CoRIPS Research Award 100
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To formulate and implement a reporting system for identification of the percentage and types of congenital heart defects undiagnosed by current antenatal ultrasound screening

Awarded £9631

Principal Aim of the Study:
To formulate and implement a reporting system for identification of the percentage and types of congenital heart defects undiagnosed by current antenatal ultrasound screening.

Primary research question;
What is the annual incidence and aetiology of postnatally presenting congenital heart defects in South Wales?.

Secondary research questions:
a. Can any sonographer training needs be identified by retrospective analysis of postnatally presenting cases?
b. Can retrospective analysis of antenatal images suggest additional techniques that could be incorporated into protocols to improve antenatal detection of those CHD cases currently presenting postnatally?

Outcomes:
A structured data management system for postnatally presenting cases of CHD, leading to improved service delivery and patient outcomes via informed identification of potential extended imaging protocols and sonographer training needs.

Review of Literature and identification of current gap in knowledge:
In the United Kingdom, ultrasound scanning in the second trimester of pregnancy to detect fetal structural abnormalities has been undertaken since the 1980's and has been useful to allow clinicians to develop pathways and care management that optimise pregnancy outcome (1).

Congenital heart disease (CHD) refers to any cardiac defect that is present from birth (2). As the majority of cases occur in low risk pregnancies, these will only be detected by examination of the fetal heart during the routine anomaly examination (1), however there is extensive literature on both the variation of detection of congenital heart disease (CDH) and the prognosis of antenatally and postnatally detected cases (3-8).

There are significant physiological changes to the fetal circulation which occur in the early post natal period which can greatly affect the prognosis of a child with CHD (9). Neonatal death within the first 24hrs of birth has been previously reported in 10% of neonates with undiagnosed CHD, while up to a half of all infantile deaths occur due to undiagnosed CHD (9,10). Antenatal detection of a fetus affected by CHD enables more specialised pregnancy management, with arrangement of delivery in a unit with the both facilities necessary to support the infant and the ability to execute prompt and correct treatment for the condition, which is often paramount to the survival (11). It can also offer the parents the choice of whether the pregnancy should continue.

Increasing the number of views imaged of the fetal heart, considerably increases the ability to antenatally diagnose CHD, especially when the cardiac outflow tracts are visualised in addition to the 4 chamber view of the heart (6). Therefore the National Institute for Clinical Excellence recommend mandatory examination and reporting of the fetal cardiac outflow tracts as part of the routine anomaly scan (12). The 2010 Fetal Anomaly Screening Programme (FASP) guidance (1) therefore includes a fetal cardiac protocol where examination of the fetal cardiac outflow tracts as mandatory from April 2011. This was reflected in the corresponding 2010 revision of Antenatal Screening Wales (ASW) standards which stated that reporting of Cardiac outflow tracts must be incorporated into the fetal anomaly scan in Wales by September 2010 (13). In response to a request from sonographers for extra training to undertake these protocols, all sonographers had specific training in the necessary techniques as part of the All Wales Fetal Cardiac Training programme, undertaken to support implementation of the improved standards.

Whilst other factors such as examination time and equipment type have been identified as explanations for increased detection (3), there is evidence to suggest that differing scanning skills can affect the rate of detection (14). Therefore, the requirement for further training has been identified as key to diagnosis, as those centres with active training programmes are reported to have increased antenatal detection rates of CHD (9). This is supported by international data on the incidence of specific abnormalities, prior to implementation of the new standards, from 2006-2010, which indicated Wales identified 93% of hypoplastic left heart syndrome, identifiable on the 4 chamber view, but only 46% of cases of transposition, the diagnosis of which requires assessment of the outflow tracts (15). As these are the only types of CHD analysed by EUROCAT, it is impossible to suggest the detection rate for other defects.

The National standards report that assessment of both the four chamber and views of the outflow tracts should exclude 50% of abnormalities (1), and since the introduction of the All Wales training programme, National Institute for Cardiovascular Outcomes Research (NICOR) has reported a Welsh national increase in the detection rate of CHD up to 2011, from 20 - 25% to 52% in cases requiring surgical intervention (16). This increase is also anecdotally supported by Congenital Anomaly Register and Information Service for Wales (CARIS) which collects data and reports on antenatal detection rates of all fetal abnormalities including CHD (17). However, due to the varied reporting systems available and the varied geographic nature of tertiary fetal and paediatric cardiac services in Wales the incidence or nature of undiagnosed conditions remains elusive, hence the need for this study to be undertaken.

