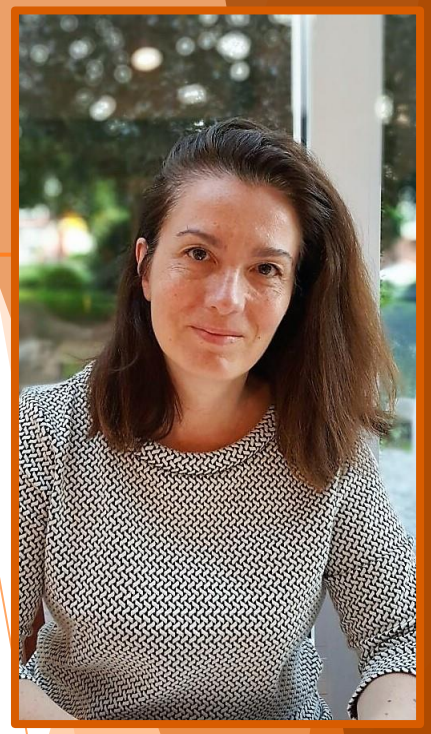
Principal Investigator: Francisca Joly

Contact: francisca.joly@aphp.fr

Partners / centres involved: Intestinal Failure Centres Worldwide

Status: In progress

Intervention: Clinical Trials



Apraglutide Open-Label Phase 3 Extension Trial (VECTIVBIO 012)

MaRDi website

Description: This is an open-label extension trial to evaluate the long-term safety of apraglutide in short bowel syndrome. The Phase 3 placebo-controlled trial TA799-007 evaluated the clinical efficacy (reduction in PS volume) of weekly SC apraglutide compared with placebo in SBS-IF subjects with either CIC or stoma. The aim of this open-label extension trial is to collect further safety, tolerability, efficacy, durability, and clinical outcomes of apraglutide for up to an additional 104 weeks.

Workstreams: Intestinal Failure; Nutrition;

Keywords: short bowel syndrome, intestinal failure, parenteral support, parenteral nutrition

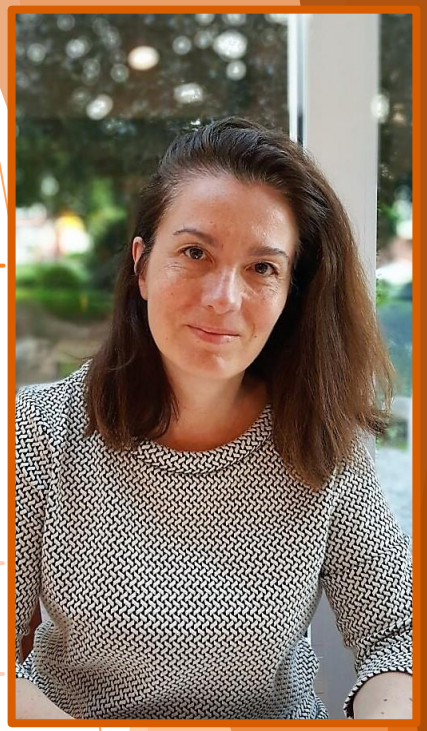
Principal Investigator: Francisca Joly

Contact: francisca.joly@aphp.fr

Partners / centres involved: Intestinal Failure Centres Worldwide

Status: In progress

Intervention: Clinical Trials



EASE SBS 1

ClinicalTrials.gov

MaRDi website

Description: A phase 3, international, multicenter, randomized, double-blind, placebo-controlled trial to evaluate the efficacy and safety of Glepaglutide in patients with short bowel syndrome (SBS) with intestinal failure.

