Review Article on Pre-natal Diagnosis in Congenital Heart Defects

Ethical issues after pre-natal diagnosis

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Abstract: Following pre-natal diagnosis of congenital heart defect parents and family face a dramatic psychological crisis because of their state of shock, contradictory information available on potential outcomes, limited availability of time for decisions and for autonomous choices. Counselling the parents can present additional difficulties due to influence of education, cultural and religious background, individual cognitive and emotional processes, and cross-cultural patient care is a challenging issue for the caregivers. Type and quality of messages transmitted by the caregivers determine the counselling process, with the risk of misunderstandings particularly high with reduced available evidence, or with different outcomes accordingly with the various alternatives of treatment. Since the introduction of pre-natal diagnosis for congenital abnormality, interruption of pregnancy became available on these grounds in many Western countries, and the numbers of babies born with congenital heart defects has declined significantly despite concomitant advances in treatment options and outcomes. Detailed and objective information, with all available options, should be provided after pre-natal diagnosis of congenital heart defect. 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. For congenital heart defects the progress of surgical treatments allowed a huge proportion of these children to reach adult life with a decent quality of life and social integration. Therefore, must be a considerable concern that universal pre-natal diagnosis widespread pregnancy interruption may obviate those gains. A reduction in the post-natal population undergoing treatment may have a significantly deleterious effect on the expertise of the caregivers, producing a reduction in outcome quality. With all respect for the parental choices and the obligations to follow the national laws, is necessary to remark that our society is genuinely ambivalent.

Keywords: Congenital heart defects; counselling; ethics; interruption of pregnancy; pre-natal diagnosis

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Introduction

Pre-natal diagnosis of congenital heart defects relies on several elements, such as equipment, specific expertise, local structure and infra-structure, all factors variable among countries and over time, as services and policies evolve, and therefore affecting rate and impact of fetal screening (1-3). In any case, the incidence of pre-natal diagnosis is influencing the trends and outcomes of congenital heart defects, and several additional factors have to be included in the evaluation of the outcomes: associated non-cardiac co-morbidities, pregnancy management and delivery planning, long-term neurodevelopmental consequences, resources utilization and costs/benefits issues.

Various reports have taken in consideration all various points listed above, and the unavoidable relationships among them (4-17); their correlation is very strict, even if it is impossible to quantify the sensitivity/specificity of the fetal screening or the subsequent outcome (18-29).

The matter becomes even more delicate when considering all potential ethical implications of fetal finding of heart malformation, particularly regarding parents

counselling and interruption of pregnancy versus treatment.

When dealing with ethical issues following pre-natal diagnosis, it should be advisable to distinguish between complex and simple cardiac malformations.

Complex congenital heart defects

The largest group of complex congenital heart defects includes all malformations with "functionally" univentricular heart, regardless their morphologic characteristics.

The results recently achieved in univentricular hearts have substantially improved (30-34), however the definition of "intractable" cardiac malformation remains attached to the diagnosis of univentricular heart, due to the questionable late outcomes (12,24). "Intractable" (12) or "uncorrectable" (24) defines a cardiac malformation not suitable to bi-ventricular repair, but only to multi-staged complex palliations towards Fontan circulation, or heart transplant. Avoidance of treatment and compassionate care remains the only available option in these patients (12,24).

Evident discrepancies exist in the literature about the various outcomes obtained with heart transplantation after the different stages of surgical management of univentricular hearts (35-38).

After fetal diagnosis of univentricular heart (4-6,8,14,15,19,21,22,24,25,28,39), the decision-making process has been oriented towards for pregnancy termination or compassionate care in 8% of cases (19), through 16% (5,6), 30 (14,15), 33% (4), reaching 47% (28), due to one or more of the following reasons: low hospital survival, impaired quality of life, child suffering (8,14), stress for the entire family, including siblings (8,24), association of genetic abnormalities and/or severe non-cardiac defects (28).

