The care of adult patients with congenital heart defects: a new challenge

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The aim of the present article was to point out some of the more common challenges and needs for adult patients with congenital heart defects. We may reasonably calculate a population approximating 80 000 to 100 000 patients in Italy. The profile of this patient population will change over the next few decades. Not all congenital heart defects require the same level of expertise; for this reason an integrated national service is required. Ideally, specialist units should be established in appropriate geographic areas; complex patients need to be grouped according to the expertise, experience and management they require. Less specialized regional centers and outpatient clinics in interconnected districts with GUCH units should be created. Specialist units should accept responsibility for educating the profession, training the specialists, and sharing particular skills between each other.

The debate on this subject is far from over; we wish to contribute to and stimulate the discussion.

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Advances in the medical and surgical management of congenital heart disease have revolutionized the prognosis of infants and children with cardiac defects so that an increasing number of patients reach adolescence and adult life, even those with complex defects. Recent data suggest that the number of adults with congenital heart disease (ACHD), whether repaired or not, approaches the number of children with the disorder[1,2]. A cure is rarely achieved and ongoing surveillance and management in conjunction with specialists in this highly specialized field is mandatory to provide optimal care for patients. While pediatric cardiology as a subspecialty is well established, it was only recently that the need for specialized units for ACHD has been recognized[3].

Current population and composition

Few data are currently available on the size and composition of the population of ACHD in Italy. In Canada, it is estimated that the number of survivors with ACHD will increase from 94 000 in 1996 to 124 000 by the end of 2006[1].

A recent English study[2] reviewed all births in one health region (Newcastle) between 1985 and 1994, and noted 1942 cases of congenital heart defects in a population of 377 310 live births (incidence of 5.2 per 1000). Of these newborn, 1514 were predicted to survive > 16 years. Also, an estimated 784 would require follow-up in adult life. These figures predict the need for the follow-up of adults with congenital heart defects, for > 200 cases per 100 000 live births, or 1600 cases every year in the UK (assuming a population of 50 million). Using a similar model, we estimate a population of about 60 000 to 80 000 patients with 1200 to 1800 new cases every year requiring follow-up in Italy.

The existence of a large number of individual lesions and of a variety of surgical treatments highlights the importance of a broad system for classification. If we classify lesions on the basis of their complexity, it is possible to identify mild, moderate, and severe categories (Tables I-III)[4].

The profile of this patient population will change over the next few decades, not only because of advancing age, but also owing to the improved survival of patients. Many adult survivors will have different

Table I. Types of congenital heart defects of great complexity: these patients should be seen regularly at adult congenital heart disease centers.

Conduits, valved or non-valved
Cyanotic congenital heart (all forms)
Double outlet ventricles
Fontan procedures
Mitral atresia
Single ventricle
Pulmonary atresia
Pulmonary vascular obstructive disease
Transposition of the great arteries
Tricuspid atresia
Truncus arteriosus
Eisenmenger syndrome

From Connelly et al.[4], modified.

Table II. Types of congenital heart defects of moderate severity: these patients should be seen periodically at regional GUCH units.

Aorto-left ventricular fistulae
Anomalous pulmonary venous drainage (partial or total)
Atrioventricular canal defects (partial or complete)
Coarctation of the aorta
Ebstein's anomaly
Significant infundibular right ventricular outflow tract obstruction
Ostium primum atrial septal defect
Patent ductus arteriosus
Pulmonary valve regurgitation (moderate to severe)
Sinus of Valsalva fistula/aneurysm
Sinus venosus atrial septal defect
Supravalvular or subvalvular aortic stenosis
Tetralogy of Fallot
Ventricular septal defect with:
Absent valve or valves
Aortic regurgitation
Coarctation of the aorta
Mitral valve disease
Right ventricular outflow tract obstruction
Straddling tricuspid/mitral valve
Subaortic stenosis

From Connelly et al.[4], modified.

hemodynamic and cardiac problems from those currently seen. For example, an infant with transposition of the great arteries will no longer have a Mustard or Senning procedure (with its late problems of systemic ventricular dysfunction, arrhythmias and pathway obstruction), but might be anticipated to have an arterial switch procedure and encounter quite different cardiac sequelae in adult life. Patients with a complex single-ventricle physiology and various modifications of the Fontan procedure will increase in number. Perhaps, with refinements in non-invasive diagnosis and with the earlier definitive repair of shunt lesions, the prevalence of pulmonary vascular disease and Eisenmenger syndrome in the adult population will diminish[1].

