

Recurrence Of Congenital Heart Disease
In Offsprings Of Mothers With Congenital Heart Disease,
By Fetal Echocardiography

Fesslova V, Inversetti A *, Sigismondi C*,
Brankovic J.

Centre of Fetal Cardiology. Policlinico San Donato IRCCS,
Milan

*Via Salute University, Ospedale San Raffaele, Milan, Italy

Background:

Maternal condition of congenital heart disease (CHD) bears a risk of recurrence and these women are nowadays routinely referred for prenatal screening.

Preconceptional counselling is very delicate in this population and the data relative to the recurrence rates are of extreme use in this field.

Objective of the study:

- to analyze the recurrence of congenital heart disease (CHD) in offsprings of mothers affected with CHD and to compare the results with the data of the literature.

Material and methods:

Retrospective-prospective study

of **310 pregnancies of 271 mothers with CHD**

studied by fetal echocardiography in our Centre between January 1995 and December 2015.

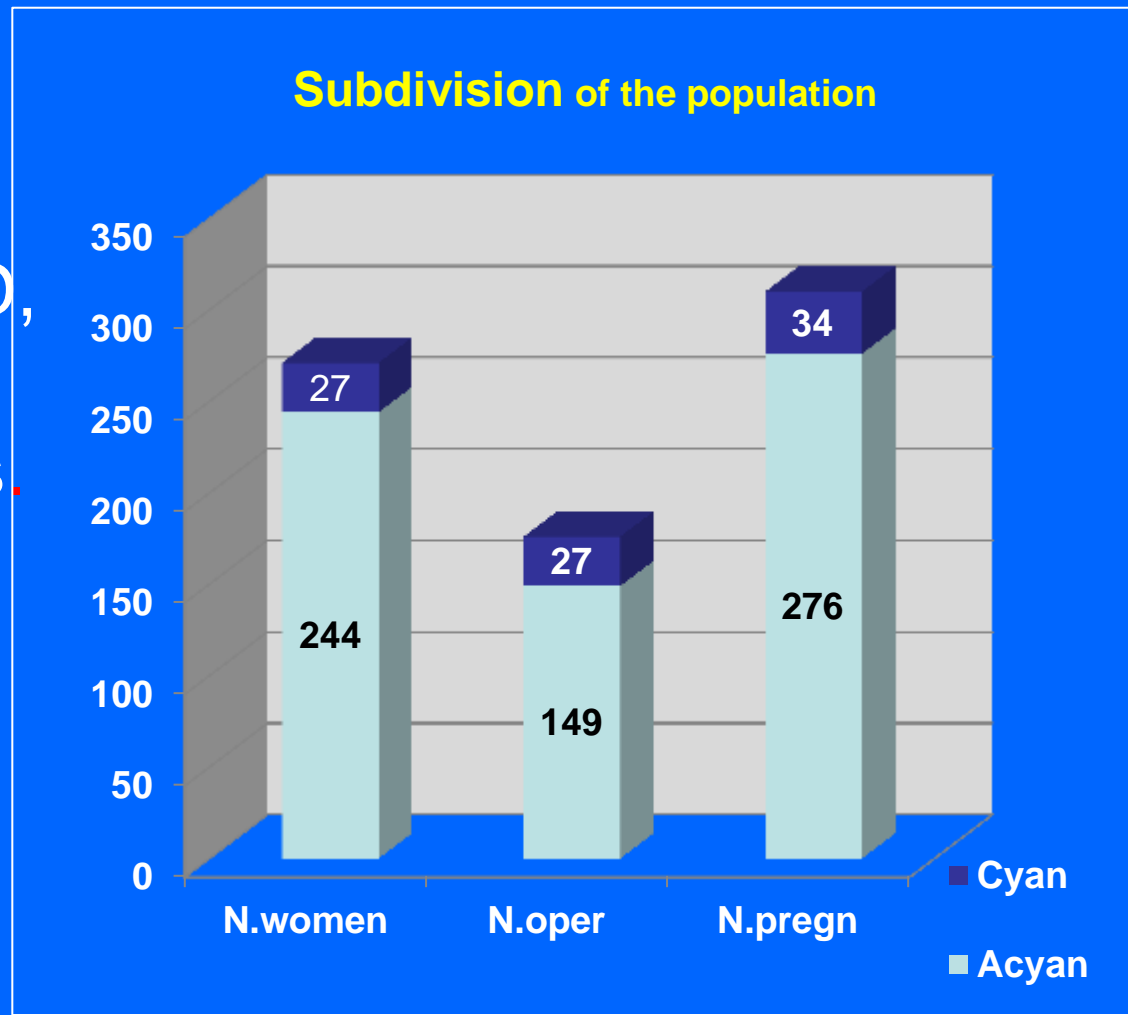
39 women -were followed-up during 2-3 pregnancies