Workstreams: Intestinal Failure; Nutrition;

Keywords: short bowel syndrome, intestinal failure, parenteral support, parenteral nutrition, GLP2 analog

Principal Investigator: Francisca Joly

Contact: francisca.joly@aphp.fr

Partners / centres involved: Intestinal Failure Centres Worldwide

Status: In progress

Intervention: Clinical Trials

EASE SBS 2

ClinicalTrials.gov

MaRDi website

Description: EASE SBS 2: Efficacy And Safety Evaluation of glepaglutide in treatment of SBS. Extension phase 3 randomized double blinded trial, evaluating safety and efficacity of long term treatment with glepaglutide in patients with short bowel syndrome and intestinal failure.

Workstreams: Intestinal Failure; Nutrition;

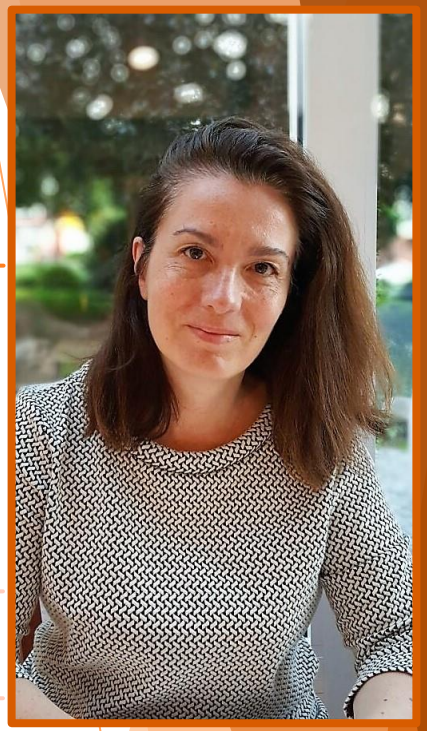
Keywords: short bowel syndrome, intestinal failure, parenteral support, parenteral nutrition, GLP2 analog

Principal Investigator: Francisca Joly

Contact: francisca.joly@aphp.fr

Partners / centres involved: Intestinal Failure Centres Worldwide

Status: In progress



Intervention

Intact Cord Resuscitation in Congenital Diaphragmatic Hernia (CHIC)

Description: Several studies have already demonstrated the benefits of late cord clamping at birth on the biological and physiological adaptation of newborns to life. Earlier work also suggests a possible benefit of this procedure for babies with CDH.

This multicenter randomized clinical trial aims to investigate the efficacy of intact cord resuscitation versus immediate cord clamping on cardiorespiratory adaptation at birth in term neonates with isolated CDH.

Workstreams: Malformations of the Diaphragm

Keywords: Congenital diaphragmatic hernia, resuscitation, delayed cord clamping, newborn

Principal Investigator: Laurent Storme

Contact: Sebastien Mur (sebastien.mur@chu-lille.fr)

Partners / centres involved: 14 participating French centers

Status: In progress

Impact: Long-term follow-up

Long term complications on bowel lengthening procedures

Description: The aim of this study is to generate on overview of long term complications after bowel lengthening procedure.

Workstream: Intestinal failure

Keywords: Short bowel syndrome, bowel lengthening procedures, complications, long term

Principal Investigator: Roel Bakx, Amsterdam UMC

Contact: r.bakx@amsterdamumc.nl

Partners / centres involved: Amsterdam UMC, Erasmus MC

Status: Preparation phase/writing protocol

Impact: Quality of Life

Cross-sectional and prospective study of chronic pain in young people (UNIDOL)

Description: UNIDOL is a study on the training on pain received by health professionals at university level. Having this information is essential in order to identify any possible areas of deficit, if there are any. It can also allow to evaluate possible alternatives that could help to improve in this area.

Workstream: Quality of Life

Keywords: Pain, Children

Principal Investigator: Francisco Reinoso Barbero

Contact: Leopoldo Martínez (leopoldo-martinez@salud.madrid.org)

Partners / centres involved: Hospital La paz

Status: In progress

Impact: Quality of Life

Non-invasive assessment of perioperative atelectasis in children: the role of Air Test in a prospective multicenter double-blind clinical trial

Description: Non-invasive assessment of perioperative atelectasis in children: the role of Air Test in a prospective multicenter double-blind clinical trial.