Although NICOR data suggests an overall increase in antenatal detection of CDH in Wales, it still identifies a wide variation in detection throughout Wales, from 5 to > 50% suggesting that many cases remain undiagnosed (15). As neither CARIS, NICOR or EUROCAT data specifically identify the aetiology of undiagnosed cases, retrospective analysis of the fetal anomaly examination of those children, undertaken with the benefit of knowledge of the confirmed condition, may give valuable insight into specific areas of fetal diagnosis during the anomaly examination that may be improved.

It is anticipated that this study could inform any need for further sonographer training, including consolidation and refining of current techniques or incorporating further techniques to aid identification of specific defects that are currently undiagnosed, so that more than 50% of cases are identified in utero in all geographical areas. However, the gathering of such data is heavily reliant on a robust reporting system to indicate when a postnatal diagnosis of CHD is made.
Therefore, this study aims to formulate and implement a reporting system for identification of the percentage and types of congenital heart defects undiagnosed by current antenatal ultrasound screening in the Cardiff referral area, as a pilot to a future all-Wales study.

Methodology

Based on local audit of the number of cases presenting postnatally in any 12 month period over the last 3 years, the anticipated whole populationfor this quantitative study will be approximately 50 cases. All babies 12 months of age or under, presenting postnatally to Cardiff Cardiac services with a CHD (excluding patent ductus arteriosus) that had an anomaly ultrasound examination in a Welsh hospital will be included in the study. Exclusion criteria will be any child over 12 months of age. Over 12 months, the nature of the CHD is unlikely to be severe enough to be detected on antenatal ultrasound examination of the fetus (16).

Maternal consent will be requested in the clinical environment by the clinician leading the consultation, where an explanation of the study aims will be given. A patient information sheet and consent form will be given to the participant for them to complete and return to the department in their own time. This will give permission for paediatric cardiologists in the study centre to report the child's presentation to the researcher via paper copy and for the researchers to access the anomaly scan images and report. This paper based process does not require any specialist technology or secure information network and is appropriate due to the close geographical proximity of the data collection and analysis sites. A similar system has previously been used effectively by Congenital Anomaly Register and Information Service for Wales (CARIS) for the purpose of data collection on the antenatal presentation and diagnosis of fetal abnormalities.

Each proforma to interrogate the data will include the following information provided by the clinician:

- Maternal details – name, date of birth, current address and address at the time of anomaly scan, NHS number, hospital number.
- Child details – name, date of birth, NHS number and postnatal diagnosis and type of

Child details – name, date of birth, NHS number and postnatal diagnosis and type of CHD

- Anomaly examination details – Date of examination, hospital where the examination was undertaken.

Information on the equipment used, number of fetuses, co-morbidities, any identified limitations of the examination will be obtained from the archived examination images and the formal, standardised All Wales RadIS II examination report.

Any antenatal images not available at the time of referral will be requested in electronic format via Patient Archive Recording System (PACS) from the respective radiology departments for all participating subjects. If electronic images are not available, anonymised, coded images will be transferred, in DICOM format, without loss of resolution, to CD to be collected by the researcher or for analysis at the research base.

For each identified child, images obtained at the time of the anomaly scan will be analysed to assess:

- Appropriateness of imaging planes used in relation to previous All Wales guidance and training protocols,
- Positional quality of still images,
- Whether Doppler techniques were used
- Whether M-Mode trace was undertaken
- Assessment for any indicators of congenital anomaly

This data will enable secondary research questions a and b to be investigated.

Anomaly scan images, will be analysed by both a sonographer with a specific interest in fetal echocardiography (principal researcher) and a fetal cardiologist (gold standard) to ensure accuracy and reliability. They will be viewed on a high resolution image reporting monitor in the cardiology department and compared to RCOG and NICE guidance for appropriate imaging of the fetal heart.

