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Cyanotic Congenital Heart Disease Modes of Presentation and Prenatal Detection

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Abstract

Prenatal detection of structural congenital heart disease (CHD) optimises cardiovascular stability pre-operatively and post-operative outcomes. We compared prenatal detection rates of critical CHD in units offering universal fetal anomaly scans with those offering imaging to selected women.

One hundred and thirteen infants met inclusion criteria. The overall pre-natal detection rate for critical CHD was 57% of liveborn infants. It was 71% (57/80) in hospitals who offered a universal anomaly scan and 29% (9/31) in centres offering a limited service. Postnatal diagnosis was associated with PICU admission (p=0.016) and pre-operative mechanical ventilation (p=0.001). One-year mortality was 10 fold higher in the postnatally diagnosed group 15% vs 1.55% (p=0.0066).

There is a significant disparity between centres offering universal anomaly versus selective screening. Prenatal detection confers advantage in terms of pre-operative stability and one year survival. Failure to deliver an equitable service exposes infants with CHD to avoidable risk.

Abreviations
CHD, congenital heart disease; PICU, paediatric intensive care unit; NWIHP, National Women and Infants Health Programme; NCHC, CHIC, National Children's Heart Centre at Children's Health Ireland at Crumlin; TAPVD, total anomalous pulmonary venous drainage, HLHD, Hypoplastic left heart disease, HRHD, Hypoplastic right heart disease, LVOTO, left ventricular outflow tract obstruction.

Introduction

Congenital heart disease occurs in 0.8% of all births and in Ireland accounts for 13.2% of infant deaths from congenital malformation[1,2]. It may occur in isolation or as part of a chromosome malformation or genetic syndrome.[3] 90% of cases of congenital heart disease occurs in pregnancies where at the time of booking for pre-natal care there are no identifiable risk factors. [1, 4, 5] Universal screening at the time of a 20-22 week anomaly scan is necessary to detect all cases.
Prenatal diagnosis enables planned delivery, at term, in a tertiary neonatal centre with early transfer to the National Children's Heart Centre. Prenatal diagnosis facilitates targeted post-natal care to ensure cardiovascular stability, improving early morbidity and mortality in critical CHD.[6, 7] In addition it facilitates prenatal counselling regarding diagnosis, postnatal therapeutic options and long-term prognosis. In some cases, this will result in a parental decision to terminate the pregnancy.

International guidelines advocate for universal provision of fetal anomaly scans between 20 and 22 weeks' gestation [8, 9] to facilitate prenatal diagnosis of CHD. Recent national studies have highlighted significant variability in access to

fetal anomaly ultrasound scans in maternity hospitals in Ireland[10]. During the time frame of data collection for this audit 23% of women booking for antenatal care in Ireland were not routinely offered an anomaly scan[10].

Recent large cohort studies have assessed prenatal detection rates and efficacy of existing screening policies internationally[5, 11, 12], in an effort to optimise access to and standardisation of fetal screening. To date in Ireland the prenatal detection rates of congenital heart disease have not been well defined.

This study determined the incidence of critical congenital heart disease in infants in Ireland and provided an assessment of prenatal detection rates within this cohort reflecting the efficacy of current prenatal screening policies. We compared site of delivery, mode of transfer to tertiary cardiology services and condition on arrival between the prenatally and postnatally diagnosed cohorts. We also compared the incidence of prenatal versus postnatal detection in the studied cohort with a control group studied over a 12-month period in 2009/10 presented to the Irish Paediatric Association in 2011 to determine changes in the prenatal detection rate over a 5-year period[13].

Methods

This was a single centre retrospective cohort study. All infants born between May 1st 2015 and May 1st 2016 and admitted to the NCHC in the first 6 weeks of life, with a diagnosis of duct dependent congenital heart disease or obstructed total anomalous pulmonary venous drainage were eligible for inclusion. The study was approved by the institutional ethics committee at CHIC.

