

## CASE REPORT

# Growing up with congenital heart disease: impact on the daily life quality from the perspective of a mother.

### Author

Francesca Maria Susin

Cardiovascular Fluid Dynamics Laboratory, Department of Civil, Environmental and Architectural Engineering – University of Padova, Italy

Email: [francescamaria.susin@unipd.it](mailto:francescamaria.susin@unipd.it)

### Abstract

Congenital heart diseases (CHDs) are the most common and severe anomalies at birth, with over 1 million newborns affected every year in the world. Thanks to the advances in surgical treatment and medical care over the past few decades, the majority of them now reach adulthood and have an acceptable life quality expectancy.

However, growing up with a congenital heart disease is far from being an easy task, although satisfactorily managed from the strictly medical point of view. Psychosocial, educational, financial issues, among the others, highly impact on the daily life of both the patients and their families, and a comprehensive and global knowledge of what living with CHDs condition does mean still has to be built in both the scientific and social community.

The present report is based on the direct experience of the author, who is mother of a 14 years old boy born with a severe heart defect. Key points on CHD-life management as experienced by the author and her son and family in the past fourteen years are proposed and discussed following a ‘from inside’ perspective. Suggestions aimed at soliciting the design and delivery of ad-hoc support programs is also given. The importance of a multidisciplinary, global approach capable of joining all the scientific, professional and socio-political parts is highlighted.

## Introduction

Congenital heart diseases affect about one over one hundred of newborns<sup>1</sup>. A large variety of malformations, either single or combined, exist, from the simplest to the most complex. The large majority of CHD can be effectively treated, and about 85% of patients now live into adulthood<sup>2</sup>. Such an important result has been obtained thanks to continuous progress in multidisciplinary research in the past three-four decades, spanning from cardiovascular physiology and surgery, to biology, pharmacology, engineering and technological sciences. However, if on one side a favourable prognosis is the obvious priority goal on both the medical and the social side, on the other that goal brings a number of different problematic situations that involve the patient and her/his family<sup>3</sup>. Indeed, as soon as CHD is diagnosed, either pre- or post-natally, not only the foetus/child becomes a patient but all her/his family does, although on different planes. From that moment on, they are all called to face the fact that daily life will also mean hard battles, fears, uncertainties, and that the conscious and informed acceptance of facts will be the elective, if not the only, way to fully live rather than to merely survive. That is the fundamental reason why CHD patients and their families require the support of specialists in the medical, psychological, social, educational and political areas. That is, an integrated treatment of CHD patients and families is required.

Researchers' awareness of the complexity of a 'CHD daily life' is increasing since the past twenty years. The recent review given in<sup>4</sup> accurately describes and analyses "*perspectives and experiences of*" a large sample of "*children and young people with CHD, to increase clinician responsiveness to patient priorities and needs*". Among the other

challenges reported in that work, it is worthwhile recalling here lifestyle restrictions, discrimination among peers, uncertainty about the future. Similar findings are outlined by<sup>3</sup>, with specific focus on limitation and frustration due to physical impairment, exclusion by others mainly due to lack of empathy, bullying, and the need of improving what people understand about the CHD condition ("*People panic*", one of the interviewed CHD adolescents says). The crucial need of accompanying adolescent CHD patients towards the still unknown world of adult health care in an efficient and gentle way is the core of the investigation performed by<sup>5</sup>. The study developed by<sup>6</sup> examines personality as a significant determinant of the ability of gaining a satisfactory adaptation to a chronic diseased status as CHD is; in a different study, the same authors show the correlation between personality and CHD outcome, irrespective of CHD complexity<sup>7</sup>. Some investigations concern parents and whole family experiences. The review given in<sup>8</sup> highlights that the incidence of anxiety, depression and stress in parents of CHD children is much larger than in the general population, also associated with a sort of 'rollercoaster emotional trend'. Reported stressors are related to surgical interventions, hospitalization, decisions about treatments, financial issues associated to job change/loss and/or high medical and assistance costs, side effects on siblings of CHD patients like worsening of school performance. Interventions proposed in the literature to mitigate parents' psychological symptoms and social isolation are also reviewed in the above work. However, the authors conclude that "*research is needed to explore parents' experiences and expectations*". Basic needs of parents have not yet been addressed although crucial for the wellbeing of the whole family, as observed for example by<sup>9</sup> who report that "*some parents wished they*

