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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.

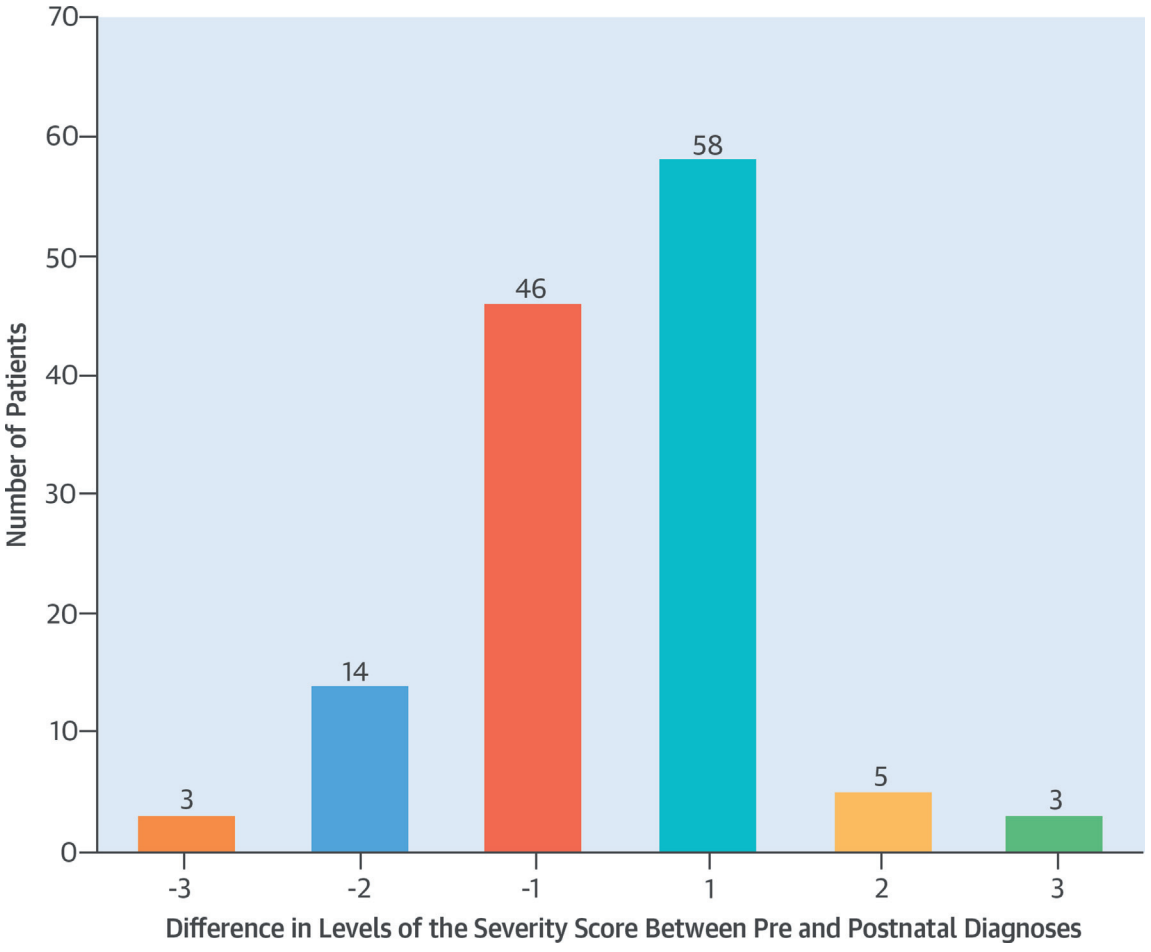
The imprecision the fetal diagnosis of the CHD was considered as having an impact on neonatal treatment if the patients were in the first 2 groups.

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

taken into account in the "comprehensive" score. We compared the scores of the intervention planned pre-natally with the scores of the intervention performed post-natally. We considered that the impact was negative if the differences between pre- and post-natal scores increased the complexity levels of the Aristotle score (1 to 5.9, 6 to 7.9, 8 to 9.9, 10 to 15). We determined that the discordance between pre- and post-natal diagnoses had a positive impact on long-term care strategy if: 1) the CHD allowed a biventricular repair whereas univentricular repair was planned pre-natally; 2) the number of necessary interventions was lower than planned; and 3) the differences between pre- and post-natal Aristotle scores reduced the level of complexity according to the 4 complexity levels of the basic score. We determined that the discordance between pre- and post-natal diagnoses had no significant impact on long-term care strategy if: 1) the diagnoses were different but the interventions performed were those planned pre-natally; and 2) the interventions performed were different from those planned pre-natally but the complexity level of the Aristotle basic score was unchanged.