All children who are live-born with a diagnosis of CHD have their care delivered in the NCHC CHIC. Hospital admission and transfer records identified eligible infants. The diagnosis was defined as the most complex cardiac anomaly identified on the first postnatal echocardiogram at NCHC. Where multiple defects were present, diagnosis was categorized by the most significant duct-dependant lesion. Hospital of booking for was identified from the NCHC fetal referral database. Demographic information, presence of additional congenital anomalies, chromosomal anomalies or genetic syndrome was recorded. Survival data at 12 months of age was reviewed.

SPSS version 24 was used for descriptive statistics. Additional subgroup analysis was performed to assess the significance of other congenital anomalies, chromosomal anomalies, or genetic syndromes. χ^2 tests were used for comparisons of prenatal detection rates. *P* value <0.05 was considered statistically significant.

Results

113 of 223 infants admitted to NCHC in the first 6 weeks had duct dependant defects or obstructed total anomalous pulmonary venous drainage (TAPVD). This gives an overall incidence of critical congenital heart disease in live born infants transferred for tertiary care in Ireland during this period of 0.2% (113/65,536).

Infant demographics including presence of extra-cardiac and chromosomal anomalies are shown in Table 1. The overall prenatal detection rate of duct dependant congenital heart disease and obstructed total anomalous pulmonary venous drainage in this cohort was 57% (66/113). Chromosomal or other genetic disorders, were present in 15/113 (13%) of our study population. Additional extra-cardiac anomalies were found in an additional 6% (7/113). There was a trend towards higher prenatal detection rates in infants with chromosomal or genetic syndromes which was not statistically significant (*P*=0.85).

Table 1. Study population characteristics

	Prenatal Diagnosis Yes (*n*=66)	**Prenatal Diagnosis No (*n*=47)**
Gestation at birth (completed weeks)	38 (28-41)	38 (29-41)
Birth weight (kg)	3.2 (1.02-4.5)	3.2 (1.03-4.18)
Apgars	9 (4-10)	9 (2-10)
Gender, N (%) male	43 (65%)	35 (76%)
Chromosomal abnormality/genetic syndrome	11 (17%)	4 (9%)
Extracardiac anomaly	3 (5%)	4 (9%)

Data for continuous variables are given as median and interquartile range and for categorical variables as N (%).

There was variation in detection rates by lesion type with a trend towards higher detection rates for transposition of the great arteries (78%), and hypoplastic left heart syndrome (62%) and lower rates for valve stenosis and extra-cardiac defects (coarctation of the aorta, TAPVD) (Table 2). However, due to the low incidence of certain defects it was not possible to evaluate statistical significance of these variances. Cardiac defects visible on a 4 chamber view were more likely to be diagnosed prenatally (36/53)68% of cases, compared with those where the anomaly was primarily detected on an outflow tract view (26/62) 42% of cases.

Table 2. Prenatal detection by cardiac diagnosis.

Diagnosis	Total	Prenatal Detection Rate
TGA	27	21 (78%)
Aortic Coarctation	27	12 (44%)
HLHS	21	13 (62%)
Critical Aortic stenosis	10	4 (40%)
HRHS	10	8 (80%)
Interrupted Aortic Arch	7	4 (57%)
Critical pulmonary stenosis	5	2 (40%)
Tetralogy of fallot/critical PS	2	2 (100%)
Obstructed TAPVD	2	0 (0%)
Severe LVOTO	1	0 (0%)
Severe Polyvalvar dysplasia	1	0 (0%)

Data given as N(%). HLHS, hypoplastic left heart syndrome; LVOTO, left ventricular outflow tract obstruction; HRHS, hypoplastic right heart syndrome; PS, pulmonary stenosis; TGA, transposition of the great arteries; TAPVD, total anomalous pulmonary venous drainage.

Table 3 demonstrates prenatal detection rates in hospitals providing routine anomaly scanning and those without provisions for universal screening. Detection of critical congenital heart disease was 71% where the patient booked for prenatal care in a hospital providing universal screening compared with 29% in centres who did not screen all patients attending ($p<0.0005$).

