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ADULT CONGENITAL HEART DISEASE – NEW GUIDELINES AND CLINICAL CARE PERSPECTIVE

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Abstract

To date, the prevalence of CHD worldwide is ~9 per 1000 newborns, with substantial geographic variation. The latest knowledge in the world for the last 50 years about their origin, diagnosis and therapy has contributed to their care. Since adult patients with CHD now present increasing numbers at advanced ages, including the elderly, the term grown-up CHD no longer appears appropriate and was therefore replaced with adult CHD (ACHD) according to the ESC guidelines published in 2020 year. Due to medical, surgical, and technological evolutions over the past decades, >90% of individuals who are born with CHD now survive into adulthood. ACHD represent a challenge for clinicians. Despite optimal medical and surgical treatment, many will experience a progressive decline in cardiopulmonary function leading to advanced heart failure. Severe ventricular dysfunction and/or pulmonary hypertension may not be amenable to corrective repair. Their early recognition and follow-up in adolescence will contribute to better care for these patients. Importantly, the care for ACHD patients is a lifelong process and requires advance care planning strategies.

Key words: Adult congenital heart disease, heart failure, pulmonary hypertension, infective endocarditis, sudden cardiac death.

Introduction

To date, the prevalence of CHD worldwide is ~9 per 1000 newborns, with substantial geographic variation. Congenital heart defects (CHD) are more common than those found in all age groups, including the fetus. The latest knowledge in the world for the last 50 years about their origin, diagnosis and therapy has contributed to their care. However, in underdeveloped countries, millions of children born with CHD do not have adequate diagnosis, therapy, or prevention.

Since 2006, The World Society for Pediatric and Congenital Heart Surgery has been promoting the care of children with CHD from fetus to adulthood, regardless of the economic status of patients, with recommendations for education, diagnostic and therapeutic care for all. Since 1970, more than 70 population epidemiological studies have been published worldwide with a questionnaire on genetics, sociodemographic, medical-obstetric data, exposure to environmental risks and drugs, risk assessment and prevention of heart defects. Since adult patients with CHD now present increasing numbers at advanced ages, including the elderly, the term grown-up CHD no longer appears appropriate and was therefore replaced with adult CHD (ACHD) according to the ESC guidelines published in 2020 year.

Methods and materials

A. Aetiology

Congenital heart defects are disorders of the anatomical structure or function of the heart and blood vessels, which are most often the result of impaired or stopped development of certain structures at the level of the embryonic or fetal phase. The exact cause of most congenital heart defects remains unknown. They are the result of the complex action of genetic and environmental factors. Knowing the aetiology is important for their prevention.

Chromosomal aberrations and gene mutations have been detected in less than 10% of congenital heart defects, whether they are isolated or associated with other genetic abnormalities in syndromes such as Down, Turner, Marfan, Noonan, Loeys-Dietz and others. There has been remarkable progress in understanding the genetic basis of cardiovascular malformations. Chromosome microarray analysis has provided a new tool to understand the genetic basis of syndromic cardiovascular malformations resulting from microdeletion

or microduplication of genetic material, allowing the delineation of new syndromes.

Enzyme deficiencies in fetal cells obtained by amniocentesis or biopsy of chorionic villi contribute to the prediction of defects, and fetal echocardiography can directly detect cardiovascular malformations during the so-called risk factors (family burden, mother's age, etc.). Teratogenic agents, which act especially during the embryonic development of the heart, have a role in the development of congenital heart defects in the first 3 months of pregnancy. They are divided into infectious, chemical and physical, and the most famous are rubella and maternal viral infections in general; drugs the mother takes, as well as heroin, cocaine, smoking, alcohol; then hypoxia, radiation, trauma, etc.

B. Classification of ACHD

The classification of congenital heart defects can be based on anatomical, functional (hemodynamic), radiological or other characteristics of defects that are often combined or complex. CHD can be classified as mild, moderate, or severe according to complexity. (**Table No. 1**)

Table No. 1. Classification of congenital heart disease complexity

MILD:
Isolated congenital aortic valve disease and bicuspid aortic disease
Isolated congenital mitral valve disease (except parachute valve, cleft leaflet)
Mild isolated pulmonary stenosis (infundibular, valvular, supra-valvular)
Isolated small ASD, VSD, or PDA
Repaired secundum ASD, sinus venosus defect, VSD, or PDA without residue or sequelae, such as chamber enlargement, ventricular dysfunction, or elevated PAP.
MODERATE: (Repaired or unrepaired where not specified; alphabetical order)
Anomalous pulmonary venous connection (partial or total)
Anomalous coronary artery arising from the PA
Anomalous coronary artery arising from the opposite sinus
Aortic stenosis - subvalvular or supra-valvular
AVSD, partial or complete, including primum ASD (excluding pulmonary vascular disease)
ASD secundum, moderate or large unrepaired (excluding pulmonary vascular disease)
Coarctation of the aorta
Double chambered right ventricle
Ebstein anomaly
Marfan syndrome and related HTAD, Turner Syndrome