Workstream: Quality of Life

Keywords: Atelectasis, Non-invasive ventilation, children

Principal Investigator: González Pizarro

Contact: Francisco Reinoso Barbero (francisco.reinoso@salud.madrid.org)

Partners / centres involved: Hospital La paz

Status: In progress

Impact: Quality of Life

TransEAsome Long Term Outcome of Easophageal Atresia : Transmics Profiles in Adolescence

Description: The aim of the project is to create a nested prospective cohort of adolescents born with EA including esophageal and blood samples biobanking in order to assess the long-term health outcome of this rare disease and multi-omics profiles.

Workstreams: Esophageal Diseases; Quality of Life

Keywords: Esophageal atresia, adolescence, gastro-esophageal reflux,dysphagia, esophageal cancer, eosinophilic esophagitis, outcome, epigenetic, proteogenomic metabolomics, quality of life

Principal Investigator: Pr Frédéric Gottrand

Contact: transeasome@chu-lille.fr

Partners / centres involved: PRISM (INSERM Nord Ouest), Go@L (Université de Lille), BiLille (Université de Lille), PedStart (INSERM Grand Ouest) and all French university hospitals

Status: Preparation phase/writing protocol

Impact: Quality of Life

Translation, validation and implementation of the SQEA questionnaire

Description: The SQEA questionnaire is a condition-specific instrument, measuring the self-perceived health-related quality of life of adults born with esophageal atresia. It was designed together with patients born with EA, and has proven to have satisfactory feasibility, reliability, and validity in the Dutch language, and to cover all domains of the International Classification of Functioning, Disability and Health framework. Ideally, the SQEA will be implemented as a standard of care during follow-up, both in the Netherlands and internationally. To that end, the SQEA questionnaire first needs to be translated and validated in different languages.

Workstream: Esophageal Diseases; Quality of Life

Keywords: SQEA questionnaire, translation, feasibility, validity, reliability

Principal Investigator: Chantal A. ten Kate

Contact: c.tenkate@erasmusmc.nl

Partners / centres involved: ERNICA centers will soon be invited to participate in this international project.

Status: Consortium building

Impact: Quality of Life

Psychometric validation of the Fench version of EA-QOL questionnaires

Description: The Esophageal Atresia Quality-Of-Life© questionnaires (Dellenmark-Blom et al., 2018) were developed for children aged 2 to 17 with EA/TEF and for their parents. They assess specific domains frequently impacted following esophageal atresia surgery. They have been translated and validated in several languages but not in French. The purpose of this study is to test the validity and the relability of the French version of the EA-QOL© questionnaires.

Workstream: Esophageal Diseases; Quality of Life

Keywords: Quality of life, health-related of quality of life, questionnaire, validation, esophageal-atresia

Principal Investigator: Nathalie Coulon, University of Lille, Laboratory PSITEC

Contact: nathalie.coulon@univ-lille.fr

Partners / centres involved: French Reference Center for Rare Esophageal Disorders (CRACMO)-CHU Lille; Queen Silvia Children's Hospital, University of Gothenburg; Association AFAO

Status: In progress

Impact

Standardized Transitional Pathways In Pediatric Achalasia: A Systematic Review And Expert Proposal

Description: This study aims to systematically review the existing literature on transition of care programs for pediatric patients undergoing achalasia surgical or endoscopic treatment.

As esohpageal dysplasia and carcinoma have been addressed as long-term risks in achalasia patients, our ultimate goal is to combine available evidence and our center's expertise in defining a transitional program proposal for pediatric patients after achalasia treatment.