Data analysis:
Descriptive statistical analysis of percentage, mean, mode and standard deviation of:

- Assessment of the number of postnatally presenting cases.
- Type of condition
- Geographical location of anomaly examination.
- The number and type of limitations of the anomaly examination

Images will be analysed for the following:

- Were standardised images taken in response to national guidelines?
- Based on the images available, subjective likert scale assessment of the likelihood of antenatal diagnosis without the availability of retrospective diagnostic information.
- With the benefit of knowledge regarding the positive diagnosis of CHD, were there any indications of the disease visible at the time of the anomaly examination?
- Could further views or techniques have potentially identified the specific type of CHD?
- Are there trends in the location of where anomaly scan examinations of identified cases were undertaken which may indicate potential sonographer training needs?

As ultrasound is a subjective dynamic examination, heavily reliant on real time data the reliability of analysis will be via double reporting of each case by two researchers, highly experienced in fetal echocardiography. The validity of the study will be measured in determining its ability to obtain the required data from the information provided. To measure this, a small pilot study of 5 cases will be undertaken to ensure that sufficient information can be obtained before proceeding to the larger study.

Data will be analysed to assess any trends in aetiology of undiagnosed conditions and also the geographical location. Any identified trends in geographical location will be reported to the superintendent sonographer of the site to inform their on-going local audit process.

A parent with a child with an antenatally undiagnosed CHD was consulted and involved in the planning of the methodology to ensure service user agreement with the study principles and process.

Ethical implications
Patient sensitivity issues regarding the nature of the research must be addressed. Maternal consent for information and participation in the study will be requested in the clinical situation which will likely be highly emotive due to the confirmation of a paediatric cardiac abnormality in

which will likely be highly emotive due to the confirmation of a paediatric cardiac abnormality in the child. Therefore in requesting consent, it is necessary for the clinician to give a clear indication of the purpose of the research and the need to retrospectively assess images, without undermining the patient's confidence in the diagnostic accuracy of the screening service. It is also imperative that the mother does not feel under duress to consent to the study, and therefore an information sheet and consent form will be given to the potential participant for future completion, thereby giving them a period of time to contemplate their participation. Three consultant clinicians will be involved in providing the study information to the patients will have extensive written and verbal information about the study and are appropriately trained to provide sufficient information that consent from the participant will be fully informed.

All paper based forms containing patient data will be secured in a locked cabinet in the Cardiff Cardiac Services office based on the clinical premises, which is key only access.

Images obtained via PACS network will not be anonymised as it will be necessary for the researcher to match maternal data to the relevant images. This data will be available to the researchers only and ultrasound images will be viewed only in the clinical environment, thereby limiting the risk of any breach of patient confidentiality. Any electronic data that may identify a participant will be held in password protected files on a password protected laptop computer, used solely for the purpose of the study.

Any examination information sent on CD will be anonymised for transfer to the study centre, but will be identified with pre-determined code known only to the researcher. This will ensure that identifiable patient data is not removed from the clinical area for transport to the research centre, but the patient information will be known to the researcher for the purposes of matching the antenatal images to the relevant case.

Data protection requirements will be adhered to in relation to each Local Health Board for the release of information/data for the NHS to the University through meetings with data protection officers and Caldicott guardians.

Individual sonographers who have undertaken the reviewed examination will not be identified to the researchers. All information pertaining to the sonographers undertaking the imaging will be anonymised for the researchers. Any suggestion of inaccuracies or shortcomings in diagnosis identified to an individual department will be reported generically to the superintendent sonographer to inform on-going local audit procedures.

Ethical approval for the study will be sought from Cardiff University and the National Research Ethics service (REC Wales). R & D passport approval will be sought from Cardiff & Vale University Health Board.

Potential impact of the study
The study can effect improvement in patient outcomes by providing a means of prospective monitoring of diagnostic accuracy of CHD by antenatal ultrasound screening, while enabling early identification of potential training needs and possible opportunity for implementation of further diagnostic techniques to improve diagnostic accuracy.

Dissemination Strategy
Articles in relevant journals: including – Radiography, Ultrasound, British Journal of Obstetrics and Gynaecology, Heart, Welsh Paediatric Journal, Cardiolgy in the Young.

Conference papers: including - BMUS Scientific Meeting, UKRC, British Maternal and Fetal Medicine Society.