would have had more help with defining normality". The present report is hence aimed at giving some suggestions about important issues on the topic, to solicit the attention of experts and the awareness of all subjects involved in congenital heart disease patients care. All raised issues come from the fourteen years' experience 'on field' of the author, who is mother of a congenital heart disease patient. The author's belief is that a comprehensive and conscious knowledge of what CHD life means, which is paramount to help patients and their families to gain a good life quality, must rely on direct sources of information. Moreover, such a knowledge can only come from the adoption a multidisciplinary and multi-subject approach, due to the wide spectrum nature of CHD-related issues and the strong interactions among them.

A description of the author's son and family case is first given to illustrate the essential of his clinical history and their daily life since his birth. Some key issues are then proposed and discussed, and some brief conclusions are drawn.

### **Case report.**

The author is the mother of a 14 years old boy (herein after called G.), born with severe aortic valve stenosis post-natally diagnosed. G. was surgically treated with the Ross procedure at the age of 2 months. ECMO was maintained for six days after surgery due to biventricular failure. Acceptable, but not full, heart function was slowly recovered in more than one year. Multiple trans-catheter procedures were performed from 1 year of age to both straight and enlarge the artificial pulmonary conduit. At the age of 13 years and 10 months, i.e. when an almost adult body size was reached, a trans-catheter prosthetic valve was implanted in the pulmonary site as a bridge to the surgical

substitution of the original artificial valve, planned for the future. Although G. recovered quite satisfactory hemodynamic conditions after surgical and trans-catheter procedures, nevertheless his growth steps were delayed with respect to peers until 12 years of age. Undertaking multiple sporting activities was allowed, but at a recreational level only and avoiding any kind of competition. G. regularly attends school; however, organization, planning and completion of homework require a professional support due to mild dyslexia and attention-deficit disorder. His social life and friendship relationships are in the norm. The family comprises the two parents and one sibling, who was 5 years old at G. birth.

The diagnosis of G. congenital heart defect was a shock for his parents and relatives. The whole family was strongly affected by the practical and psycho-emotional consequences of the CHD condition management in both routine daily life and periods of high stress levels like surgeries and hospitalizations. Psychological therapies, support groups, yoga and mindfulness practices, ad-hoc educational programs, professional nurse counselling and assistance from a private paediatric cardiologist were some of the strategies adopted during years by G. parents to help their children and/or themselves in coping with anxiety, fears, and problematic situations. The associated economic burden significantly impacted the family financial situation. Grandparents' contribution to CHD related expenses was also required.

### **Discussion**

Some key issues in CHD condition management are here proposed and discussed. They all come from the direct experience of the author and her family.

*Medical and nursing care.* Raising a child means also learning to recognise the signals that not everything is ok. For CHD children parents, the latent thought that something is wrong with her/his cardiovascular condition is always present, even when usual and usually trivial facts like lack of appetite occur. Hence, distinguishing between what is 'normal' and what is not is crucial for a good quality daily life of the whole family, for example to avoid needless trips to ER or, on the opposite, to not underestimate significant symptoms. A reference subject with specific expertise in paediatric cardiology can be very supportive in helping parents to become confident in their child's daily care. The presence of such an experienced professional should be guaranteed as a fundamental service for at least the first three years of life of the CHD child.

On the clinical side, the formation of care units specifically devoted to GUCH (Grown Up with Congenital Heart defects) patients is undelayable. Continuous interaction among the various professionals is essential in that case, as well as the deep and detailed knowledge of the patient's clinical history. It is the author's opinion that the peculiar nature of any CHD and of its development in years deserves a 'Patient-Specific Medicine'.

*Psychological support.* CHD diagnosis usually traumatizes parents and possible siblings. Specific psychological support is required to help them accepting in deep the chronic nature of CHDs, and focusing on potentialities rather than on difficulties/limits to gain a fruitful life. A holistic perspective able to conjugate conventional and 'alternative' approaches might be of great help. Practices like, for example, yoga, meditation and mindfulness can act as effective de-stressors. Moreover, they can be adopted to train CHD patients in gaining full confidence in their body

responses to either physical and mental requests since the young age. Scientific research is widely confirming the benefits of the above body-mind practices in general well-being as well as in pain and related disorders relief in diseased patients<sup>10, 11</sup>. Randomized controlled trials on patients with diagnosed cardiac disease also give favourable results<sup>12</sup>. Specific studies focused on CHD population might provide useful results too and should be hence highly encouraged.