CENTRAL ILLUSTRATION **Accuracy of Fetal Cardiac Diagnosis: Impact on the Severity of Projected Outcome**



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Difference in levels between pre- and post-natal diagnoses according to the fetal cardiovascular disease severity scale (20) in the 368 cases of discordance. Each congenital heart defect with discordance was classified in 1 of 7 severity levels of the score for the pre- and post-natal diagnosis. The difference in the score (either positive or negative) is reported in the figure. The discordances modified the level in 129 of 368 cases (10.3% of the total population). Most of the differences were within 1 level to each other.

IMPACT ON THE SEVERITY OF PROJECTED OUTCOME. To assess the global impact of the discordances between pre- and post-natal diagnoses, we used the fetal cardiovascular disease severity scale developed by Davey et al. (20). We classified all CHD in 1 of 7 levels of severity according to pre- and post-natal diagnoses. We determined that the difference between the level of severity between pre- and post-natal was positive if the level of severity was lower, and negative if it was higher.

TERMINATION OF PREGNANCY. In the group of TOP and IUFD, we compared the pre-natal diagnosis with the final diagnosis obtained by autopsy when available. The discordances between pre-natal diagnosis and autopsy were classified into 3 groups: 1) different CHD diagnosis; 2) partially different diagnosis with a potential impact on neonatal treatment or on long-term cardiac care strategy; and 3) partially different diagnosis with minor differences with no potential impact on either neonatal treatment or on long-term cardiac care strategies. The hospital's local committee on clinical investigation approved the review of medical records.

STATISTICS. Descriptive data are expressed as numbers and percentages.

TABLE 1 Pregnancy Outcomes and Verification Rates According to Heart Defect Categories

	IUFD		TOP		Live Birth	
	n	Verified Diagnosis	n	Verified Diagnosis	n	Verified Diagnosis
Total	73		799		1,258	
Transposition of the great arteries and variants	1	1 (100)	13	3 (23)	328	328 (100)
Coarctation of the aorta and variants	6	0 (0)	33	16 (48)	168	168 (100)
Single ventricle and functionally univentricular heart	12	1 (8)	320	81 (25)	148	148 (100)
Tetralogy of Fallot and variants	6	1 (17)	90	23 (26)	133	133 (100)
Atrioventricular septal defect and atrioventricular valves anomalies	13	0 (0)	97	8 (8)	60	60 (100)
Double outlet right ventricle	5	0 (0)	31	5 (16)	50	50 (100)
Pulmonary stenosis or pulmonary atresia (suitable for biventricular repair)	0	0 (0)	5	2 (40)	45	45 (100)
Ventricular septal defect	5	1 (20)	16	4 (25)	40	40 (100)
Congenitally corrected transposition of the great arteries	0	0 (0)	21	3 (14)	31	31 (100)
Aortic stenosis	0	1 (0)	29	7 (24)	25	25 (100)
Heterotaxy and isomerism	3	2 (0)	45	13 (29)	24	24 (100)
Common arterial trunk	0	3 (0)	26	7 (27)	22	22 (100)
Tumors	2	4 (0)	26	10 (38)	19	19 (100)
Interrupted the aortic arch	2	5 (0)	22	1 (5)	18	18 (100)
Anomaly of the venous return	1	6 (0)	0	0 (0)	17	17 (100)
Anomaly of the pulmonary return	0	7 (0)	1	1 (100)	10	10 (100)
Other	17	1 (6)	24	3 (13)	120	120 (100)

Values are n or n (%).
IUFD = intrauterine fetal death; TOP = termination of pregnancy.