Workstream: Esophageal Diseases;

Keywords: achalasia, endoscopy, Heller myotomy, transition of care, long-term follow-up

Principal Investigator: Piergiorgio Gamba, Padua University Hospital

Contact: Rebecca Pulvirenti (rebeccapulvirenti@gmail.com)

Partners / centres involved: Department of Surgical, Oncological and Gastroenterological Sciences, Padua University Hospital

Status: In progress

Impact:

National Esophageal Atresia Registry

Description: The current project aims to set up a national registry (metropolitan France + Dom Tom) to measure the prevalence of esophageal atresia among live births, their phenotypic characteristics, the circumstances of their diagnosis, and their initial short-term outcome during the first year of life, when the vast majority of deaths and complications in this condition occur. Completion of this registry over will enable us to assess the influence of advances in management (antenatal diagnosis, advances in surgery and neonatal resuscitation) on the incidence and prognosis of the condition.

Workstreams: Esophageal Diseases

Keywords: esophageal atresia, comorbidity, prevalence

Principal Investigator: Pr Frédéric Gottrand

Contact: Katialine Groff (clinical research assistant, katialine.groff@chu-lille.fr)

Partners / centres involved: Numerous centres in France

Status: In progress

Impact

Diagnosis and treatment of congenital esophageal stenosis: the European experience

Description: This is a multi-center observational study, aiming to retrospectively collect data on the diagnostic-therapeutic process of congenital esophageal stenosis. Aspects to be evaluated involve the symptoms' onset and characteristics, the diagnostic tests used, the type of treatment performed, and the patients' condition at follow-up. Additional objectives are to define the effectiveness of the different therapeutic approaches and identify predictive factors enabling a tailored treatment. The ultimate goal is using the retrieved data as a basis for a diagnostic-therapeutic consensus statement.

Workstreams: Esophageal Diseases;

Keywords: congenital esophageal stenosis, endoscopic dilatation, surgery, outcome

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Rebecca Pulvirenti (rebeccapulvirenti@gmail.com)

Partners / centres involved: Padua University Hospital, Italy, Erasmus MC, The Netherlands

Status: Pending ethical approval

Impact

Transition of care in pediatric achalasia: an international multicentric study of the ERNICA Study Group

Description: The main objective of the study will be to define the prevalence of the diagnosis of esophageal achalasia in children in ERNICA centers, the treatment performed, and the short-term outcomes and complications. It will be fundamental to describe the age of onset of symptoms, associated comorbidities, and the reasons for choosing a particular treatment.

Workstreams: Esophageal Diseases; Transition

Keywords: Esophageal achalasia, pediatric achalasia, transition of care, rare diseases

Principal Investigator: Renato Salvador; Co-PI: Giovanni Capovilla

Contact: renato.salvador@unipd.it; giovanni.capovilla@aopd.veneto.it

Partners / centres involved: UOC Chirurgia Generale I, Azienda Ospedale Università di Padova; UOC Chirurgia Pediatrica, Azienda Ospedale Università di Padova; other centers have not yet been contacted because of pending ethical approval

Status: Pending ethical approval

Impact

Histological reparative changes in infants with Necrotizing Enterocolitis (NEC)

Description: This study aims to assess the impact of reparative changes of the affected bowel on postoperative recovery and long-term outcomes, including neurodevelopmental assessment beyond 3 years, in infants with NEC.

Workstreams: Intestinal Diseases;

Keywords: NEC, Histology, reparative changes, long-term outcomes

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: Padua University Hospital, Italy

Status: In progress

Impact

Non-invasivE multimodal monitoring tools for NeCrotizing enterocolitis in preterm infants: A piLot MulticEnteR sTudy (NEC-ALERT)

Description: This study aims to provide a prompt and accessible multimodal analysis tool based on artificial intelligence approach to predict the risk and severity of NEC in preterm babies. This approach integrates antenatal findings, clinical data, inflammatory biomarkers, near-infrared spectroscopy, imaging data, and omic-analysis of plasma and faecal sample in premature infants

Workstreams: Intestinal Diseases;

Keywords: NEC, diagnostic tools, omic analysis, Artificial intelligence

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: ERNICA Partners

Status: Consortium building

Impact

Near-Infrared Spectroscopy as a predictive tool for the development and progression of Necrotizing Enterocolitis in preterm neonates: A pilot Study

Description: This study investigates whether parameters from abdominal and cerebral NIRS (tissue oxygen saturation (CrS02, ArS02); oxygenation variability; and fractional-tissue oxygen extraction (FTOE)) might predict NEC onset. Furthermore, the role of ArS02 as predictive indicator for severe NEC is being explored.