Patient follow-up care can be stratified into three levels: 1) patients who require care exclusively in a grown-up congenital heart (GUCH) unit; 2) patients in whom shared care can be established with the appropriate general adult cardiac services; 3) patients who can be managed in "non-specialist" clinics. The majority of patients may only need to be reviewed in the outpatient clinic. A minority of grown-up patients require hospital admission.

Table III. Types of simple congenital heart defects: these patients can usually be cared for in the general medical community.

Isolated congenital aortic valve disease
Isolated congenital mitral valve disease
Isolated patent foramen ovale or small atrial septal defect
Isolated small ventricular septal defect
Mild pulmonary stenosis
Previously ligated or occluded ductus arteriosus
Repaired atrial septal defect without residua
Repaired ventricular septal defect

From Connelly et al.[4], modified.

The major reason for cardiac medical admission is for the control of arrhythmias and for interventional/diagnostic cardiac catheterization. These services need to be available in the specialist center. Other reasons for hospital admission include the investigation and assessment of non-cardiac problems, both surgical and medical, as well as of infections, endocarditis, and pregnancy.

Medical issues

Arrhythmias. Arrhythmias are a major cause of morbidity and mortality and are one of the most important reasons for hospitalization[3]. The etiology is multifactorial with electrical disturbances that form part of the defect (e.g. preexcitation in Ebstein's anomaly), a consequence of the operation (e.g. atrial arrhythmias following Senning/Mustard repair for transposition of the great arteries), or the result of hemodynamic abnormalities during follow-up (e.g. ventricular tachycardia with pulmonary regurgitation after repair of tetralogy of Fallot). Arrhythmias, which might be considered benign in patients with normal hearts, may lead to a catastrophic hemodynamic decline and be life-threatening in patients with surgical residua.

Supraventricular arrhythmias are more frequent than ventricular arrhythmias, and are becoming more frequent as the length of follow-up increases. Sinus node dysfunction is most common after atrial surgery[5-7]. Pharmacological treatment may be limited by hemodynamic side effects, concomitant sinus node dysfunction and by the desire for pregnancy[8]. Catheter ablation and surgical approaches have been increasingly applied[9], but the success rates remain lower than those in structurally normal hearts[10].

Pacing in these patients is often difficult due to the limited, abnormal access to the heart as well as the abnormal cardiac anatomy itself[11,12]. The implantable cardioverter-defibrillator will be used with increasing frequency in patients with congenital heart defects who are considered at risk of sudden death.

Interventional/diagnostic cardiac catheterization. Despite new diagnostic imaging techniques, cardiac catheterization remains an important part of the assessment and management of ACHD. The rapid increase in the type and application of interventional techniques in childhood congenital heart disease has been mirrored in the adult population. There is an increasing need for therapeutic interventions; an interventional program is central to any GUCH unit. Transcatheter atrial septal defects, balloon valvuloplasty and stent implantation are procedures frequently performed with very good results in the cath lab of units for adults with congenital heart defects[13-16]. Embolization of venous and arterial collaterals, fistulae and patent ductus arteriosus are also frequently needed. The most recent technique introduced in the cath lab is percutaneous valve implantation into the right ventricular outflow[17].

Hematological problems. Hematological problems are the most frequently encountered in cyanotic patients. The hyperviscosity syndrome, the iron deficiency, the reduced platelet count and abnormal platelet function together with clotting factor deficiencies, are the major concerns for these patients[18]. Some patients may require venesection for symptoms due to hyperviscosity but it is important to avoid iron deficiency that may result from over-zealous treatment. Hyperuricemia is common and may produce frank gout.

Infective endocarditis. Most, but not all, patients have a life-long risk of endocarditis[19]. Appropriate information and health patient education on this subject are mandatory. Antibiotic prophylaxis is recommended after the surgical repair of most conditions[20]. The portals of entry for infection are more numerous than generally appreciated and include acne, body piercing, and tattooing.