RESULTS

Over a 10-year period, 1,258 neonates with pre-natally diagnosed CHD were included into the study. In addition, we compared pre-natal diagnosis of CHD with autopsy findings for 189 TOP and IUFD performed at our center during the same period (**Central Illustration**). **Table 1** shows the different rates of live births, TOP, and IUFD according to the different categories of CHD, as well as the rates of verification. All live births had post-natal verification. Pre- and post-natal diagnoses were identical in 890 of 1,258 live births (70.7%).

There was no disagreement between the 2 reviewers for assessment of the impact on neonatal and long-term care strategies. For 53 of 380 (14%) cases the reviewers disagreed in level of the fetal cardiovascular disease severity scale. Consensus was achieved after third evaluation.

Median gestational age at diagnosis was 26 weeks (range from 14 to 40 weeks) with no statistical difference between correct diagnoses and discordances. We identified 368 discordances between pre- and post-natal diagnoses of the CHD (29.3%) (**Figure 1**).

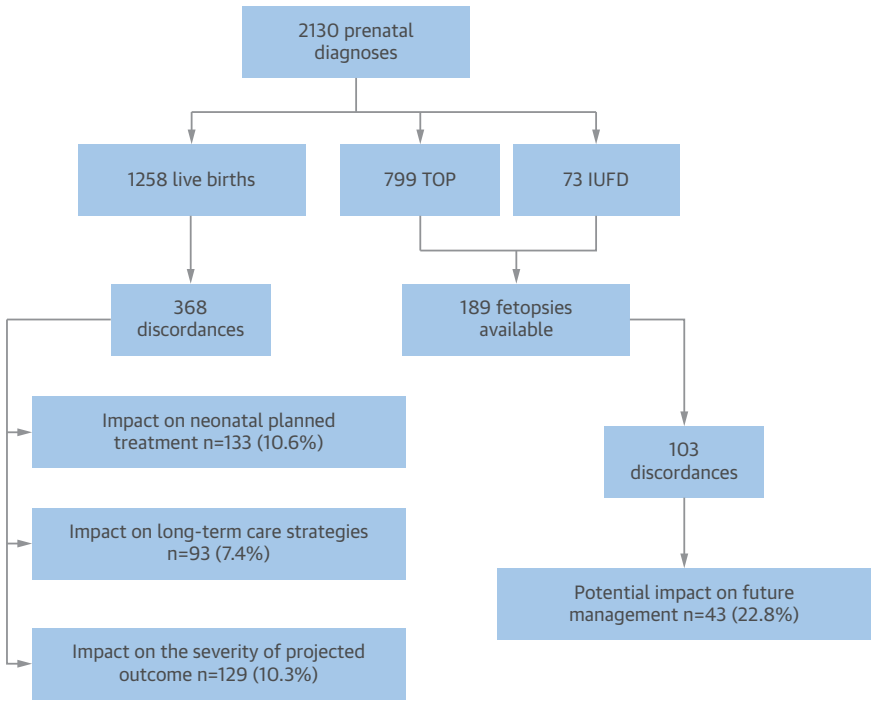
IMPACT ON NEONATAL PLANNED TREATMENT OF THE CHD. The discordances between pre- and post-natal diagnoses had an impact on neonatal planned treatment of the CHD in 10.6% of the cases (**Table 2**). The changes in the neonatal interventions were related to the need of prostaglandin E1 in 67 cases, Rashkind procedure in 17 cases, and urgent surgery in 20 cases.

IMPACT ON LONG-TERM TREATMENT OF THE CHD. **Table 3** describes the impact (negative, positive, or insignificant) of the discordances founded between the pre- and post-natal diagnoses on long-term treatment strategy. Overall, the discordances between pre- and post-natal diagnoses had an impact (positive or negative) on long-term care strategy of the CHD in 7.4% of cases. In 19 cases, the impact could not be measured because the neonates died before the start of the project.

IMPACT ON THE SEVERITY OF PROJECTED OUTCOME. The discordances between pre- and post-natal diagnoses modified the level of the fetal cardiovascular disease severity scale in 129 of 368 cases (10.3% of the total population to 35% of the discordances). The severity score was lower in 63 cases (−1 level in 46, −2 levels in 14, and −3 levels in 3). The severity score was higher in 66 cases (+1 in 58, +2 in 5, and +3 in 3) (**Central Illustration**).

