Introduction to the series: pre-natal diagnosis in congenital heart defects

One of the major achievements of pediatric medicine in the last 50 years is the increased understanding of the pathogenetic causal mechanisms of congenital heart defects as well as its treatment, and any resultant sequelae. In particular, for neonates and infants born with congenital heart defects, the progress of the interventional and surgical treatments has allowed a huge increase in the proportion of these children that will reach adult life with a decent quality of life and social integration.

Within the last few decades the pre-natal diagnosis of congenital heart defects has made substantial progresses, allowing the recognition of virtually almost all heart malformations between the 16th and 18th week of pregnancy, with a sensibility over 96% and a specificity close to 100% (1,2).

The pre-natal echocardiographic screening was, by necessity, the first step in the process of pre-natal diagnosis and management of congenital heart defects, and several studies were reported in the literature, focused on the pre-natal screening, with considerations on general and specific issues (3-19).

General non-cardiac issues

In fetuses with congenital heart defects a high incidence of chromosomal abnormalities was always observed (4,7,13), as well as reduced fetal body weight and growth (5,17), associated non-cardiac malformations (13), and presence of situs inversus or heterotaxy (14).

General cardiac issues

The high incidence of complex congenital heart defects observed in the pre-natal cardiac screening was confirmed in the post-natal diagnosis (4-6,8,10,14,16,18).

The pre-natal cardiac screening proved to be useful to detect fetal arrhythmias (10).

The major contribution provided by the availability of pre-natal diagnosis of complex congenital heart defect was the possibility of introducing fetal interventions (9,20-23).

Of course, imaging plaid a vital role in the diagnosis and treatment planning for fetal cardiac abnormalities discovered in utero, with ultrasound as the primary modality for evaluating the fetus due to its spatial and temporal resolutions, widespread availability, and ease-of-use. Nevertheless, there was a growing interest in magnetic resonance imaging as an adjunct diagnostic tool for the fetal heart, brain, lungs, liver, and other organs when ultrasound is limited by maternal obesity, oligohydramnios, multiple gestations, fetal diaphragmatic hernia, or fetal bone in late gestation (24).

Although in the past magnetic resonance imaging in the setting of fetal cardiac interventions has received less attention than interventions for other organ malformations, recent advances in fetal cardiac magnetic resonance technology contributed more significantly to the diagnosis, planning, and monitoring of fetal cardiac interventions (9,22,23). Furthermore, fetal cardiac magnetic resonance provided additional physiologic information on the distribution of the fetal circulation and fetal oxygen transport, helpful in the management of other fetal cardiac conditions in which fetal treatment is under investigation (22).

Overall, the potential impact of pre-natal diagnosis of congenital heart defects included:

- better knowledge of the natural history of the congenital heart defects “in utero”;
- potential pre-natal medical or interventional cardiology therapeutic interventions in the case of diagnosed heart failure, arrhythmias, or malformations with poor neonatal prognosis;
- safer management of the pregnancy itself;
- organization of the peri-natal period in or close to institutions with facilities available for the immediate management in the case of life-threatening heart malformations;
- parental counselling.

In the literature strong disagreements persists about the potential impact of the pre-natal diagnosis on the early and late

outcomes of complex congenital heart defects, despite positive recent reports (9,22,23).

Of course, it has to be taken into account the difficulty of having meaningful inferences from the literature, because of different inclusion criteria, relatively small numbers of patients, different peri-operative managements, different endpoints, and frequently insufficient statistical analysis.

This purpose of this series on "pre-natal diagnosis in congenital heart defects" is to collect and share with the readers of "Translational paediatrics" the current state-of-the-art knowledge on the topic, with the updated information provided by the world experts in this matter.

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