Editorial on Pre-natal Diagnosis in Congenital Heart Defects

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.

Acknowledgments

Funding: None.

Footnote

Provenance and Peer Review: This article was commissioned by the editorial office, Translational Pediatrics for the series “Pre-natal Diagnosis in Congenital Heart Defects”. The article did not undergo external peer review.

Conflicts of Interest: The author has completed the ICMJE uniform disclosure form (available at http://dx.doi.org/10.21037/tp-20-140). The series “Pre-natal Diagnosis in Congenital Heart Defects” was commissioned by the editorial office without any funding or sponsorship. AFC served as the unpaid Guest Editor of the series, and serves as an unpaid editorial board member of Translational Pediatrics from Apr 2020 to Mar 2022. The author has no other conflicts of interest to declare.

Ethical Statement: The author is accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Open Access Statement: This is an Open Access article distributed in accordance with the Creative Commons Attribution-NonCommercial-NoDerivs 4.0 International License (CC BY-NC-ND 4.0), which permits the non-commercial replication and distribution of the article with the strict proviso that no changes or edits are made and the original work is properly cited (including links to both the formal publication through the relevant DOI and the license). See: https://creativecommons.org/licenses/by-nc-nd/4.0/.

References

1. Allan LD, Crawford DC, Chita SK, et al. Prenatal screening for congenital heart disease. Br Med J (Clin Res Ed) 1986;292:1717-9.
2. Bull C. Current and potential impact of fetal diagnosis on prevalence and spectrum of serious congenital heart disease at term in the UK. British Paediatric Cardiac Association. Lancet 1999;354:1242-7.
3. Tworetzky W, McElhinney DB, Reddy VM, et al. Improved surgical outcome after fetal diagnosis of hypoplastic left heart syndrome. Circulation 2001;103:1269-73.
4. McBrien A, Sands A, Craig B, et al. Major congenital heart disease: antenatal detection, patient characteristics and outcomes. J Matern Fetal Neonatal Med 2009;22:101-5.
5. Levey A, Glickstein JS, Kleinman CS, et al. The impact of prenatal diagnosis of complex congenital heart disease on neonatal outcomes. Pediatr Cardiol 2010;31:587-97.
6. McBrien A, Sands A, Craig B, et al. Impact of a regional training program in fetal echocardiography for sonographers on the antenatal detection of major congenital heart disease. Ultrasound Obstet Gynecol 2010;36:279-84.
7. Rychik J, Szewast A, Natarajan S, et al. Perinatal and early surgical outcome for the fetus with hypoplastic left heart syndrome: a 5-year single institutional experience. Ultrasound Obstet Gynecol 2010;36:465-70.
8. Trivedi N, Levy D, Tarsa M, et al. Congenital cardiac anomalies: prenatal readings versus neonatal outcomes. J Ultrasound Med
2146

9. Van Aerschot I, Rosenblatt J, Boudjemline Y. Fetal cardiac interventions: myths and facts. Arch Cardiovasc Dis 2012;105:366-72.
10. Emam SM. High prevalence of complex congenital cardiac anomalies detected by fetal echocardiography in a cohort of Saudi women referred for prenatal assessment. J Egypt Soc Parasitol 2012;42:281-90.
11. McCandless RT, Puchalski MD, Minich LL, et al. Prenatally diagnosed coarctation: a more sinister disease? Pediatr Cardiol 2012;33:1160-4.
12. Enzensberger C, Vogel M, Degenhardt J, et al. Fetal pulmonary venous flow and restrictive foramen ovale in hypoplastic left heart. Ultraschall Med 2012;33:E38-45.
13. Axt-Fiedner R, Enzensberger C, Fass N, et al. Fetal diagnosis of hypoplastic left heart, associations and outcomes in the current era. Ultraschall Med 2012;33:E51-6.
14. Beaton AZ, Pike JI, Stallings C, et al. Predictors of repair and outcome in prenatally diagnosed atroventricular septal defects. J Am Soc Echocardiogr 2013;26:208-16.
15. Natarajan S, Szust A, Tian Z, et al. Right ventricular mechanics in the fetus with hypoplastic left heart syndrome. J Am Soc Echocardiogr 2013;26:515-20.
16. Asplin N, Dellgren A, Conner P. Education in obstetrical ultrasound—an important factor for increasing the prenatal detection of congenital heart disease. Acta Obstet Gynecol Scand 2013;92:804-8.
17. Cnota JF, Hangge PT, Wang Y, et al. Somatic growth trajectory in the fetus with hypoplastic left heart syndrome. Pediatr Res 2013;74:284-9.
18. Oster ME, Kim CH, Kusano AS, et al. A population-based study of the association of prenatal diagnosis with survival rate for infants with congenital heart defects. Am J Cardiol 2014;113:1036-40.
19. Axt-Fiedner R, Tenzer A, Kawecki A, et al. Prenatal assessment of ventriculo-coronary connections and ventricular endocardial fibroelastosis in hypoplastic left heart. Ultraschall Med 2014;35:557-63.
20. Szust A, Tian Z, McCann M, et al. Vasoreactive response to maternal hyperoxygenation in the fetus with hypoplastic left heart syndrome. Circ Cardiovasc Imaging 2010;3:172-8.
21. Co-Vu J, Lopez-Colon D, Vyas HV, et al. Maternal hyperoxygenation: A potential therapy for congenital heart disease in the fetuses? A systematic review of the current literature. Echocardiography 2017;34:1822-33.
22. Marini D, Xu J, Sun L, et al. Current and future role of fetal cardiovascular MRI in the setting of fetal cardiac interventions. Prenat Diagn 2020;40:71-83.
23. Pickard SS, Wong JB, Bucholz EM, et al. Fetal Aortic Valvuloplasty for Evolving Hypoplastic Left Heart Syndrome: A Decision Analysis. Circ Cardiovasc Qual Outcomes 2020;13:e006127.
24. Roy CW, van Amerom JFP, Marini D, et al. Fetal Cardiac MRI: A Review of Technical Advancements. Top Magn Reson Imaging 2019;28:235-44.

Antonio F. Corno
Corno AF. Introduction to the series: pre-natal diagnosis in congenital heart defects. Transl Pediatr 2021;10(8):2144-2147. doi: 10.21037/tp-20-140

View this article at: http://dx.doi.org/10.21037/tp-20-140