DISCORDANCES BETWEEN PRE-NATAL DIAGNOSIS AND AUTOPSY AFTER TOP. Pre-natal diagnosis and autopsy findings were identical in 86 of 189 cases

FIGURE 1 Flow Diagram of the Results



All neonates with a pre-natally diagnosed congenital heart disease born in our center were included over a 10-year study period. All available fetal autopsies following termination of pregnancy (TOP) or intrauterine fetal death (IUFD) during the same period were reviewed. Discordances in the diagnoses and impact on care strategies were evaluated in both groups.

(45.5%). In 16 cases (8.5%), the diagnosis was completely different. In 27 cases (14.3%), the discordances were partial with a potential impact on neonatal treatment or on long-term cardiac care strategies. In 60 cases (31.7%), the discordances were minor with no potential impact on either neonatal treatment or on long-term cardiac care strategies.

TYPE OF ANATOMICAL DISCORDANCES. The anatomical discordances are shown in Table 4. More than 1 anatomical discordance could be identified in the same case. The most frequent discordances interested the outflow tract anatomy: ventriculoarterial connections, as well as right and left ventricular outflow tract (18.8%). These discordances had the greatest impact on neonatal treatment. Anatomical differences with the greatest impact on long-term treatment were the anomalies of the right outflow tract, the anomalies of the pulmonary artery branches, and the anomalies of the pulmonary veins.

TABLE 2 Impact of the Discordances on Planned Neonatal Treatment

	n	% of the Total Population	% of the Discordances
Different diagnosis	36	2.9	9.8
Partially different diagnosis with major (positive or negative) impact on the planned neonatal treatment	97	7.7	26.3
Partially different diagnosis with minor (positive or negative) impact on the planned neonatal treatment	235	18.7	63.9

DISCUSSION

In our series, in 10.6% of the cases, the discordances between pre- and post-natal diagnoses led to incorrect prediction of neonatal potential intervention. In addition, the long-term care strategy was changed in 7.7% of the cases. Finally, the global severity scale, designed to predict the "quality of life" after a fetal diagnosis of CHD was modified in one-tenth of the patients. Our results suggest that the most accurate precision is needed for pre-natal diagnosis of CHD as anatomical details may modify the therapeutic plans and the need for in utero transfer. These limitations should be part of the information given to the parents during pre-natal interviews. Of note, all fetal

TABLE 3 Impact of the Discordances on Long-Term Care Strategies

		n	Total N	% of the Total Population	% of the Discordances
Negative impact on long-term care strategy	More severe CHD leading to unplanned compassionate care whereas active treatment was planned pre-natally	15	62	4.9	16.8
	Biventricular repair contraindicated although planned pre-natally	4			
	Higher number of necessary interventions than planned pre-natally	30			
	Increase of the complexity level of the Aristotle score	13			
Positive impact on long-term strategy	Biventricular repair allowed whereas univentricular repair was planned pre-natally	2	31	2.5	8.4
	Lower number of necessary interventions than planned pre-natally	19			
	Reduction of the complexity level of the Aristotle score	10			
No significant impact on long-term care strategy	Different diagnoses but same interventions performed	192	275	21.9	74.7
	Different diagnoses, different interventions performed but unchanged level of complexity of the Aristotle score	64			
	Impact could not be measured	19			

echocardiographies were performed by expert fetal cardiologists after a 3-step process (screening, CHD confirmation, and expertise), and prognostication of the outcomes was defined with the final diagnosis. As the discrepancies between first-line sonographers and expert fetal cardiologists are well known, this reinforces the need for expertise of fetal CHD before any information shall be given on potential outcomes (21,22).

The most frequent discordances between pre- and post-natal diagnoses interested the left and right outflow tracts. Adding the 3 vessels view to the analysis of the fetal heart is necessary to evaluate these segments, but it is not systematically performed in the current screening for fetal CHD (23–25). In tetralogy of Fallot, recent studies showed that the anatomy of the pulmonary valve and the direction of flow in the arterial duct are reliable tools for predicting neonatal need for ductal patency as well as the need for early surgical treatment (26–28). Hirji et al. (28) showed that pulmonary-aortic annulus ratio was reliable to predict the type of surgical repair in tetralogy of Fallot (valve-sparing repair or transannular patch). These findings were not useful in CHD with univentricular physiology and pulmonary obstruction (26). Certainly, the progression of the right outflow tract obstruction during pregnancy is a limitation to anticipate neonatal physiology.

