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Diagnosis of severe congenital heart defects in Norway 2016

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BACKGROUND

Most structural congenital heart defects can be identified prenatally through ultrasound examination in pregnancy or via routine examinations during hospital maternity stays, but in some cases, heart defects are not discovered prior to discharge. There has been little previous research into detection rates with the various methods available. In this study, we have examined the timing and method of diagnosis of severe congenital heart defects.

MATERIAL AND METHOD

All children with severe heart defects born in Norway in 2016 and registered at Oslo University Hospital were included in this study. In addition, information on committee-handled abortions (after the 12th week of pregnancy) was obtained from the Medical Birth Registry of Norway.

RESULTS

In total, 105 of 181 (58 %) severe heart defects were diagnosed prenatally, and 51 (28 %) pregnancies were terminated. Among the 73 live-born children with severe heart defects that went unrecognised prenatally, 33 (45 %) of the heart defects were discovered outside of routine examinations and 9 (12 %) after discharge from hospital. Coarctation of the aorta was the most common diagnosis in cases of late-detected heart defects.

INTERPRETATION

This first national study of the diagnosis of severe congenital heart defects in Norway shows that most severe congenital heart defects are discovered prior to discharge from hospital after birth. However, almost half are diagnosed outside of routine examinations, and in some cases the diagnosis is not made until after discharge. The results indicate a need for new studies and for a quality registry of congenital heart defects to further improve diagnosis and early treatment.

Congenital heart defects are defined as congenital structural abnormalities of the heart and/or major intrathoracic vessels with functional or potential functional significance (1). Heart defects are the most common congenital malformation and affect approximately 1 in 100 live-born children (2). Most congenital heart defects have limited clinical significance, but approximately one-quarter are severe defects that require early identification and treatment (2, 3). Despite major advances in diagnostics and treatment, congenital heart defects are still a key cause of childhood morbidity and mortality (3, 4).

Many severe congenital heart defects can be detected prenatally through routine ultrasound examination in pregnancy (5). In Norwegian centres, prenatal detection rates

for congenital heart defects have varied from 25 % to 70 % in previous reports (5–7). After birth, all neonates undergo a clinical examination to uncover disorders such as congenital heart defects. Nevertheless, Meberg *et al*. found that approximately 25 % of heart defects in children in Vestfold in 1982–96 were discovered after discharge from hospital (8). Similar findings have been reported in other countries (9, 10). Some of these children have severe heart defects and die before accessing treatment (11). Recent decades have seen significant developments in prenatal diagnostics, and in 2013 routine measurement of oxygen saturation was introduced for all neonates in Norway to help detect heart defects prior to hospital discharge. Pulse oximetry has better predictive value for detecting heart defects than clinical examination alone and has been reported to reduce the number of missed cases by approximately 10 % (12, 13). However, pulse oximetry has low sensitivity for detecting certain severe heart defects, including coarctation of the aorta (14). Severe heart defects may still therefore be missed, and screening of all neonates with echocardiography has been proposed (15).

The purpose of this study was to examine the timing of diagnosis and the diagnostic method used to detect severe congenital heart defects in Norway in 2016.

Material and method

Information on committee-handled abortions approved in Norway in 2016 because of heart defects was obtained from the Medical Birth Registry of Norway statistics bank (16). Data on live births in 2016 of children with severe congenital heart defects were obtained from Oslo University Hospital's Clinical Registry for Congenital Heart Defects. Data were extracted on 31 December 2017 in order to capture late-detected heart defects. Oslo University Hospital has national responsibility for the treatment of children with severe congenital heart defects, and all patients treated at the Department of Paediatric Cardiology are entered into the registry. Data from the registry were supplemented with information obtained by reviewing medical records.

All congenital heart defects were classified according to the International Paediatric and Congenital Cardiac Code (17). We further divided congenital heart defects into severe and non-severe heart defects (3). The following congenital heart defects were classified as severe: Atrioventricular septal defect, truncus arteriosus, total anomalous pulmonary venous return, Tetralogy of Fallot, pulmonary atresia, hypoplastic left heart syndrome, congenitally corrected transposition of the great arteries, transposition of the great arteries, double outlet right ventricle, coarctation of the aorta, other isolated valve defects and other severe heart defects. Non-severe congenital heart defects were not included. Other serious diagnoses with major implications for the child's health and/or development, such as other congenital malformations and syndromes, were also recorded. All medical records were reviewed by two experienced paediatric cardiologists, and cases of doubt were classified by consensus among the authors.

Continuous variables are presented as median (interquartile range) and categorical variables as number and percentage.

The study is considered to be a quality assurance project and is therefore exempt from the requirement for regional ethics committee approval. Data collection and handling of personal data have been approved by the research Data Protection Officer at Oslo University Hospital.

