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UK cohort study of congenital anomalies potentially requiring surgery in children (Surgical-PEARL study)

Submission date
26/04/2022

Recruitment status
Recruiting

Registration date
28/04/2022

Overall study status
Ongoing

Last Edited
19/03/2025

Condition category
Genetic Diseases

[X] Prospectively registered
[X] Protocol
[ ] Statistical analysis plan
[ ] Results
[ ] Individual participant data
[ ] Record updated in last year

Plain English summary of protocol

Background and study aims
Around 1 in 60 babies are born with congenital anomalies (a structural condition) that require surgery to survive but we don't really know why congenital anomalies occur. We are setting up a study to collect information about the health and quality of life of children with congenital anomalies that may require surgery. We are asking parents whose unborn baby or child has been diagnosed with a structural condition to take part in this study. We hope to look at risk factors by collecting samples from children and their parents. By monitoring children's blood, urine and stool during their care in the hospital we may be able to work out better treatments for them.

Who can participate?
Children ages 0-5 years old, diagnosed with a congenital anomaly that might require surgery to correct.
The study will also invite the biological parents to collect data and samples.

What does the study involve?
Participants can be approached before or after birth. Once the congenital anomaly has been diagnosed, the parents will be approached by the study team and given a participant information leaflet. If the parents consent to their child taking part in the study, details about their delivery and surgery will be collected by the research team. If the parents also consent to samples, these will be collected after delivery (placenta, umbilical cord blood), where applicable, and during their surgical admission (blood, urine, stool and waste tissue). Parents will also be asked to complete a questionnaire 12, 24, 36 and 48 months after delivery. Questionnaires will be completed online and include questions about risk factors, their quality of life and if they have been admitted to the hospital or seen a GP since their surgery.
Biological parents will also be approached and given a participant information leaflet. If they are happy to take part in the study, data on potential risk factors will be collected from medical notes and/or questionnaires, and blood and urine samples will be collected.

What are the possible benefits and risks of participating?
There are no direct benefits of participating in the study, but we hope that the results from this

study may benefit future care and outcomes for patients and the NHS. Samples will be taken from the children from lines that are already in place, so no extra needles will be needed. There is very minimal risk associated with taking blood samples from the biological parents.

Where is the study run from?
The study is run from the University of Bristol and will take place in hospitals around the UK.

When is the study starting and how long is it expected to run for?
April 2021 to January 2032

Who is funding the study?
The study is currently funded by the Oyster Foundation Charity, the British Heart Foundation and the Bristol Biomedical Research Centre (UK).

Who is the main contact?
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Additional identifiers

Integrated Research Application System (IRAS)
302251

Central Portfolio Management System (CPMS)

51673

Protocol serial number
2021-4859

Study information

Scientific Title
Surgical Paediatric congEnital Anomalies Registry with Long term follow-up (Surgical-PEARL study)

Acronym
Surgical-PEARL

Study objectives
Congenital anomalies or rare diseases affect 1 in 64 live births. Those with structural anomalies often require surgical interventions to survive. The potential causes of congenital anomalies potentially requiring surgical interventions (CAPRS) and the factors influencing their prognosis are not well understood. It is likely that they occur as a result of interacting molecular, genetic and environmental risk factors. This study aims to create a data registry tracking CAPRS from diagnosis through to long term outcomes including routinely collected clinical data on interventions and survival, as well as collecting biological samples from patients and their biological parents.

Ethics approval required
Old ethics approval format

Ethics approval(s)
Approved 15/03/2022, South East Scotland Research Ethics Committee 01 (2nd Floor, Waverley Gate, 2-4 Waterloo Place, Edinburgh, EH1 3EG, UK: +44 131 536 9000; Sandra.Wyllie@nhslothian.scot.nhs.uk), ref: 22/SS/0004

Primary study design
Observational

Study design
Multi-centre prospective cohort study

Study type(s)
Other

Health condition(s) or problem(s) studied
Congenital anomalies potentially requiring surgical intervention