After fetal diagnosis of complex congenital heart defect a comprehensive multi-specialties pre-natal screening, including chromosomes assessment, complete intra- and extra-cardiac evaluation, to provide the family with an appropriate explanation of the expected future (5-7,14-17,19,21,24,29,39). The parents and their families have the right to receive all data useful to prepare and support them (24).

Following pre-natal diagnosis of congenital heart defect parents and family face a dramatic psychological crisis because of their state of shock, contradictory information available on potential outcomes, limited availability of time for decisions and for autonomous choices (8,39). Furthermore, not all the parents wish to perform autonomous choices due to family or other societal pressures (8).

Counselling the parents, already complicated by the communications between healthcare providers and families, can present additional difficulties due to education, cultural and religious background, individual cognitive and emotional processes (9,10,24,26,39). Cross-cultural patient care is a challenging issue for the caregivers: dealing with families rejecting medical treatments because of their educational and/or religious backgrounds requires an open mind to successfully propose a medical plan (10,26,39).

Nowadays many families can become aware of the best outcomes of the top hospitals, while the same excellence not necessarily can be achieved by their local services. At this point it can be quite uncomfortable for a doctor to explain to prospective parents that the local results for complex congenital heart defects may be less satisfactory than the gold standards reported on internet, and this can also skew subsequent discussions (39).

Quite familiar is the notion of how influent are the modalities of communications transmitted by the physicians during the counselling process, as well as the perceiving mode of the parents. While it is not surprising that optimistic surgeons are capable of attracting the parents approval for surgical approach, totally unexpected is the negative reaction not infrequently shown by families after the advice received by physicians (16,17,22-24,29,39).

The belief of caregivers can range between extreme optimism on the possible long-term outcomes and advice to seriously take the possibility of abortion into consideration, with obvious influence on the decisions of the parents (11,20,22,25,27,39).

Fully focused attention should be given to the sensitivity of each case, particularly considering that within each couple of parents and family there are various degrees of capability to accept a poor quality of life.

The suggestions made by the doctors are generally on line with the goal of maximizing the possibilities of achieving the best outcomes, without always providing details about the chances of negative outcomes; furthermore, their knowledge on expected outcomes are biased by their previous clinical experiences (11,39).

The risk of misunderstandings in the transmission of information is particularly high with reduced available evidence, or with different treatments available with variable reported results (11). Finally, due to the difficulty of pre-natal screening in complex congenital heart defects and the large variation in the reported results, it can be extremely challenging for the physicians to maintain a scientific and

Table 1 Results of the "Audience Response Survey"

Options	Patient	Family member
Norwood	73%	26%
Heart transplant	0%	1%
Compassionate care	6%	5%
Pregnancy interruption	21%	68%

objective approach untainted by their personal feelings and opinions (39).

Sometimes doctors are asked the following question: "What if it were your child?" (13). At the first joint meeting of the European Association Congenital Heart Surgeons and the "Congenital Heart Surgeons Society" of North America, the "Audience Response Survey" tried answering the above question interrogating experienced professionals in congenital cardiac surgery, with their answers reported in *Table 1* (18,39).

The congenital heart surgeons participating to the survey provided striking different answers between the option suggested for a pre-natal diagnosis of hypoplastic left heart syndrome in a patient versus a hypothetical close member of their family (18,39).

In the all parents, families, and all the health professional involved, the fetal diagnosis of complex congenital cardiac malformation is always followed by important consequences (39).

The counselling of the parents and the entire decision-making process is very challenging due to the large variability of malformations with different clinical presentation, association of genetic abnormalities, presence of other intra- or extra non-cardiac defects, and the reported poor outcomes. Evidence provided by studies on the managements used nowadays should give adequate knowledge to facilitate the parents counselling after pre-natal diagnosis of complex congenital heart defects.

Simple congenital heart defects

The ethical considerations become even more controversial in the presence of fetal diagnosis of simple cardiac malformations.

