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Discordances Between Pre-Natal and Post-Natal Diagnoses of Congenital Heart Diseases and Impact on Care Strategies

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ABSTRACT

BACKGROUND Pre-natal diagnosis of congenital heart disease (CHD) allows anticipation of urgent neonatal treatment and provides adequate information to the parents on cardiac outcomes.

OBJECTIVES This study sought to analyze the discordances between expert fetal cardiac diagnosis and final diagnosis of CHD and their impact on neonatal and long-term care strategies.

METHODS We included 1,258 neonates with a pre-natally diagnosed CHD and 189 fetopsies following termination of pregnancy at our tertiary center over a 10-year period. Pre-natal echocardiographic and final diagnoses were compared.

RESULTS For live births, we identified 368 (29.3%) discordances between pre- and post-natal diagnoses. The pre-natal diagnosis was different from the post-natal diagnosis in 36 cases (2.9%) and partially different with a major impact on neonatal treatment of the CHD in 97 cases (7.7%). In 235 cases (18.7%), the diagnosis was partially different with no impact on neonatal planned treatment. The discordances had a negative impact on late care strategy in 62 cases (4.9%): more complex CHD that was unsuitable for biventricular repair, leading to unplanned compassionate care, additional surgery or increase of the complexity level of the Aristotle score. A positive impact was found in 31 cases (2.5%): less complex CHD that allowed biventricular repair, fewer surgical procedures, or decrease of the complexity of the Aristotle score. For 275 patients (21.9%), there was no impact on late care strategy. Of the 872 terminations of pregnancy and intrauterine fetal deaths, 189 fetopsies were available: 16 (8.5%) different diagnoses, 27 (14.3%) major differences, and 60 (31.7%) minor differences.

CONCLUSIONS Correcting fetal cardiac diagnosis after birth can lead to significant changes in neonatal (10.6%) and late (7.4%) care strategies. Tools should be developed to try to improve the accuracy of pre-natal diagnosis of CHD. Clinicians should be cautious when predicting required treatment and outcomes during pre-natal counseling.
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The pre-natal diagnosis of congenital heart defects has contributed significantly to improving outcomes of high-risk congenital heart disease (CHD) through their prompt medical care or surgical treatment in specialized centers at birth (1-3). Pre-natal ultrasound screening for the detection of CHD is now offered to the majority of women in most countries of Europe, although the

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ABBREVIATIONS AND ACRONYMS

CHD = congenital heart disease

CMR = cardiac magnetic resonance

IUFD = intrauterine fetal death

TOP = termination of pregnancy

pre-natal screening policies and pre-natal detection rates vary greatly (4,5). The accuracy of fetal diagnosis of CHD allows anticipation of urgent treatment immediately after birth (6–9). Another important aim of fetal diagnosis of CHD is to give adequate information to the parents on cardiac and noncardiac outcomes (10–12). Finally, accompanying families who choose termination of pregnancy (TOP) is also a major issue. Few studies have evaluated the accuracy of fetal cardiac diagnoses (13–16). Here, we sought to analyze the discordances between the fetal diagnosis and the final cardiac diagnosis of CHD after birth or after TOP and intrauterine fetal death (IUFD), but we also analyze the impact of these discordances on planned neonatal and long-term care strategies.

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METHODS

POPULATION. Over a 10-year period, all neonates with a pre-natal diagnosis of CHD at our center, and who were delivered onsite were retrospectively included into the study. Expert fetal cardiologists performed all fetal echocardiographies. The last fetal echocardiography was used for comparisons, but all available anatomical details that had been described during the fetal cardiac follow-up were included in the final fetal cardiac diagnosis. The final post-natal diagnosis of the CHD was based on ultrasound, computed tomography/cardiac magnetic resonance (CMR) imaging, and surgical reports. For TOP and IUFD, a specialist in cardiac anatomy reviewed the autopsies.

Comparisons between fetal diagnosis and confirmed post-natal/autopsy diagnosis were performed by 2 authors (M.B. and D.B.). In case of discordance between the 2 reviews, a third evaluation of discordances was done in common between the 2 reviewers.

We intentionally removed from the analysis all false-positive cases of CHD seen at the screening level (i.e., normal fetal echocardiography at the expert level). Indeed, the aim of the study was to not to evaluate the correlation between first-line sonographers and pre-/post-natal expert diagnosis. We also excluded false-positive diagnoses of isolated coarctation of the aorta. Indeed, this could not be considered a discordance but only a limitation of the performance of the echocardiographic predictors of neonatal coarctation largely described elsewhere (17,18). If the risk of coarctation was associated with another pre-natally diagnosed CHD, we included false-positive/false-negative diagnoses of coarctation of the aorta in our analysis.

IMPACT ON NEONATAL TREATMENT OF THE CHD. The discordances between pre- and post-natal diagnoses were classified into 3 groups according to their impact on immediate neonatal care. The first group has a different diagnosis (completely different CHD). The second has a partially different diagnosis with a major (positive or negative) impact on the treatment of the CHD. This group included patients for whom the change in diagnosis leads to a change in medical care (prescription of prostaglandin), an unexpected need for interventional or surgical intervention during the first days of life, or conversely, no intervention needed, although pre-natally planned. The third group has a partially different diagnosis with no major impact on the planned treatment of the CHD.

The imprecision the fetal diagnosis of the CHD was considered as having an impact on neonatal treatment if the patients were in the first 2 groups.

IMPACT ON LONG-TERM CARE STRATEGY OF THE CHD. We compared the pre- and post-natal diagnoses according to their impact on care strategies after the neonatal period. We collected the planned treatment program according to pre-natal diagnosis (performed by cardiologist expert in fetal echocardiography) and from the report from the pre-natal meeting between the pediatric cardiologist and the parents. The data retrieved from the pre-natal files were: 1) whether or not the CHD was suitable for a biventricular repair (excluding borderline cases); and 2) the number and type of planned interventions.

After birth, we collected all interventions performed until last follow-up. We did not take into account unplanned interventions secondary to post-natal interventions. If the pre-natal number of interventions was uncertain, we determined that the post-natal number of interventions was increased if the modification of the diagnosis was responsible of an additional intervention. We determined that the discordance between pre- and post-natal diagnoses had a negative impact on long-term strategy if: 1) the CHD complexity was profoundly underestimated and led to proposing compassionate care whereas active treatment was planned pre-natally; 2) the CHD contraindicated a biventricular repair although biventricular repair was planned pre-natally; 3) the number of necessary interventions was higher than planned; and 4) in the other cases for which planned and post-natal diagnoses were different but not included in the first 3 groups, we used the Aristotle score (19) to compare the complexity of the procedures. We used the basic score to overcome the noncardiac items