Interventions
Participants can be recruited antenatally or postnatally. Once the congenital anomaly has been diagnosed, the parents will be approached and given a Participant Information Leaflet. When a parent consents to their child taking part, data from their delivery and surgical admission will be collected. Where parents have also consented to samples, placenta and umbilical cord samples will be collected after delivery (if patient has been approached antenatally) and blood, urine,

stool and waste tissue will be collected during their surgical admission. These samples are stored for future analyses to identify potential markers of post-operative complications and recovery. Parents will also be asked to complete a questionnaire on behalf of the children 12, 24, 36 and 48 months after delivery or surgery. Questionnaires will be completed online and include questions about risk factors, quality of life (PEDSQL) and resource use.
Participants also consent for details to be collected from NHS Digital and other clinical databases. Patient involvement in the study finishes once all questionnaires have been submitted.
Biological parents will also be invited to take part in the study to provide data and samples. If the parents consent, blood and urine samples will be taken and they will be asked to complete a questionnaire on potential risk factors during pregnancy.
Where consent has been obtained, pseudonymised samples may be made available to future Research Ethics Committee (REC) approved studies.

Intervention Type
Other

Primary outcome(s)
1. Aggregated routinely collected clinical data from review of medical notes: operation details, PICU/NICU, HeartSuite, Hospital episode statistics (HES), HER, Magnetic Resonance Imaging (MRI), Computerised tomography (CT), Echocardiogram (ECHO), ultrasounds. Assessed at baseline and then yearly for 5 years.
2. Phenotypic and genetic analysis of biomaterials that would normally be discarded during surgery as well as blood, urine and stool samples from patients and their biological parents. Biological mothers will provide baseline blood and urine samples, and optional amniotic fluid sample (if patient is recruited antenatally). Biological fathers will provide baseline blood and urine samples. Patients will provide pre-operative blood, urine, stool and tissue when available, on arrival to PICU: blood and urine samples, 24 hours post-surgery: blood and urine samples.

Key secondary outcome(s)
1. Demographics collected via questionnaires at baseline
2. Maternal and paternal demographics and information on potentially modifiable risk factors associated with CAPRS, assessed using questionnaires developed specifically for this study and accessing relevant pregnancy medical records at baseline.
3. Short, medium and long-term clinical outcomes in patients born with CAPRS, monitored by reviewing the patient's medical notes yearly for 5 years.
4. NHS resource use, monitored by reviewing the patient's medical notes yearly for 5 years and using questionnaires developed specifically for this study
5. Patient's and parents' quality of Life (QoL) measured using PEDSQL questionnaires 12, 24, 36 and 48 months post-delivery (for patients recruited antenatally) or post-consent (for patients recruited postnatally).
6. Genetic and phenotypical characterisation of patients and their mothers and fathers.
7. MicroRNA analyses and isolation of progenitor cells when sufficient tissue, plasma and serum are available

Completion date
16/01/2032

Eligibility

Key inclusion criteria

Patients must be:
1. Fetus (2nd and 3rd trimester) or aged between 0 months and 5 years of age
2. Diagnosed with CAPRS

The person giving consent must:
3. Have parental responsibility for the participant

Biological mothers and fathers:
4. Have a biological child enrolled in PEARL
5. Have capacity to consent

Participant type(s)
Patient

Healthy volunteers allowed
No

Age group
Child

Lower age limit
0 Months

Upper age limit
5 Years

Sex
All

Key exclusion criteria
1. Unable to give informed consent and/or assent
2. Main residence is outside the UK

Date of first enrolment
19/05/2022

Date of final enrolment
01/01/2032

Locations

Countries of recruitment
United Kingdom

England

Study participating centre
University Hospitals Bristol and Weston NHS Foundation Trust
Trust Headquarters

Marlborough Street
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BS1 3NU

Sponsor information

Organisation
University of Bristol

ROR
https://ror.org/0524sp257

Funder(s)

Funder type
Charity

Funder Name
Oyster Foundation Charity

Funder Name
British Heart Foundation

Alternative Name(s)
The British Heart Foundation, the_bhf, BHF

Funding Body Type
Private sector organisation

Funding Body Subtype
Trusts, charities, foundations (both public and private)

Location
United Kingdom

Funder Name
NIHR Bristol Biomedical Research Centre

Alternative Name(s)

NIHR Biomedical Research Centre Bristol, National Institute for Health Research Bristol Biomedical Research Centre, NIHR Bristol BRC, Bristol BRC, Bristol Biomedical Research Centre

Funding Body Type
Private sector organisation

Funding Body Subtype
Research institutes and centers

Location
United Kingdom

Results and Publications

Individual participant data (IPD) sharing plan
The datasets generated during and/or analysed during the current study are/will be available upon request from pearl-study@bristol.ac.uk.

IPD sharing plan summary
Available on request

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Protocol article		09/12/2022	06/01/2023	Yes	No