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Pregnancy outcome in correlation with prenatal detection of congenital heart disease

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Objective

The study aims to evaluate the impact of a prenatally diagnosed heart defect on the further course of pregnancy, depending on the type of defect and associated fetal extracardiac diseases.

Methods

20-year (2003-2022) retrospective study analyzed a set of 51,321 examined pregnant women (51,222 fetuses) to whom fetal echocardiography was performed in the first and second trimesters of pregnancy. For each CHD identified, a principal cardiac diagnosis was established and associated genetic and morphologic extracardiac pathologies were recorded. Pregnancy outcome was evaluated as termination of pregnancy, intrauterine death and live birth.

Results

Overall, CHD was diagnosed in 1 % (520/51,222) of fetuses. Of this number, CHD occurred as an isolated anomaly in 66 % (342/520), in 24 % (125/520) the occurrence of CHDs was combined with genetic involvement, and in 10 % (53/520) with another morphological anomaly and a normal karyotype. The most frequently detected CHD was atrioventricular septal defect (n=85), which also showed the greatest number of genetic disorders in 59 % (50/85). The second most common defect was hypoplastic left heart syndrome (n=69), which, on the other hand, was dominated by isolated disabilities in 78 % (54/69). 50 % (260/520) of the parents decided to terminate the pregnancy, of this group, 49% (127/260) had CHDs isolated, 36 % (95/260) genetically and 15 % (38/206) otherwise extracardiacally affected fetuses. Only 2 % (13/520) of the fetuses died in utero, with a predominance of isolated CHDs in 70 % (9/13). There were 48 % (247/520) of live-born newborns with CHDs, with a predominance of isolated CHDs in 84 % (206/247).

Conclusion

In half of the prenatally detected CHDs, the parents decided to terminate the pregnancy. The decision depends on the severity of the defect and the associated anomalies of the foetus. Intrauterine foetal death with CHD is rare, and the causes are not quite clear, some of these defects are isolated, and hemodynamic consequences or unidentified genetic and metabolic diseases are possible. Isolated cardiac anomalies predominate in continuing pregnancies towards delivery.

Association of CHDs and genetic and morphological impairments from 2003–2022

CHD	n	Isolated (%)	Chromosomal aberrations (%)	Extra-cardiac anomalies (%)
Atrioventricular septal defect	85	22 (26)	50 (59) 39x Trisomy 21 6x Trisomy 18 2x Trisomy 13 1x Smith-Lemli-Opitz syndrome 1x trans 13/14 1x Smith-Magenis syndrome	13 (15) (situs anomalies, brain, gastrointestinal, kidney, lung abnormalities, limb defects)
Hypoplastic left heart syndrome	69	54 (78)	10 (15%) 4x Monosomy X 2x Edwards 1x Deletion 22q11 1x t 13/14 1x Tollner syndrome 1x mutation NOTCH1	5 (7) (situs anomalies, gastrointestinal, kidney, lung abnormalities)
Ventricular septal defect	58	37 (64)	14 (24) 7x Trisomy 18, 2x Trisomy 21 2x Deletion 22q11 2x Trisomy 13 1x Smith Magenis syndrome	7 (12) (limb defects, cleft face, kidney abnormality)
Tetralogy od Fallot	51	27 (53)	20 (39) 5x Trisomy 21 5x Deletion 22q11 2x Noonan sy 1x Trisomy 18 1x 47 XX+22, 1x 47 XX+9 5x others	4 (8) (situs anomalies, limb defects gastrointestinal abnormalities)
Transposition of great arteries	36	32 (89)	1 (3) 1x Mikrodeletion 15q11.2	3 (8) (lung ang genital abnormalities)
Coarctation of aorta	34	26 (76)	6 (2) 2x Monosomy X 2x Trisomy 18 1x Trisomy 13 1x Trisomy 9	2 (6) (gastrointestinal lung abnormalities)
Double outlet right ventricle	33	21 (64)	6 (18) 2x Trisomy 18 2x Trisomy 13 1x Deletion 22q11 1x Jacobsen syndrome	6 (18) (situs anomalies, brain/, gastrointestinal, kidney, lung abnormalities, limb defects, spina bifida)
Pulmonary stenosis	30	27 (90)	2 (7%) 1x Monosomy X/inv. 4 1x Triploidy 69	1 (3) (lung and kidney abnormalities)
Aortic stenosis	26	26 (100)	0	0
Tricuspid atresia	18	13 (72)	5 (28) 3x Trisomy 18 1x Monosmy X microduplication 22q11.2	0
Pulmonary atresia/ventricular septal defect	15	10 (67)	4 (27%) 3x Deletion 22q11 1x Trisomy 13	1 (6) (gastrointestinal abnormality)
Single ventricle	15	11 (73)	1 (7%) 1x Trisomy 18	3 (20) (gastrointestinal, multiple abnormalities)
Common arterial trunk	14	9 (64)	3 (22%) 1x Deletion 22q11 2x Trisomy 13 1x partial deletion 13q	2 (14) (kidney, multiple abnormalities)
Ebstein's anomaly	12	12 (100)	0	0
Pulmonary atresia/ intact ventricular septum	12	7 (59)	1 (8) 1x Triploidy 69	4 (33) (situs and multiple anomalies)
Corrected transposition of great arteries	5	3 (60)	0	2 (40) (situs anomalies)
Mitral atresia	4	4 (100)	0	0
Interruption of the aortic arch	3	1 (33)	2 (67) 1x Deletion 22q11 1x sy cri du chat	0
Total	**520**	**342 (66)**	**125 (24%)**	**53 (10)**