.

Material and methods 2/

27 women, all operated, had cyanotic CHD, in 34 pregnancies,

and 244 women had acyanotic CHD, operated in 149, in 276 pregnancies.

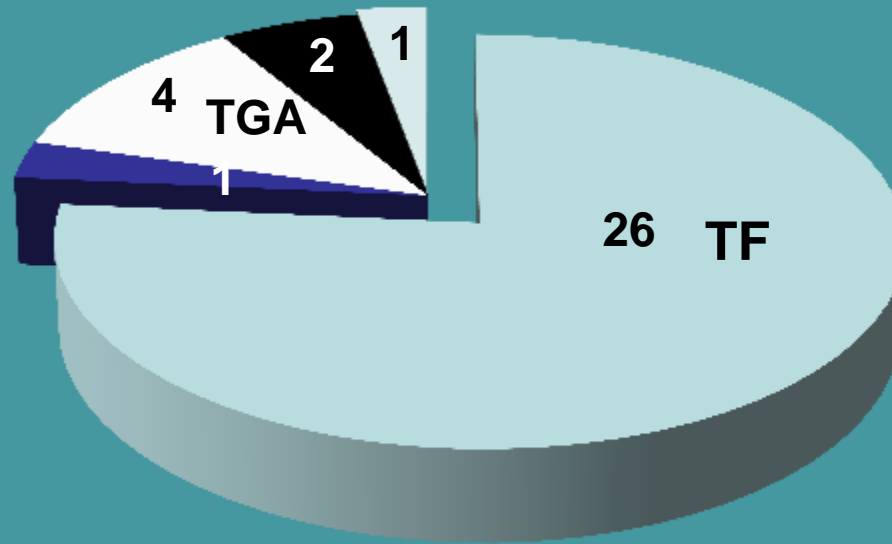


Material and methods 3/

36 women in 38 pregnancies

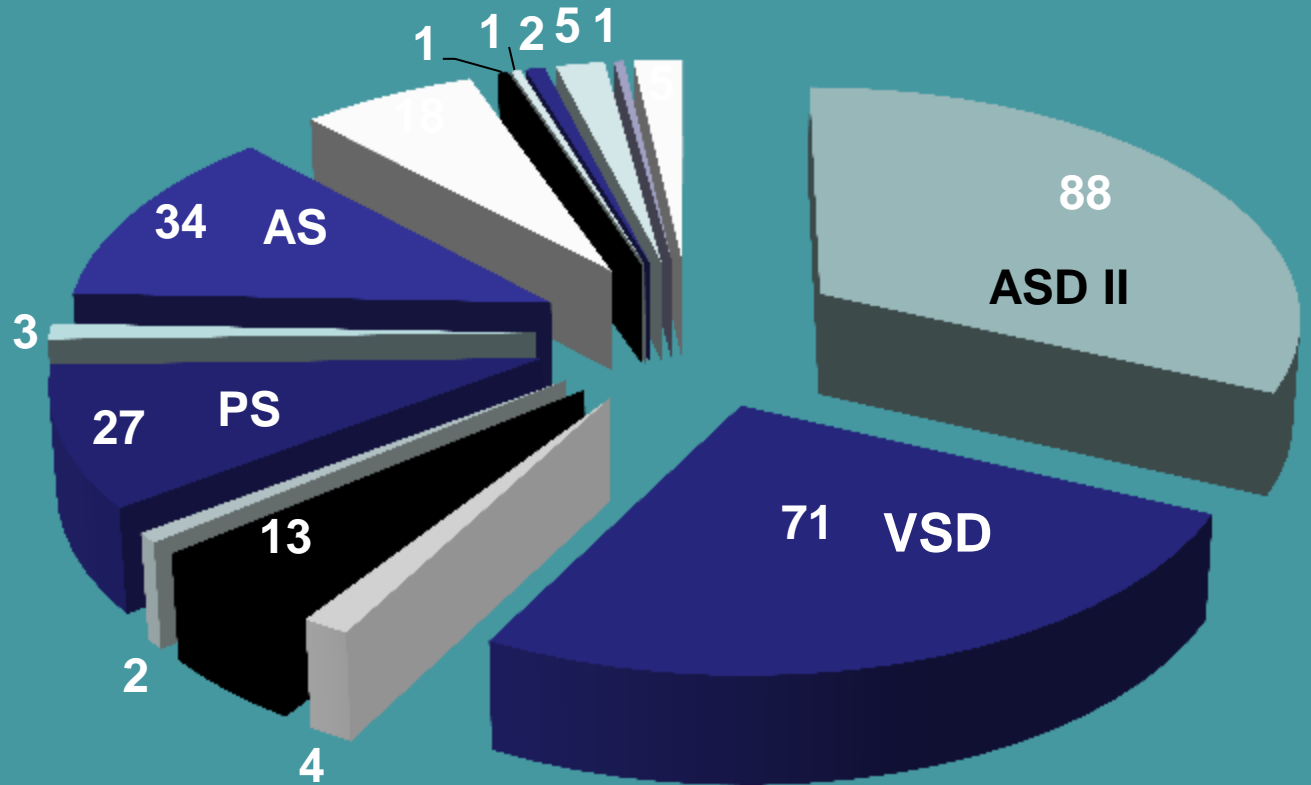
had *multiple familial risk (2-5 relatives)*