PDA, moderate or large unrepaired (excluding pulmonary vascular disease)
Peripheral pulmonary stenosis
Pulmonary stenosis (infundibular, valvular, supra-valvular), moderate or severe
Sinus of Valsalva aneurysm/fistula
Sinus venosus defect
Tetralogy of Fallot – repaired
Transposition of the great arteries after arterial switch operation
VSD with associated abnormalities (excluding pulmonary vascular disease) and/or moderate or greater shunt
SEVERE: (Repaired or unrepaired where not specified; alphabetical order)
Any CHD (repaired or unrepaired) associated with pulmonary vascular disease (including Eisenmenger syndrome)
Any cyanotic CHD (unoperated or palliated)
Double-outlet ventricle
Fontan circulation
Interrupted aortic arch
Pulmonary atresia (all forms)
Transposition of the great arteries (except for patients with arterial switch operation)
Univentricular heart (including double inlet left/right ventricle, tricuspid/mitral atresia, hypoplastic left heart syndrome, any other anatomic abnormality with a functionally single ventricle)
Truncus arteriosus
Other complex abnormalities of AV and ventriculoarterial connection (i.e., crisscross heart, heterotaxy syndromes, ventricular inversion).

ASD = atrial septal defect; AV = atrioventricular; AVSD = atrioventricular septal defect; CHD = congenital heart disease; HTAD = heritable thoracic aortic disease; LV = left ventricle/ventricular; PA = pulmonary artery; PAP = pulmonary artery pressure; PDA = patent ductus arteriosus; VSD = ventricular septal defect. Source: 2020 ESC Guidelines for the management of adult congenital heart disease. *European Heart Journal*, Volume 42, Issue 6, 7 February 2021, Pages 563–645,

The classification into defects without cyanosis and with cyanosis is common. Cyanosis is divided into defects with left-right shunt and without shunt. Defects with a shunt are further divided according to the level of communication between the systemic and pulmonary circulation, and defects without a shunt depending on whether they refer to the structures of the inflow or outflow through the left or right heart. Cyanotic defects are divided into those with increased and reduced pulmonary flow, based on which the cause of cyanosis (mixing of arterial and venous blood or reduction of pulmonary flow) is clarified.

Congenital heart defects in adults include: atrial septal defect (ASD), atrial septal defect and anomalous pulmonary venous connection, ventricular septal defect (VSD), atrioventricular septal defect (AVSD), patent ductus arteriosus (PDA), left ventricular outflow tract obstruction (LVOTO), valvular

aortic stenosis, supraventricular aortic stenosis, subaortic stenosis, coarctation of the aorta (CoA), Aortopathies including Marfan syndrome, right ventricular outflow tract obstruction (RVOTO), Ebstein's anomaly, Tetralogy of Fallot, Pulmonary atresia with ventricular septal defect, transposition of the great arteries, congenitally corrected transposition of the great arteries (ccTGA), univentricular heart, patients after Fontan operation and coronary anomalies. To date, ~90% of patients with mild, 75% with moderate, and 40% with complex heart defects reach the age of 60 years.

The frequency of individual defects is different. Although over a hundred anomalies have been described, 85% are due to 8 congenital heart defects that also occur in adults: atrial septal defect, ventricular septal defect, patent ductus arteriosus, congenital aortic stenosis, aortic coarctation, pulmonary stenosis, tetralogy of Fallot, transposition of great arteries.