Workstreams: Intestinal Diseases;

Keywords: NEC, Near Infrared spectroscopy (NIRS), predictive indicators, NEC severity

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: Padua University Hospital

Status: In progress

Impact

Hirschsprung's associated enterocolitis (HAEC): putative histopathological and clinical predictive factors

Description: This study aims to identify potential predictive factors for post-operative HAEC in Hirschsprung's disease patients, with a particular focus on histopathological findings including the length of segments with altered neuro-enteric system and mucosal inflammatory changes in the resected bowel specimen.

Workstreams: Intestinal Diseases;

Keywords: HAEC, Transition zone, histological inflammation

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: Miriam Duci (ducimiriam@gmail.com)

Partners / centres involved: Padua University Hospital

Status: In progress

Impact

LHR trend as congenital diaphragmatic hernia (CDH) outcome predictor

Description: The study aims to evaluate LHR and LHR O/E trend during prenatal ultrasound evaluations and compare the ratio course with postnatal outcomes. Ultimate goal is to assess their reliability at different prenatal stages as good outcome predictors.

Workstreams: Malformations of the Diaphragm and AWD

Keywords: Congenital diaphragmatic hernia, prenatal ultrasound, LHR, outcome

Principal Investigator: Francesco Fascetti-Leon, Padua University Hospital

Contact: francesco.fascettileon@unipd.it

Partners / centres involved: Padua University Hospital

Status: Preparation phase/writing protocol

Impact

Current outcomes of total colonic aganglionosis in Europe

Description: This retrospective observational study aims to address the current number of patients followed-up, management strategies and outcomes of TCA in European Reference Network (ERNICA) centers. This research brings new information on outcomes of TCA and allows to compare outcomes and treatment results between centers. The results may be used to improve care of TCA patients. The study is currently ongoing, participating center are applying local ethical approval and data collections is aimed to finish during 2024.

Workstreams: Intestinal Diseases

Keywords: Hirschsprung, total colon aganglionosis, long-term outcomes

Principal Investigator: Mikko Pakarinen

Contact: Annika Mutanen (first investigator, annika.mutanen@hus.fi)

Partners / centres involved: Multiple centres in Europe have expressed interest in participating

Status: Pending ethical approval

Impact

A multicentric cross-sectional observational study on intestinal inflammation in total colonic aganglionosis

Description: Inflammatory Bowel Diseases can occur in association with Hirschsprung disease. Based on this consideration, we will perform a prospective study in order to provide prevalences and demographics on inflammatory bowel disease-like lesions in children with total tolonic aganglionosis treated in ERNICA expertise centers. This study is ongoing, protocol and application of ethical approval will be finalized during 2024.

Workstreams: Intestinal Diseases

Keywords: Total colon aganglionosis, Hirschsprung, bowel inflammation, inflammatory bowel disease

Principal Investigator: Mikko Pakarinen

Contact: Annika Mutanen (first investigator, annika.mutanen@hus.fi)

Partners / centres involved: Participating center list is not ready

Status: Preparation phase/writing protocol

Impact

Pediatric intestinal pseudo-obstruction an international survey on diagnostic and management strategies in ERNICA IF Teams

Description: The aim of this study was to outline the current diagnostic and management strategies in intestinal failure (IF) teams of the European Reference Network for rare Inherited and Congenital Anomalies (ERNICA) and to compare these practices to the latest PIPO international guidelines. An online survey on institutional diagnostic and management strategies of PIPO was conducted among the ERNICA IF teams. This study showed, that numbers of PIPO patients are low and management strategies are diverse among ERNICA IF teams. To improve PIPO patient care, regional reference centers with specialized multidisciplinary IF teams and continuous collaboration across centers are needed.