Non-cardiac medical problems. There is increasing need for specialist advice in other disciplines:
- endocrinology: in case of thyroid dysfunction in patients on amiodarone;
- rheumatology: in case of gout and arthropathy in cyanotic patients;
- neurology and orthopedics: in case of trauma and spinal disorders;
- nephrology: in case of proteinuria or a reduction in the glomerular filtration rate in cyanotic patients.

Counseling regarding contraception and pregnancy is required and it is important that it is begun before conception and taking into consideration the risks to both fetus and mother[21,22]. Genetic counseling may now be based on the results of trials which have shown that the transmission rate is higher if the mother rather than the father has congenital heart disease[23].

Surgical issues

ACHD fall into three categories: those who have not previously undergone operation, those who have had palliative surgery, and those who have had reparative surgery. There is a wide range of conditions which require specialist surgical intervention (Table IV).

Table IV. Most frequent surgical procedures needed in adult patients with congenital heart defects.

Secundum atrial septal defect closure
Ross operation or aortic valve replacement
Native aortic coarctation and re-coarctation
Modified Blalock-Taussig shunt
Mitral valve repair
Ostium primum atrial septal defect closure
Complete repair of tetralogy of Fallot
Ventricular septal defect closure
Patent ductus arteriosus ligation
Pulmonary artery banding
Fontan conversion

Surgery can be performed safely only by teams with a vast experience in the management of congenital heart defects in infants and children as well as knowledge of the principles of conventional adult cardiac surgery[24-26]. These patients challenge the surgeon because they have unique surgical problems[24]. Replacement of conduits may require modification of both surgical and bypass techniques depending on the position of the conduit and on the presence or absence of associated defects. Hemostasis may be difficult during and after surgery, particularly in case of reoperation and in cyanotic patients with abnormal clotting factors. Myocardial dysfunction and pulmonary vascular disease are often present in patients who may have undergone primary repair at an older age. Patients with cyanotic heart disease present the most problems and their stay in intensive care and hospital is considerably longer than for adults undergoing valvular or coronary arterial surgery.

Other needs

In addition to provision of care for complex medical and surgical problems, it is important to provide support for the many psychosocial problems in this population[26], such as employment, insurance, physical activity, and sport[27,28].

Conclusions

In Italy the population of grown-ups with congenital heart disease is increasing; for this reason a review of their needs is mandatory. In Italy the healthcare needs of many grown-ups with congenital heart disease are not currently being met. The obstacles to the improvement of their care include the critical shortage of trained and experienced professionals and the inadequate number of centers of excellence to lead the efforts necessary to provide high-quality care.

Specialists for the management of ACHD require knowledge of congenital cardiac malformations (their treatment in infancy and childhood), and knowledge of general medicine; training in adult cardiology including coronary arterial disease is also mandatory. The surgeons need expertise and training in both adult and pediatric cardiac surgery.

Adults with congenital heart defects quite often necessitate support by specialties such as electrophysiology, radiology, intensive care, anesthesia, dental service, and magnetic resonance imaging; for this reason the GUCH unit should be located in an adult medical environment with multidisciplinary specialty provision and be associated with a pediatric cardiology group.

There is an urgent need to create dedicated specialist units and defined referral links. Ideally specialist units should be established in appropriate geographic areas; complex patients need to be grouped for expertise, experience, and optimal management. Less specialized regional centers and outpatient clinics in interconnected districts with GUCH units should be created. Specialist units should accept responsibility for educating the profession, training the specialists, and sharing particular skills between each other.

Training programs for specialist staff should be defined and implemented[24-26]. This training is indicated for both the adult and pediatric cardiologist; it requires the following skills: 1) knowledge of congenital cardiac malformations and their treatment in infancy and childhood; 2) knowledge of general medicine; 3) knowledge of adult cardiology.

In a specialized center for grown-ups with congenital heart disease, it may be advantageous to appoint consultants with both backgrounds. It may be necessary to consider a specific training program, but we do not have any suggestions regarding the Italian training system.

The creation of specialist units sharing their knowledge with regional units should reduce the number of useless hospitalizations with a significant economical benefit for the national health system.

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