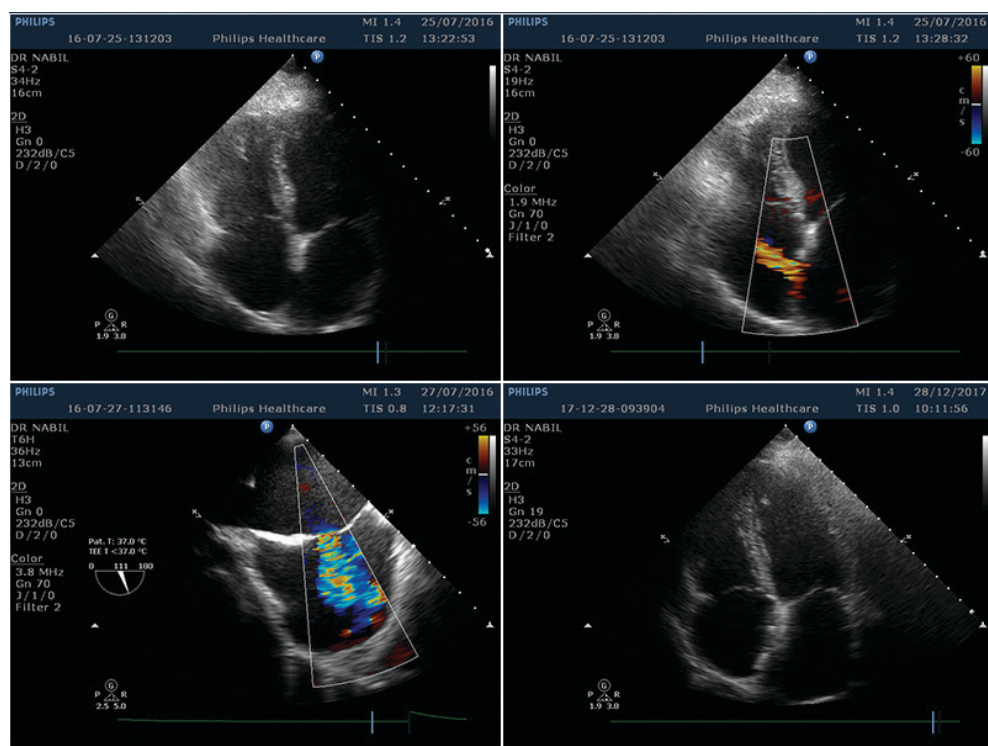


Figure 1. A secundum atrial septal defect (ASD) in 46 years old female patient with left-to-right shunt was confirmed by 2-dimensional transthoracic echocardiogram (TTE) (Panel A), 2-dimensional TTE color Doppler (Panel B). Transesophageal bicaval view with color Doppler (TEE) (Panel C) and TTE echocardiography of the final result after transcatheter closure with Amplatzer occluder (Panel D).

Secundum ASD (80% of ASDs; located in the region of the fossa ovalis and its surrounding). The ASD type secundum is the communication between the left and right atria placed lower towards the mitral valves. It is often associated with anterior mitral valve fissure and consequent mitral regurgitation (MR). Device closure has become the first choice for secundum defect closure, when feasible, based on the morphology (includes stretched diameter ≤ 38 mm and sufficient rim of 5 mm except towards the aorta).

Primum ASD [15%; synonyms: partial AV septal defect [atrioventricular septal defect (AVSD) with communication on the atrial level only], partial AV canal; located near the crux, AV valves are typically malformed, resulting in various degrees of regurgitation. The shunt volume depends on RV/LV compliance, defect size, and LA/RA pressure. If the defect is large, it burdens the pulmonary circulation and gives symptoms. It can be treated by surgical or catheter interventional treatment.