Table 2. Prenatally detected CHDs: termination pregnancy, intrauterine death, and delivery; divided to genetic and morphological pathologies

Congenital heart defect	Termination (%)				Intrauterine death (%)				Delivery (%)				Total
	n	CHD I	CHD+G	CHD+M	n	CHD I	CHD+G	CHD+M	n	CHD I	CHD+G	CHD+M	
Atrioventricular septal defect	**56**	8 (14)	36 (64)	12 (22)	**3**	1 (33)	2 (67)	0	**26**	13 (50)	12 (46)	1 (4)	**85**
Hypoplastic left heart syndrome	**59**	45 (76)	10 (17)	4 (7)	**2**	1 (50)	0	1 (50)	**8**	8 (100)	0	0	**69**
Ventricular septal defect	**11**	0	7 (64)	4 (36)	**2**	2 (100)	0	0	**45**	35 (78)	7 (15)	3 (7)	**58**
Tetralogy od Fallot	**19**	5 (26)	14 (74)	0	**0**	0	0	0	**32**	22 (69)	6 (19)	4 (12)	**51**
Transposition of great arteries	**10**	8 (80)	1 (10)	1 (10)	**0**	0	0	0	**26**	24 (92)	0	2 (8)	**36**
Coarctation of aorta	**8**	2 (25)	6 (75)	0	**0**	0	0	0	**26**	24 (92)	0	2 (8)	**34**
Double outlet right ventricle	**17**	7 (41)	4 (24)	6 (35)	**0**	0	0	0	**16**	14 (87)	2 (13)	0	**33**
Pulmonary stenosis	**2**	0	2 (100)	0	**2**	1 (50)	0	1 (50)	**26**	26 (100)	0	0	**30**
Aortic stenosis	**9**	9 (100)	0	0	**1**	1 (100)	0	0	**16**	16 (100)	0	0	**26**
Tricuspid atresia	**17**	12 (71)	5 (29)	0	**0**	0	0	0	**1**	1 (100)	0	0	**18**
Pulmonary atresia/ventricular septal defect	**11**	7 (64)	3 (27)	1 (9)	**1**	1 (100)	0	0	**3**	2 (67)	1 (33)	0	**15**
Single ventricle	**6**	3 (50)	1 (17)	2 (17)	**0**	0	0	0	**9**	8 (89)	0	1 (11)	**15**
Common arterial trunk	**11**	6 (55)	3 (27)	2 (18)	**0**	0	0	0	**3**	3 (100)	0	0	**14**
Ebstein's anomaly	**3**	3 (100)	0	0	**2**	2 (100)	0	0	**7**	7 (100)	0	0	**12**
Pulmonary atresia/intact ventricular septum	**12**	7 (59)	1 (8)	4 (33)	**0**	0	0	0	**0**	0	0	0	**12**
Corrected transposition of great arteries	**3**	1 (33)	0	2 (67)	**0**	0	0	0	**2**	2 (100)	0	0	**5**
Mitral atresia	**3**	3 (100)	0	0	**0**	0	0	0	**1**	1 (100)	0	0	**4**
Interruption of the aortic arch	**3**	1 (33)	2 (67)	0	**0**	0	0	0	**0**	0	0	0	**3**
Total	**260**	**127 (49)**	**95 (36)**	**38(15)**	**13**	**9 (70)**	**2 (15)**	**2 (15)**	**247**	**206 (84)**	**28 (11)**	**13 (5)**	**520**