Since the introduction of pre-natal diagnosis for congenital abnormality, interruption of pregnancy has become available on these grounds in many Western countries. As a consequence, in some of these countries, the numbers of babies born with simple congenital heart disease has declined significantly despite concomitant advances in treatment options and outcomes, as reported on the database of the European Congenital Heart Surgery Association (https://echsacongenitaldb.org) and of the North America STS Congenital Heart surgery database (www.sts.org/registries-research-center/sts-national-database/sts-congenital-heart-surgery-database). This is the result of policies applied and reported, with debatable decision-making processes after fetal diagnosis of simple congenital heart defects (40).

The discrepancy between pre- and post-natal diagnosis of congenital heart defects occurs with a relatively low incidence but, when present, may change both the treatment both immediately after birth and the long-term management (41). Therefore, clinicians should show some caution during pre-natal counselling regarding the planned treatment based on the fetal echocardiogram, particularly when interruption of pregnancy is under consideration (41).

The reported study (40) reflects the experience in a country with a level of socio-economic and health organization among the most advanced in the world, with a particular social demographic which may be quite different even from many other high-income countries. The facts presented may well pose significant questions to be accepted or indeed understood in other health care systems with completely different levels of socio-economic status and health care, and among other cultures, religions, educations, and ethnicities (9,10,24,26,39).

In particular, interruption of pregnancy has been offered for pre-natal diagnosis of congenital heart defects not as complex as the "functionally" univentricular hearts, but such as aortic coarctation, atrioventricular septal defect, tetralogy of Fallot, double outlet right ventricle, truncus arteriosus, transposition of the great arteries, and aortic arch interruption (40), all malformations with recognized low STAT risk stratification (42) and excellent outcomes, with reported mortality lower than 5% (43).

When the decision-making has to take in account scientific as well as ethical aspects, the process becomes complicated by different perspectives.

Since the natural history of some congenital heart malformations may results in spontaneous abortion, before the availability of this information families and caregivers have to choose between medical management and abortion. The availability of termination of pregnancy completely influences the approach of physicians in these situations (44).

In a genuinely ambivalent society praising the option that

in the near future doctors will suppress patients even with very simple congenital heart defects instead than offering them treatment, the medical profession has to deal with the opinion that some life is not worthy to be lived (44).

To be a doctor is not privilege: it is a commitment, with patients, families and community, requiring every possible effort to help the diseased people, and to push the boundaries of knowledge, with a work ethic far beyond that of research alone (45).

Interruption eliminates the fetus with a malformation, it doesn't cure the underlying malformation. Thus, interruption of pregnancy doesn't address the defect, and doesn't care the affected individuals (45). Our responsibility is to develop a science without cultural stagnation, but to encourage research, and to involve not only patients and families, health organizations, and society at large (45).

Pre-natal diagnosis of congenital heart defects produces an ethical dilemma for both parents and physician. The right to decide should be left to the parents, while the caregivers have to support them with nondirective counselling, with the information necessary for parental decision (39,46).

When individuals have been asked about a choice for their own life, the preference was always for some life, even if short and with poor quality, than no life at all (39,46).

Full information about a fetal congenital heart defects, particularly in the case of simple defects, has to be provided with all available data obtainable with the currently available treatments (42,43). The way the family is perceiving the information received is certainly influenced by the modality used by the counsellor to transmit the information (39,46).

Conclusions

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 infants with congenital heart defects, the progress of the surgical treatment 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. Therefore, must be a considerable concern that universal pre-natal diagnosis widespread pregnancy interruption may obviate those gains. Furthermore, such a reduction in the post-natal population undergoing treatment may have a significantly deleterious effect on the experience and expertise of those involved in the care, which might conceivably produce a reduction in outcome quality which cannot be considered positively.

With all respect for the parental choices and the obligations to follow the national laws, however the feeling is the necessity to remark that our society is genuinely ambivalent.

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