Defining the respective position of the great vessels and their spatial relationship with the ventricular septal defect is also challenging in double outlet right ventricles. Indeed, the neonatal physiology might be completely different if the ventricular septal defect is subaortic or subpulmonary and the planned therapeutic interventions could be completely modified. In a series of double outlet right ventricles, Gedikbasi et al. (29) showed that the type of ventricular septal defect and the type of malposition of the great arteries could be defined in approximately 80% of the cases. This was confirmed in another series including different types of conotruncal defects (30). New imaging techniques, such as 3-dimensional echo and fetal CMR (31,32), may help to precisely define this cardiac segment.

We also found a significant number of undetected abnormal pulmonary venous connections. This is not surprising, as diagnosing these anomalies remains difficult (33,34), particularly in complex CHD such as heterotaxy syndromes. As abnormal pulmonary venous connection is currently seen in these conditions, particular attention should be given to identifying pulmonary veins to prevent early neonatal demise.

Finally, pulmonary and aortic valve anomalies that were not seen pre-natally had a limited impact on planned treatment. Certainly, the sensitivity of fetal echocardiography to detect bicuspid aortic valve, for example, is limited and information on its frequent association with left outflow tract anomalies should be given to the parents.

Cardiac anomalies that could theoretically be diagnosed with the 4-chamber view were not rare. These undetected anomalies were mainly atrioventricular valve malformations that had consequences on late care strategies but not on neonatal treatment. Mainly, they compromised the biventricular repair or led to a higher complexity (**Table 1**). Certainly, straddling of the tricuspid valve or of the mitral valve could be more accurately diagnosed to prevent this change in planned treatment. It is potentially more

challenging for mitral valve anomalies in left-right asymmetry as these defects may be evolutive pre-natally and also after birth. Therefore, there is a need for a more accurate analysis of the mitral valve and for a precise definition of the left heart structures measurements to predict biventricular repair. Indeed, echocardiographic factors that can predict biventricular repair in fetuses with small left ventricles are limited, whereas they are numerous in neonates (35). In a recent study of fetal "borderline" left ventricles, a mitral valve annulus Z-score below −1.9 and a ratio of tricuspid-mitral annulus below 1.5 had a sensitivity of 100% to predict biventricular repair. Conversely, a ratio of right-left ventricular end-diastolic diameters above 2.1 had a 95% specificity to predict univentricular palliation (36). Along the same line, right ventricular-left ventricular end-diastolic dimension ratio between 2 and 4 SD for gestational age was the only predictor for biventricular repair for borderline left ventricles in pre-natally diagnosed atrioventricular septal defect and double outlet right ventricle (37). In addition to precise measurements of the left heart structures, a precise description of mitral valve and left outflow tract should be a priority, as outcomes can widely vary according to associated anomalies of these structures (38).

The proportion of discordances between fetal diagnosis and autopsy reports were more frequent than for live births. This difference can easily be explained by the early term for termination of pregnancy in the majority of cases. Indeed, when pregnancy continued, the fetal diagnosis of CHD could be more accurate with sequential surveillance and repeated check for associated anomalies. Only 189 fetopsies were available. Fetal autopsies are always proposed in our institution but performed only after parental approval. This may represent a biased sample of the population (probably more complex cardiac defect).

Pre-natal counseling following a pre-natal diagnosis of CHD is complex and involves several variables aside from the cardiac diagnosis such as the gestational age at the diagnosis, association with extracardiac malformations, and natural potential evolution of the cardiac defect during the fetal life (10). In addition, this work shows us that there is a limit to the precision and to the predictive value of fetal echocardiograms. This should encourage the clinicians to be extremely cautious when giving parents predictions of the neonatal or late planned care strategies and outcomes.