Support groups can also be of great help for parents in recovering 'the sense of normality' in daily life. Being among peers is essential to feel less lonely and isolated, share openly feelings and experiences, build up confidence in one's own perspective, improving skills to cope with challenges, exchanging practical information about social, educational, economic resources. A professional facilitator trained in CHD issues is highly recommended to make the group as incisive as possible. For what young CHD patients are concerned, it is the author's experience that support groups mainly based on sport and recreational activities have highly positive effects on both the psychological and physical health. Athletic programmes specifically conceived by sports medicine and cardiology professionals help the children and the adolescents to fully and safely live their physical potential, avoiding imaginary limits and respecting the real ones.

Finally, professional psychologists with specific skills in CHD issues are required. Parents need support along the journey since a number of demanding question arise from their growing child, and answers can highly impact on her/his development. However, it is worthwhile pointing out that neither all the questions can be answered by the parents, nor they must be. The passage from childhood to adult age hence requires dedicated professional

support to CHD patients, when needed, also to facilitate the achievement of autonomy.

*Social impact.* People is unaware of what congenital heart disease means. Moreover, they often fail to perceive the real condition of a CHD patient since she/he is apparently completely healthy. In that sense, the disability related to congenital heart disease should be termed 'hidden disability'. Behaviours due to physical limits, for example, are often seen like signs of laziness, or as consequences of excessive parents' protection. Fears that a CHD child may experience as she/he develops rational awareness of her/his body signals, even when benign, are often misunderstood and classified as 'dramatics'. Feelings of being different from the majority and emargination from peers may arise as a result. Another important social issue concerns the poor empathic understanding that parents, mothers especially, encounter in their daily life<sup>13</sup>. Other family members, friends, colleagues, and generic 'others' not seldom minimize the psycho-physical burden associated with raising a child with a congenital heart defect. As a result both practical and emotional support often lacks. Social isolation may even occur, undertaken by either the mother (father) or the outside world. Specific publicity campaigns and educational meetings can promote the spreading of awareness on CHDs. Reinforcement of emotional intelligence at the social level and enhancement of an inclusive social perspective can also give great benefits.

*Educational issues.* An increasing number of scientific investigations reports that among CHD patients '*many will have impaired neurodevelopmental outcome across a wide spectrum of domains*'<sup>14</sup>. For what the potential impact on school performance is concerned, it is recognized that '*children with CHD display a neuropsychological profile with mainly mild*

*motor deficits and subtle difficulties with language tasks. Attention/executive functioning and memory also appear involved but to a lesser degree*'<sup>15</sup>. For complex CHDs surgically treated, '*children experience greater executive functioning difficulties than healthy peers, with metacognition particularly impacted and particularly relevant for day-to-day school performance. Especially in older children, clinicians should watch for metacognitive deficits, such as problems with organization, planning, self-monitoring, and follow-through on tasks*'<sup>16</sup>. However, in the author's experience, the above reported scientific evidences are almost completely ignored by school's professionals. As a consequence, ad-hoc treatments and/or educational plans are often undertaken with years of delay, when not at all, and usually only after persistent requests coming from parents. Training programs devoted to teachers' education in the area of CHD related neurodevelopmental issues are urgent.

*Financial issues.* Surgical procedures, hospitalization, and cardiologic follow-up related to the CHD condition are free of charge in the author's country. Nevertheless, the disease often has major negative effects on the family's financial situation. Increased expenses (e.g., travels to the health care center, parents accomodation during hospitalization, day-care for siblings, professional psychological support, extra medical assistance) and lost income due to job reduction or change for one or both of the parents are the two main causes of financial burden. Moreover, worries about the work ability of their child once adult and planning for her/his financial future usually afflict the parents. Accurate and extended estimation of CHDs related direct and indirect costs is required, as a first step to solicit policies of financial support especially for low

socioeconomic status families. Moreover, specific training on CHDs and related impairments must be encouraged for medical committees deputed to CHDs childrens' disability evaluation. It is the author belief that such a kind of interventions can highly contribute in reducing the overall socioeconomic impact of congenital heart diseases.