Workstreams: Intestinal Failure

Keywords: Intestinal failure, multidisciplinary, pediatric intestinal pseudo-obstruction, parenteral nutrition

Principal Investigator: Annika Mutanen

Contact: annika.mutanen@hus.fi

Partners / centres involved: Department of Pediatric Gastroenterology, Hepatology and Nutrition, Emma Children's Hospital, Amsterdam UMC; Department of Pediatric Surgery, University Hospital Mannheim, University of Heidelberg; 11 ERNICA centers.

Status: Completed/published

Impact

Outcomes of pediatric intestinal pseudo-obstruction in ERNICA

Description: Pediatric patients with pseudo-obstruction (PIPO) experience significant morbidity and mortality while lack of knowledge in all aspects of PIPO from diagnostics to management brings significant challenges in clinical care. To improve care of patients with PIPO, multicenter studies are warranted. This retrospective multicenter European Reference Network for Rare inherited Congenital Anomalies (ERNICA) collaborative study will provide unique data on pediatric PIPO incidence, diagnostic modalities, and management and further benchmarking of outcomes in ERNICA centers. This is a retrospective multicenter study in the ERNICA centers including all patients diagnosed with PIPO and currently under follow up in ERNICA centers.

Workstreams: Intestinal Failure

Keywords: Intestinal failure, pediatric intestinal pseudo obstruction, parenteral nutrition

Principal Investigator: Annika Mutanen

Contact: annika.mutanen@hus.fi

Partners / centres involved: ERNICA IF centers, list of participating centers still not ready

Status: Consortium building

Impact

A nordic multicenter study on contemporary outcomes of pediatric short bowel syndrome in 208 patients

Description: Despite advances in the management of short bowel syndrome related intestinal failure (SBS-IF), large-scale contemporary pediatric studies are scarce. The aim of this multicenter study was to assess key outcomes and clinical prognostic factors in a recent Nordic pediatric SBS-IF population. Patients with SBS-IF treated during 2010-2019, whose parenteral support (PS) started at age <1 year and continued >60 consecutive days were included and retrospectively reviewed. All six participating centers followed multidisciplinary SBS-IF management. In this study, we showed that although with current multidisciplinary management, prognosis of pediatric SBS is encouraging, septic complications and IFALD still associated with the remaining low mortality rate.

Workstreams: Intestinal Failure

Keywords: intestinal failure, short bowel syndrome, parenteral nutrition

Principal Investigator: Annika Mutanen

Contact: annika.mutanen@hus.fi

Partners / centres involved: Department of Pediatric Gastroenterology, Hepatology and Nutrition, Emma Children's Hospital, Amsterdam UMC; Department of Pediatric Surgery, University Hospital Mannheim, University of Heidelberg; 11 ERNICA centers.

Status: Completed/published

Impact

Pediatric Short Bowel Syndrome in Europe: can we do better?

Description: The aim of our study is to prospectively collect data on the parenteral nutrition weaning strategies adopted by the ERNICA centres, compare them with the current literature, and explore the specific areas where treatments are based on local practices rather than evidence-based approaches. This could help broaden the discussion, share protocols, and promote studies to provide further insights into these topics, ultimately improving the care of pediatric patients with short bowel syndrome.