Results

In the period from 1 January 2016 to 31 December 2016, 59 852 live births, 404 stillbirths and 272 committee-handled abortions were registered in Norway (16). Heart defects were reported in 51 of the committee-handled abortions. In the same period, 130 children with a severe congenital heart defect were registered in Oslo University Hospital's Clinical Registry for Congenital Heart Defects. The prevalence of severe congenital heart defects in Norway in

2016 was thus 299 per 100 000 pregnancies and 217 per 100 000 live births.

In addition to the 51 abortions owing to heart defects, congenital heart defects were diagnosed prenatally in 54 of the 130 affected live births. The estimated overall prenatal detection rate was therefore 58 %.

The distribution of the various types of severe congenital heart defects in live-born children is shown in Table 1. Other congenital malformations and syndromes occurred frequently (39 of 130 (30 %) children), but to varying degrees with different heart defects.

Table 1

Distribution of severe congenital heart defect diagnoses and other serious comorbid diagnoses (e.g. congenital malformations and syndromes) in Norway in 2016. Number (n).

Diagnosis	Number of children	Other serious comorbid diagnoses
Coarctation of the aorta	26	2
Other severe heart defects	26	7
Atrioventricular septal defect	21	15
Transposition of the great arteries	14	0
Other isolated valve defects	13	6
Tetralogy of Fallot	12	3
Double outlet right ventricle	7	3
Pulmonary atresia	6	3
Hypoplastic left heart syndrome	5	0
Total	130	39

The time of diagnosis for the various heart defects in live-born children is shown in Table 2. In 73 of 130 (56 %) children, the diagnosis was made after birth, with nine (12 %) children being diagnosed after discharge from hospital. The reasons for discovery of the heart defect after birth are shown in Figure 1. In 35 of the 73 (48 %) children, the heart defect was detected during a routine examination. Of these, 16 (46 %) children were diagnosed during a medical examination prior to discharge from hospital after birth. The most common clinical finding in the examination that indicated a heart defect was a heart murmur upon cardiac auscultation (15 of 16 (94 %) children). Pulse oximetry revealed a heart defect in 11 of 35 children whose heart defect was detected during a routine postnatal examination. Fewer than five children born in 2016 had a heart defect diagnosed in connection with follow-up at a child health centre. A heart murmur was the primary reason for referral in all those referred from child health centres.

Table 2

Timing of diagnosis in live-born children with severe congenital heart defects in Norway in 2016. Number (n).

Diagnosis	Number of children	Prenatal diagnosis	Before discharge from hospital	After discharge from hospital	Unknown time of diagnosis
Coarctation of the aorta	26	5	14	7	0
Other severe heart defects	26	11	12	2	1
Atrioventricular septal defect	21	10	10	0	1

Diagnosis	Number of children	Prenatal diagnosis	Before discharge from hospital	After discharge from hospital	Unknown time of diagnosis
Transposition of the great arteries	14	7	7	0	0
Other isolated valve defects	13	2	11	0	0
Tetralogy of Fallot	12	8	4	0	0
Double outlet right ventricle	7	3	3	0	1
Pulmonary atresia	6	4	2	0	0
Hypoplastic left heart syndrome	5	4	1	0	0
Total	130	54	64	9	3

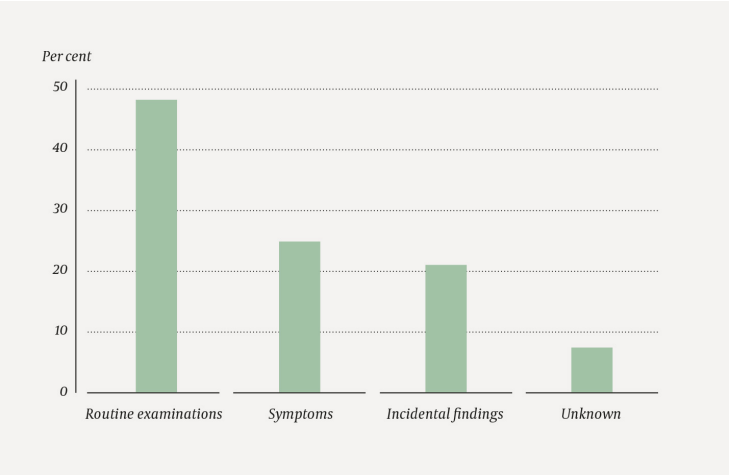

Figure 1 *Reasons for identification of severe congenital heart defects after birth in Norway in 2016.*

The median time to diagnosis of a severe congenital heart defect after birth was 2 (1–6) days. The median time to diagnosis in the nine children diagnosed following discharge from hospital was 114 days (18–318 days). Coarctation of the aorta was the most common late-detected heart defect (7 of 9 (78 %) children). In four of the seven children with late-detected coarctation, the heart defect was discovered following auscultation of a heart murmur, while the other cases were detected as a result of symptoms.