Cyan. CHD in mothers



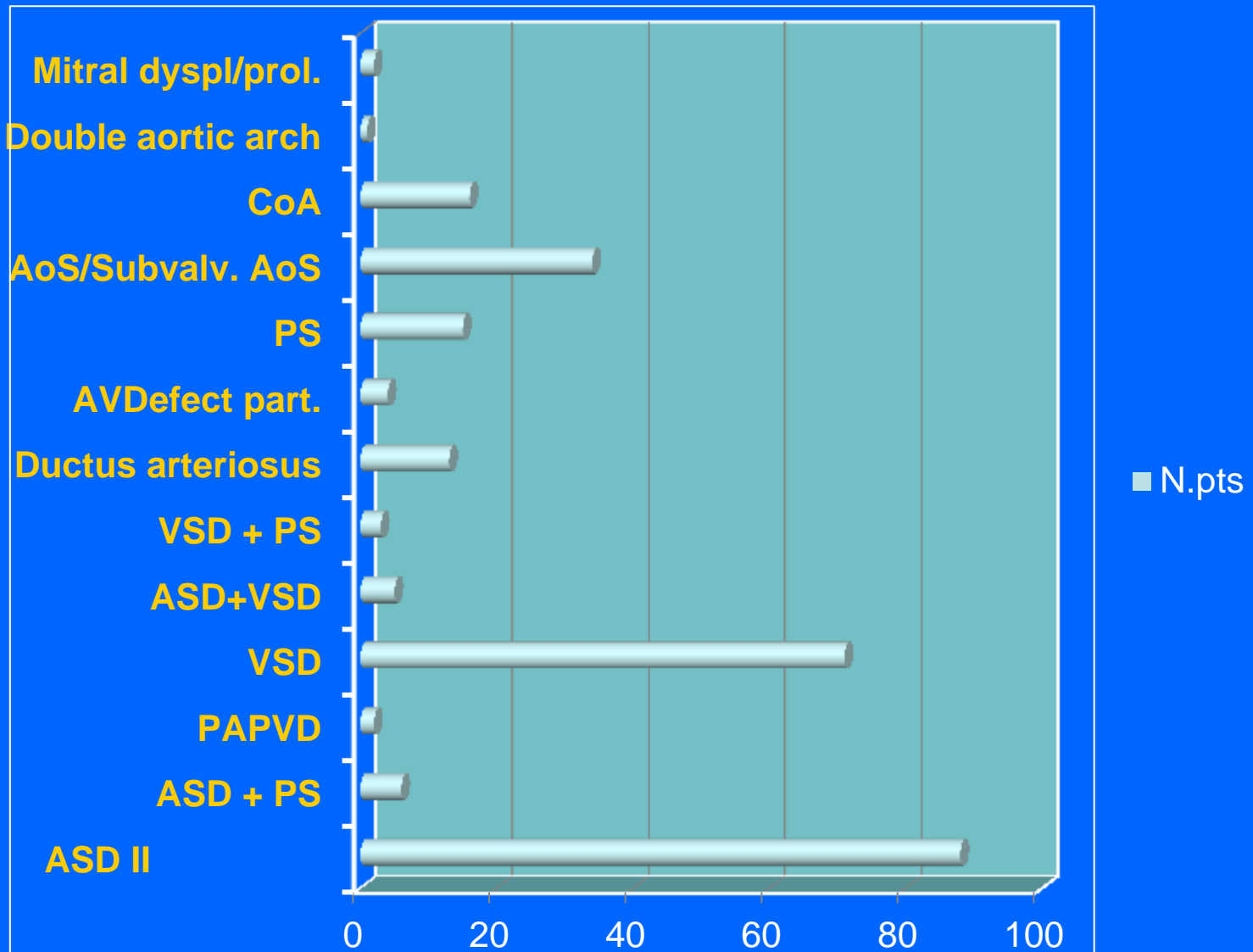
■ TF ■ PAttrVSD ■ TGA ■ TGAc ■ TGAc+Patr+dxc

Acyan. CHD in mothers



- | | | | |
|------------|--------------|----------|---------|
| ■ ASD II | ■ VSD | ■ AVD | ■ PDA |
| ■ ASD+VSD | ■ PS | ■ VSD+PS | ■ AS |
| ■ CoA | ■ UVH | ■ DORV | ■ PAPVD |
| ■ dxcs svi | ■ N-Ebst+ASD | ■ misc | |

Types of acyanotic diseases in mothers



Results:

23 probands had CHD

total recurrence rate 23 / 310 = 7.4%,

1 / 34 pregnancies with cyanotic CHD - 2.9%,

22 / 244 pregnancies with acyanotic CHD - 9 %

Results 2/:

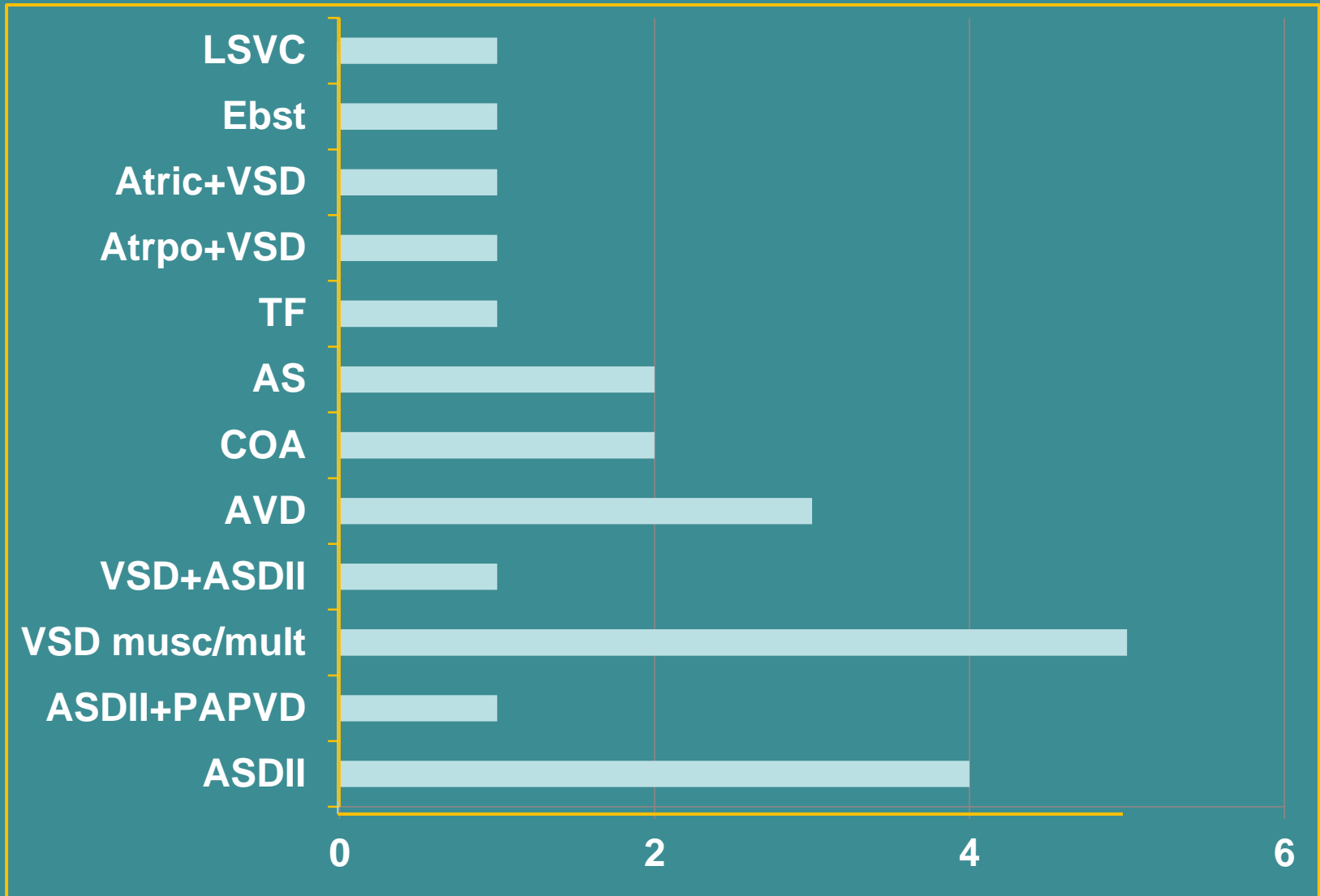
When mother alone was affected,

the recurrence was $21 / 235 = 8.9 \%$

When mother and another relative were affected

the recurrence rate was $2 / 36 = 5.5 \%$

Types of CHD in probands



Results 3/ :

The recurrence was higher in

-ASD 7 / 88 = 7.9 %, N.B ASD considered > 1yr,
diam. >8mm

- VSD (7 / 71= 9.8 %),

- ductus arteriosus (1 / 12=8.3)

and in AVSD (1 / 3).

Concordant lesions occurred in 9 cases,
partially concordant in 7,
discordant in 6 cases.