Workstreams: Intestinal Failure

Keywords: Intestinal failure, short bowel syndrome, parenteral nutrition, weaning strategies

Principal Investigator: Giovanna Verlato, Padua University Hospital

Contact: giovanna.verlato@aopd.veneto.it

Partners / centres involved: Padua University Hospital, Italy; Universitätsklinik Mannheim: Uniklinik Mannheim, Germany; Motol University Hospital, Czech Republic; Akademiska sjukhuset i Uppsala, Sweden; Amsterdam UMC – Emma children's hospital, The Netherlands; Centre Hospitalier Universitaire de Lille, France; Hopital Robert Debré, France; Great Ormond Street Hospital, United Kingdom

Status: Consortium building

Impact

Engagement in Physical Activities and Sports in Adolescents With Diaphragmatic Hernia (CHASAM)

Description: The aim is to understand how a population of adolescents – who have undergone diaphragm surgery in the first month of life (i.e. subjects with a rare disease with an impact such as CDH) - engages in physical and sporting activities, and what factors may hinder and facilitate these practices. - engage in physical activities and sports, and what factors may hinder or facilitate these practices.

Based on self-questionnaires and semi-structured interviews, this is qualitative research in the humanities and social sciences.

Workstreams: Abdominal wall defects

Keywords: Diaphragmatic Hernia, physical activity, sports, childhood, adolescence, sociological psychological environment

Principal Investigator: Sebastien Mur

Contact: sebastien.mur@chu-lille.fr

Partners / centres involved: a French association of CDH patients, Lille Medical Center

Status: In progress

Impact

BELAPS National Prospective Multicentre Registry on Hirschsprung's Disease

Description: In order to increase efforts to deliver optimal surgical care to pediatric patients in Belgium, the Belgian Association of Pediatric Surgery decided to start a voluntary prospective registry on Hirschsprung's disease, including pre-, intra- and short- and long-term postoperative data on this patient population. Participating surgeons will be included in the analysis, presentation and discussion of the results. All pediatric surgeons in Belgium are invited to join the registry and document consecutively treated patients

Workstreams: Intestinal Diseases

Keywords: Hirschsprung's Disease, registry, surgery, outcome

Principal Investigator: Marc Miserez

Contact: Colette Barlé (kim.vanderlinden@uzbrussel.be)

Partners / centres involved: AZ St Jan Brugge, CHR La Citadelle Liège, CHR Namur, CHU Charleroi, CHU St Pierre Brussels, GHdC Charleroi, GZA Antwerpen, HUDERF Brussels, UZ Antwerpen, UZ Brussel, UZ Gent, UZ Leuven, ZNA Paola Antwerpen

Status: In progress

Comorbidities

BIOPTIC I and II Study

Description: Biomarker for Outcome Prediction in Neonates with Congenital Diaphragmatic Hernia Study. This studies was initiated in 2014 and prospectively enrolls CDH neonates. Blood biomarkers are measured from blood taken at predefined timepoints. Biomarker levels are correlated with respiratory and cardiovascular outcome, such as pulmonary hypertension, need for ECMO, Chronic lung diseases, cardiac dysfunction, mortality.

Workstreams: Malformations of the Diaphragm and AWD

Keywords: Congenital diaphragmatic hernia, ECMO, biomarker, pulmonary hypertension

Principal Investigator: Florian Kipfmueller

Contact: florian.kipfmueller@ukbonn.de

Partners / centres involved: Bonn

Status: In progress

Comorbidities

Malignant Hyperthermia Risk Population: Ambispective Cohort

Description: This ambispective cohort design study of patients with hypermetabolic skeletal muscle response will assess MH susceptibility in these patients. of skeletal muscle will assess the MH susceptibility of these patients by performing IVCT by performing IVCT, genetic study, and skeletal muscle hypermetabolic lymphocyte B cell hypermetabolic B-lymphocyte response (TRHLB). In addition, gene frequencies will be evaluated in the Spanish population that are associated with positive responses

Workstreams: Quality of Life

Keywords: Malignant Hyperthermia, Children, Risk

Principal Investigator: Pascual Sanabria Carretero

Contact: pascual.sanabra@salud.madrid.org

Partners / centres involved: Hospital Infantil La Paz

Status: In progress