Discussion

This study on the detection of severe congenital heart defects in Norway in 2016 showed that 58 % of heart defects were discovered before birth. In 48 % of the children who received a postnatal diagnosis, the heart defect was detected during a routine examination. In 12 % of live births with no known heart defect, the condition was discovered only after discharge from hospital. Coarctation of the aorta was the most common diagnosis in cases of late-detected heart defects.

Routine ultrasound examination in pregnancy identifies many fetuses with heart defects. This enables the birth of children with severe heart defects to take place at centres with paediatric cardiology and cardiac surgery expertise, but it has also led to the termination of many pregnancies after the discovery of heart defects (3). New international guidelines for ultrasound examinations in 2013 included several different views as standard (18). This was shown in an American study to increase the detection rate from 44 % to 69 % (19). This national study confirms that the prenatal detection rate is comparable to previous findings from Norwegian regional centres and from other countries including Denmark (5, 6, 20), but the study is too small to be able to examine the impact of the new guidelines.

Since severe heart defects cannot always be diagnosed prenatally, some children will still be born with these conditions. For these children, rapid diagnosis and appropriate treatment

are critical for a successful outcome. Many of the heart defects discovered postnatally were identified during a medical examination prior to discharge from the hospital maternity unit. This confirms the importance of routine paediatrician-led examination of all neonates, and argues against early discharge from maternity units. It is also important to emphasise the need for prompt clarification with an echocardiogram should the clinical examination raise suspicion of a heart defect.

Auscultation of a heart murmur was the most common clinical finding that indicated a heart defect. However, it is important to be aware that murmurs are often present in children without heart defects, and about half of all children with heart defects do not have murmurs (21).

Screening with pulse oximetry is a simple and inexpensive test that was introduced in Norway in 2013 (22), and which has been shown in several studies to be cost-effective (23). In the current study, 15 % of heart defects with postnatal diagnosis were identified as a result of pulse oximetry. Given that some of these heart defects would probably also have been detected by routine medical examinations, we do not know exactly to what extent this technique has reduced the number of children with missed diagnoses.

Pulse oximetry has low sensitivity for left-sided obstructions, particularly coarctation of the aorta where oxygen saturation is usually normal or near normal (14). In this study, coarctation was the most common unrecognised heart defect upon discharge from maternity units. In children with a narrow coarctation, the systemic circulation is dependent on a persistent arterial duct, with symptoms first arising when the duct closes. This usually happens during the first few days of life, but may occur after several weeks. In the event of a critical stenosis, the condition can be life-threatening with rapid development of severe metabolic acidosis, cardiogenic shock and cardiac arrest. Immediate diagnosis and treatment to maintain blood flow through the arterial duct is life-saving. Similar symptoms may emerge in other duct-dependent heart defects including critical aortic stenosis, hypoplastic left heart syndrome, transposition of the great arteries, and tricuspid or pulmonary atresia.

Some studies have shown that routine echocardiography of all neonates reduces the number of unrecognised severe heart defects, but the procedure is technically demanding and resource intensive, and has an approximately 5 % false positive rate (24, 25). Standard Norwegian practice is to reserve echocardiography for children with a prenatal diagnosis or findings in a routine examination, or where there is clinical suspicion of a heart defect. The current study did not examine the usefulness of echocardiography.

Our study encompasses a nationwide cohort of children with severe congenital heart defects, and includes both committee-handled abortions and live-born children, but the study also has several weaknesses. The study included only patients born in 2016, and the number of individuals was relatively small. Non-severe heart defects, which are probably more likely to be detected later in life, were not included. We did not have the opportunity to quality assure the data on the number of abortions of fetuses with heart defects. We also have no information on the type of heart defect or on any comorbidities in this group. Unfortunately, Norway does not have a national registry of patients with congenital heart defects. Although Oslo University Hospital has national responsibility for the treatment of severe congenital heart defects in children and adolescents, some patients may have been examined and treated by local hospitals without the involvement of Oslo University Hospital. These individuals will therefore not be included in this study.

In summary, this study shows that most children with severe congenital heart defects are diagnosed during prenatal ultrasound examination or routine examinations prior to discharge from the maternity unit. However, almost half of children with severe heart defects that were detected postnatally were diagnosed outside of routine examinations, either as a result of symptoms or as an incidental finding. In some children, especially those with coarctation of the aorta, the diagnosis was not made until after discharge from the

hospital maternity unit. It is important to emphasise the need for immediate hospitalisation and rapid assessment by a paediatric cardiologist of young children with suspected severe heart defects. This study also shows that there is a need for continuous quality assurance of the methods for detecting heart defects. Establishing a national quality registry of congenital heart defects will be a vital part of this work.

MAIN MESSAGE

The prenatal detection rate for severe congenital heart defects was 58 % in Norway in 2016

Forty-five per cent of severe congenital heart defects diagnosed postnatally were discovered outside established routine examinations

Twelve per cent of children with a severe congenital heart defect that went unrecognised prenatally were discharged from the hospital maternity unit after birth without a correct diagnosis

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