Recurrence in different types of maternal CHD

Type of CHD	N. preg	N. probands affected	CHD	Recurrence rate
ASD II	88	7	3 ASD II, (1mr) 1 ASD+VSD 1 ASD,PAPVD, PDA, 3 VSD musc	7.9 %
VSD	71	7	3 VSD 2 AVD complex 1 TF, 1 Patr+VSD	9.8 %
AVSD	4	2	1 AVD partial (multiple risk-5x) , 1 Tric.atr+VSD	50 %
PDA	12	1	1 ASD	8.3%
TF	24	1	1 persistent LSVC	4.2 %
Subaort.st/ AS	34	3	1 Ebstein, 2 AS	8.8 %
CoA	17	2	Coa	11.7%

Comparison with the literature

Burn et al., 1998 provided counselling figures for different types of cardiac lesions in siblings and offsprings of cases with CHD,

reporting **a higher recurrence rate for children of affected mothers**

compared to the cases with an affected father (**4.1% and 2.2%**, respectively).

Burn J, Brennan P, Little J, Holloway S, Coffey R, Sommerville J, Dennis NR, Allan L, Arnold R, Deanfield JE, Godman M, Houston A, Keeton B, Oakley C, Scott O, Silove E, Wilkinson J, Pembrey M, Hunter AS. Recurrence risks in offsprings of adults with major heart defects: results from first cohort of British collaborative study. *The Lancet* 1998, 351: 311-316

Comparison with the literature 2/

Higher recurrence rates in the offspring of mothers with CHD with and without surgical treatment (of **16.1%**) were reported by **Whittemore et al. 1982**,

while **Rose et al., 1985** found a **9%** recurrence in parents with specific defects.

Whittemore R, Hobbins JC, Engle MA. Pregnancy and its outcome in women with and without surgical treatment of congenital heart disease. Am J Cardiol 1982; 50: 641-651

Rose V, Gold RJM, Lindsey G, Allen M. A possible increase in the incidence of congenital heart disease among the offsprings of affected parents. J Am Coll Cardiol 1985; 6: 376-382

Recurrence of Congenital Heart Disease in Cases with Familial Risk Screened Prenatally by Echocardiography
V.Fesslova, et al. J. Pregn. 2011, Article ID 368067,
doi:10.1155/2011/368067

Mother alone	250	13	5.2%
<i>Mother + another relative</i>	<i>26</i>	<i>2</i>	<i>7.7%</i>
Father alone	93	7	7.5 %
<i>Father + another relative</i>	<i>19</i>	<i>1</i>	<i>5.3 %</i>

Comparison with the literature 3/

The **preponderance of affected offsprings of mothers with CHD** might be explained by **cytoplasmatic inheritance- transmission of maternal cytopathy**

This mechanism is more evident in some specific maternal cardiac lesions such as AS, CoA, PS, VSD, AVSD.

Mothers are likely to be more vulnerable to teratogens than fathers

Nora JJ, Nora AH. Maternal transmission of congenital heart disease: new recurrence risk figures and the question of cytoplasmic inheritance and vulnerability to teratogens. Am J Cardiol 1987; 59: 459-463

Moon et al. 2008. Primary diagnoses in 921 children

	Birth prevalence per 1,000
1. Hypoplastic Left Heart Syndrome	0.09
2. Univentricular physiology	0.08
3. Tricuspid atresia	0.04
4. Tetralogy of Fallot	0.47
5. Pulmonary atresia with VSD	0.05
6. Pulmonary atresia without VSD	0.05
7. Double outlet right ventricle	0.12
8. Double inlet left ventricle	0.02
9. Truncus arteriosus	0.06
10 TGA	0.26
11 Congenitally corrected TGA	0.03
12 Coarctation of the aorta	0.41
13 Atrioventricular septal defect	0.33
14 Atrial septal defect, type I	0.01
15 Ebstein malformation	0.03
16 Pulmonary valve abnormality	0.79
17 Aortic valve abnormality	0.32
18 Aortic abnormality	0.08
19 Left ventricle outflow tract obstruction	0.07
20 Atrial septal defect, type II	1.46
21 Ventricular septal defect	2.72
22 Mitral valve abnormality	0.26
23 Pulmonary vein abnormality	0.10

Conclusions:

Our data confirm a relevant recurrence of CHD in affected mothers.

This fact has to be taken in account during the prenatal counselling.