Table 3. Detection rates by hospital of booking

	Prenatal Diagnosis Yes (*n*=66*)	Prenatal Diagnosis No (*n*=45*)
Booking hospital providing routine 20-week anomaly scan	57 (71%)	23 (29%)
Booking hospital not providing routine 20-week anomaly scan	9 (29%)	22 (71%)

*Data given as N(%). *Data on hospital of booking was not accessible on 2 patients.*

Pre-operative stability and mortality at 1 year are demonstrated in Table 4. Postnatally diagnosed patients were more likely to require ICU admission 40% vs 20% (p=0.016), mechanical ventilation 34% vs 8% (p=0.001), and high dose prostaglandin 21% versus 6% (p=0.061). They were less likely to have access to the National Neonatal Transport Program. Early survival was reduced in the postnatally diagnosed cohort with a 1-year mortality 15% versus 1.5% in the prenatally diagnosed group (p=0.006).

Table 4. Pre-operative study population

	Prenatal Diagnosis Yes (*n*=66)	Prenatal Diagnosis No (*n*=47)	*P*
Ventilated for transfer	5 (8%)	16 (34%)	0.001
On Prostin	49 (74%)	26 (55%)	0.036
>5nanograms/kg	4 (6%)	10 (21%)	0.061
NNTP	59 (90%)	30 (63%)	0.006
PICU admission	13 (20%)	19 (40%)	0.016
Mortality	1 (1.5%)	7 (15%)	0.006

Data given as N(%). NNTP; National Neonatal Transport Program.

Discussion

Duct dependent congenital cardiac anomalies and obstructed TAPVD are fatal in the neonatal period in the absence of surgical or catheter intervention. This is reflected in our cohort; postnatal diagnosis was associated with increased morbidity, and transport out of hours by a non-standardised transfer team. One-year mortality was 10-fold greater in the postnatally diagnosed group.

Overall prenatal screening in Ireland currently detects 58% of duct dependant cardiac lesions and obstructed TAPVD. This represents a marked improvement over the past 5 years from 25% in our 2009/2010 cohort[13]. Although not statistically significant in our sample, there was a trend towards higher prenatal detection rates in fetuses with extracardiac malformations and genetic disorders than in isolated cardiac defects. There is a higher prenatal detection rate in cardiac defects visible on the 4 chamber view, and lower detection rates of lesions of isolated valvar stenosis and of the great vessels. This is consistent with previous studies [5, 12] and reflects the increased training demands of the addition of the outflow tract view as well as the development of these lesions late in pregnancy after the routine anomaly scan.

Rates of prenatal detection in those Irish centres providing universal fetal anomaly screening approach 80%, this compares favourably to existing European data. However, there is disparity in the detection of CHD prenatally in Ireland with significantly lower detection rates in those centres not offering routine universal screening. Universal access to a 20-22 week anomaly scan is a minimum standard of prenatal care as per international guidelines[8, 9]. During the study period, 23% of women in Ireland were not offered a routine anomaly scan[10]. This deficit has been improved by the HSE NWIHIP but remains a risk.

This is the first study to detail national prenatal detection rates for critical CHD in Ireland. While the number of maternity hospitals offering routine anomaly ultrasound scans has increased since 2017 this study highlights that universal access to routine anomaly screening would improve prenatal detection of critical congenital cardiac disease and impact prenatal counselling, morbidity and mortality in this cohort. National healthcare policy guidelines should align with the resources required to achieve this goal.

While every effort has been made to include all prenatally diagnosed infants there may be some infants with an aneuploidy diagnosis that was considered lethal and who were therefore not referred for prenatal cardiac imaging. The numbers presented also exclude women who travelled outside the state for a termination of pregnancy following prenatal diagnosis.

Declaration of Conflicts of Interest:
There are no conflicts of interest to disclose.

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