Our results are similar to previous studies with regard to the number of discordances between pre- and post-natal diagnoses. Indeed, Cha et al. (16) found 15 of 148 minor differences (10.1%) and 2 of 148 major discordances (14%) in global treatment plans in their series. Clur et al. (13) found a correlation between pre- and post-natal Aristotle scores in 81% of their 231 cases. Berkley et al. (15) found 3 of 53 minor and 3 of 53 major differences in analyzing pre- and post-natal diagnoses and the impact of these discordances on delivery plans, neonatal planned treatment, and location of delivery. Recently, van Velzen et al. (14) found 8.1% of 708 cases with no similarity between pre- and post-natal diagnosis and 9.9% of discrepancies that did not result in a different treatment. In our series, we confirmed that accuracy of pre-natal diagnosis of CHD should be improved, not only to predict early neonatal demise but also to gain accuracy on prediction of late outcomes. New tools should be used to augment precision of the anatomical diagnosis.

Recent advances in 3-dimensional ultrasound technologies and post-processing visualization modalities has led to the development of new techniques allowing real-time volumetric 3-/4-dimensional reconstruction of heart anatomy (39–41). Although echocardiography remains the gold standard for fetal heart imaging, its limitations are well known: operator dependency; maternal obesity; position of the fetus; or ossification. Fetal CMR can be a reliable second-line tool (42–44). However, CMR has some limitations such as the fetal movement, the high fetal heart rate, and the difficulties in heart gating (45).

STUDY LIMITATIONS. Our study has some limitations particularly for the prediction of complexity. First, we defined the importance of the discordances between pre- and post-natal diagnoses according to our own definition. Second, we used the Aristotle score that incorporates expected mortality, anticipated morbidity, and technical difficulty. The changes in the Aristotle complexity levels with negative impact were limited but could have been underestimated because of the design of the score. Indeed, when 2 simultaneous procedures were performed after birth, if the more complex was already planned pre-natally while the second was unexpected, this had no impact on the complexity level while subjectively the risk was higher (19).

For global severity comparisons, we used the fetal cardiovascular disease severity scale (20). This could be in contradiction with our own classification of positive or negative impact on outcomes, as this scale does not consider univentricular physiology as being always worse than biventricular physiology. Indeed, it is a combination of anatomical description of the CHD, anticipated treatment, and overall prognosis including quality of life. We finally used 3 different approaches to analyze the impact of discordant

TABLE 4 Types of Anatomical Discordances*

		Neonates		TOP		Neonates + TOP	Neonates + TOP
		Total N	n	Total N	n	Total N (%)	n
Situs and position of the heart	Situs inversus: false negative	5	3	2	1	7 (1.2)	4
	Situs inversus: false positive		2		1		3
Pulmonary venous return	Pulmonary vein obstruction: false negative or positive	32	4	7	0	39 (7.1)	4
	Partial anomalous pulmonary venous return: false negative		12		4		16
	Partial anomalous pulmonary venous return: false positive		4		0		4
	Total anomalous pulmonary venous return: false negative		8		2		10
	Total anomalous pulmonary venous return: false positive		2		1		3
	Other		2		0		2
Systemic venous return	Left superior vena cava: false negative or positive	29	24	23	23	52 (9.4)	47
	Azygos continuation of the inferior vena cava: false negative		5		0		5
Interatrial septum	Ostium primum defect: false negative	3	2	0	0	3 (0.6)	2
	Common atrium: false negative		1		0		1
Interventricular septum	Wrong location of VSD	64	4	12	1	76 (13.8)	5
	Wrong number of VSD		6		0		6
	VSD: false negative		35		5		40
	VSD: false positive		19		6		25
Atrioventricular junction and valves	Anomaly of the mitral subvalvular apparatus: false negative or positive	52	22	19	3	71 (12. 9)	25
	Mitral cleft: false negative		8		1		9
	Straddling or overriding of the mitral valve: false negative		3		0		3
	Mitral atresia or stenosis: false negative		3		4		7
	Mitral stenosis: overestimated		1		6		7
	Tricuspid atresia or stenosis: false negative		1		0		1
	Tricuspid atresia: false positive		1		1		2
	Ebstein anomaly or dysplastic tricuspid valve: false negative or positive		8		4		12
	Straddling or overriding of the tricuspid valve: false negative or positive		4		0		4
	Common atrioventricular orifice: false negative		1		0		1
Atrioventricular connections	Atrioventricular discordance: false negative†	6	4	1	1	7 (1.3)	5
	Atrioventricular discordance: false positive‡		2		0		2
Global intracardiac anatomy	Incorrect intracardiac anatomy in functionally univentricular heart	23	14	19	15	42 (7.6)	29
	Incorrect estimation of right of left ventricle size		4		4		8
	Other		5		0		5
Ventriculoarterial connections	Subaortic conus in outflow tract defect: false negative or positive	34	9	8	3	42 (7.6)	12
	Subpulmonary conus in outflow tract defect: false negative or positive		8		1		9
	Incorrect VSD location in DORV		15		1		16
	Incorrect position of the great arteries in single ventricle		2		3		5
Right ventricular outflow tract	Obstruction: false negative	42	22	11	6	53 (9.6)	28
	Obstruction: underestimated		6		4		10
	Obstruction: overestimated		3		1		4
	Obstruction: false positive		11		0		11
Left ventricular outflow tract	Obstruction: overestimated	3	1	6	0	9 (1.6)	1
	Obstruction: false positive		1		0		1
	Obstruction: underestimated		0		5		5
	Obstruction: false negative		1		1		2
Arterial valves	Bicuspid pulmonary valve: false negative	64	10	6	0	70 (12.7)	10
	Bicuspid aortic valve: false negative or positive		54		6		60