*Scientific research advancements.* Diagnosis and treatment of cardiovascular diseases can greatly benefit from multidisciplinary research. In particular, a large amount of work has been done by research groups that join cardiologists, heart surgeons and engineers in the past twenty years. Improved medical imaging techniques and supercomputers are among the factors that have promoted innovative investigations. For example, in-vitro and in-silico fluid dynamics models of blood circulation have been built, from the simplest to the most complex, and have proved their reliability as tools for the assessment of diseases<sup>17,18</sup>, the development of safe and effective prosthetic devices<sup>19,20</sup>, and the presurgical planning<sup>21</sup>. The belief of the author of this report, who has a more than ten-year's experience in the cardiovascular fluid dynamics field, is that fluid dynamics modelling shows its full potential when applied to congenital heart defects, which often occur with complex and/or patient-specific configurations that require ad-hoc analysis and detailed description<sup>22,23</sup>. Three dimensional printing is a further innovative technological tool that can make the difference in CHDs surgical treatment. Physical replica of the anatomical district that requires correction, in fact, facilitate a deeper and immediate understanding of the patient lesion and can thus play an important role in orienting surgical decisions<sup>24</sup>. Public and private funding of multidisciplinary research in the CHDs area is

now paramount to achieve new goals, since many questions are still unanswered (e.g. how hemodynamic, mechanical and biological factors interact in Fontan circulation up to its failure). Moreover, dissemination of research results and their positive socioeconomic impact has to be promoted among the scientific community and the administrative and political world, in particular to facilitate the adoption of innovative technological tools in healthcare.

## **Conclusion**

Congenital heart disease patients and their families go through challenging experiences that may highly impact on the quality of their daily life. Despite a large amount of literature on the topic exists, to the author's knowledge no comprehensive report from the perspective of a mother, as the author herself is, has been proposed yet. The 'from inside' perspective has the advantage of the direct knowledge of the entire spectrum of events that occur when caring a child with a cardiovascular defect, from emergency to routine and at all different levels. This allows the author to identify the psycho-emotional wellbeing as the 'fil rouge' that drives the life quality of the family members. The main limit of the 'from inside' perspective is the lack of distance, which may affect objectivity. However, the majority of the issues proposed in this report have been discussed by the author with a large number of parents of CHDs patients. Similar experiences have been reported by all the parents, and a wide consensus about the relevance of the above issues has emerged. Moreover, the author finds significant correspondence between her own experience and what reported in the literature.

CHDs related issues are reported in the discussion section as a list. This choice has been adopted for sake of clarity. However, all

these issues are part of a single reality and have to be read as strongly interconnected. For example, psychological stress of parents may require professional support with an impact on the financial condition of the family, and may cause social isolation to all family members, which may act as a further stressor. Indeed, close and continuous interaction between CHDs related issues and between involved subjects as well (the patient, the parents, the siblings) is the reason why any particular issue has to be addressed by following an integrated approach. Cardiologists, heart surgeons, paediatricians, nursing professionals, psychologists, neuroscientists, sport doctors, should all join in multidisciplinary teams devoted to healthcare programs tailored on the patient. Moreover, a global perspective has to be adopted to plan effective and comprehensive strategies for CHDs patients and family overall support. Pedagogists, health economists, communications experts, policy-makers and

administrators, among the others, are also fundamental to accomplish the task. The task is urgent, as the number of congenital heart defect patients increases at the rate of about 1.3 millions of newborns per year worldwide. This also means that about three times that number of people will be exposed to some kind of stress sooner or later, with enormous socioeconomic impact. Hence, institutional coordination is paramount to delineate appropriate interventions and provide quality services.

Finally, ethical reasons should also play a major role in designing and delivering ad-hoc support programs for patients and caregivers. In fact, if on one side it is true that in the mid- or long-term it is possible to adapt successfully to living with a CHD condition, it is also true that adaptation comes at the expenses of an incredible amount of physical, mental and emotional resources, often experiencing prolonged loneliness.

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