Continued on the next page

diagnoses on care strategies, and we approximately found the same proportions for each of them. Lastly, our present study excluded all false-negative cases (after nonexpert cardiac evaluation) that represent the most important issue in pre-natal diagnosis of complex or life-threatening CHD. The rates of pre-natally diagnosed CHD in a population-based registry in our area has already been reported (4) and

TABLE 4 Continued

		Neonates		TOP		Neonates + TOP	Neonates + TOP
		Total N	n	Total N	n	Total N (%)	n
Aorta and aortic arches	Interruption of the aortic arch: false negative	28	6	19	0	47 (8.5)	6
	Interruption of the aortic arch: false positive		3		5		8
	Risk of coarctation: nonanticipated		3		1		4
	Right aortic arch: false negative		9		7		16
	Aortopulmonary window: false negative or positive		5		0		5
	Anomaly of the aortic arch: false negative		2		6		8
Main pulmonary artery and branches	MAPCA: false negative	30	15	4	1	34 (6.2)	16
	Pulmonary artery sling: false negative		1		0		1
	Disconnected pulmonary arteries: false negative		4		0		4
	PA-VSD instead of common arterial trunk or conversely		2		1		3
	Pulmonary arteries stenosis: false negative or positive		5		1		6
	Main pulmonary trunk in PA-VSD: false negative or positive		3		1		4
Coronary arteries	Coronary fistula: false negative or positive	9	9	0	0	9 (1.6)	9
Total		415		137		552	

*1 patient can have several discordances. †Nonseen atrioventricular discordance with normal ventriculoarterial connection or congenital corrected TGA instead of TGA. ‡TGA instead of congenitally corrected TGA.
DORV = double outlet right ventricle; MAPCA = major aortopulmonary collateral artery; PA = pulmonary atresia; TGA = transposition of great arteries; TOP = termination of pregnancy; VSD = ventricular septal defect.

clearly shows that progress is still needed in the screening for CHD in low-risk pregnancies.

CONCLUSIONS

Discordances between pre- and post-natal diagnoses of CHD are limited but might change the treatment both immediately after birth and for the long term. There is still room for improvement of pre-natal diagnosis of CHD not only to define the indication for in utero transfer or to predict the need for neonatal interventions, but also to better plan the future treatment of the cardiac defect as well as the quality of life of the child. Accordingly, clinicians should show some caution during pre-natal counseling regarding the planned required treatment and the outcomes based on the fetal echocardiogram.

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PERSPECTIVES

COMPETENCY IN PRACTICE-BASED LEARNING: Fetal echocardiography lacks precision for evaluation of anomalies of pulmonary venous return, ventricular outflow tract, and atrioventricular junctions and valves. Expert sonographers should address these competency gaps and through enhanced education improve performance of front-line sonographers.

TRANSLATIONAL OUTLOOK: Prospective studies are needed to develop standardized echocardiographic measurements that enhance diagnostic precision for assessment of congenital anomalies of the ventricular outflow tract.

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