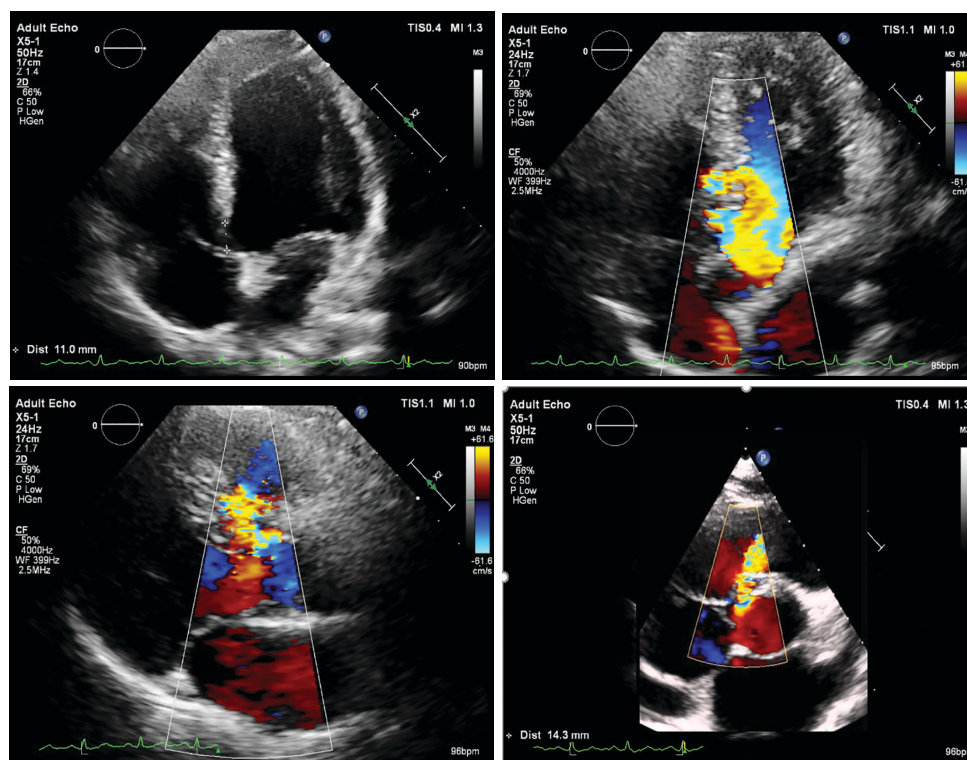


Figure 2. A perimembranous ventricular septal defect with small muscular VSD in 34 years old male with left - right shunt was confirmed by 2-dimensional transthoracic echocardiogram (TTE) (Panel A) and 2-dimensional TTE color Doppler (Panel B, C and D).

VSD is mostly diagnosed and, when indicated, treated before adulthood. Spontaneous closure is frequent in childhood. Several locations of the defect within the interventricular septum are possible, and these can be divided into four groups according to their location within the RV: perimembranous /paramembranous/subaortic/conoventricular (most common, ~80% of VSDs), muscular/trabecular (up to 15-20%), Outlet (with or without malalignment of the outlet septum) and Inlet/AV canal/AVSD type. Due to the increased blood flow on the left side, the pulmonary circulation is burdened. Surgical closure can be performed with low operative mortality (1–2%) and good long-term results. Transcatheter closure has become an alternative, particularly in residual VSDs, in VSDs that are poorly accessible for surgical closure, and in muscular VSDs that are located centrally in the interventricular septum.

Patent ductus arteriosus (PDA) is the persistent communication between the proximal left PA and the descending aorta just distal to the left subclavian artery. It can be associated with a variety of CHD lesions, however, in adults, it is usually an isolated finding. PDA originally results in L–R shunt and LV and LA volume overload. In adults, calcification of the PDA may cause a problem for surgical closure. Device closure is the method of choice, even if cardiac operations are indicated due to other concomitant cardiac lesions and can be successfully performed in the vast majority of adults with a very low complication rate.

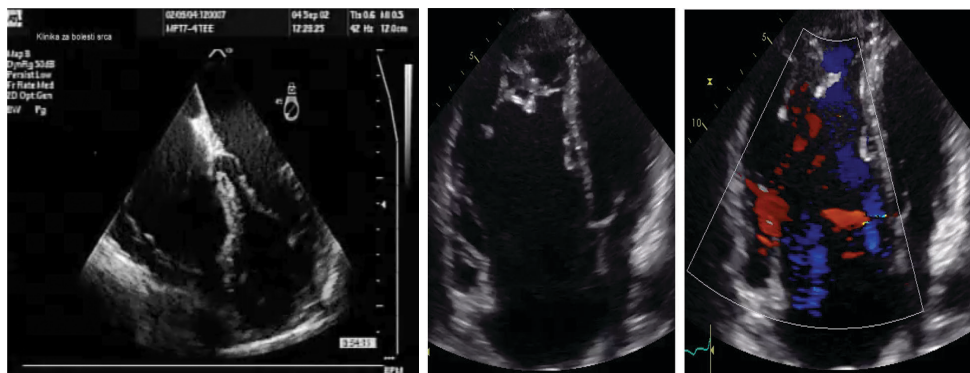


Figure 3. Ebstein's Anomaly with atrialization of the right ventricle in 56 years old female patient with history of supraventricular paroxysmal tachycardia and ischemic cerebrovascular insult.

Ebstein's anomaly is characterized by abnormally formed and apically displaced leaflets of the tricuspid valve. The anterior leaflet usually originates

at the annular level but is enlarged and sail-like, while the septal and posterior leaflets are displaced towards the RV apex and often tethered to the endocardium. Clinical symptoms determine the treatment. Conservative therapy can alleviate symptoms temporarily and create a beneficial basis for the following operation. Surgical repair remains challenging and should only be performed by surgeons with specific experience in this lesion.

CoA is considered as part of a generalized arteriopathy, and not only as narrowing of the aorta. It occurs as a discrete stenosis or as a long, hypoplastic aortic (arch) segment. Typically, CoA is located in the area where the ductus arteriosus inserts, and only in rare cases occurs ectopically (ascending, descending, or abdominal aorta). Associated lesions include bicuspid aortic valve (up to 85%), ascending aortic aneurysm, subaortic stenosis or supraortic stenosis and (supra)mitral valve stenosis. Patients with CoA who reach adolescence demonstrate particularly good long-term survival up to age 60 years. The natural course may be complicated by left heart failure, intracranial haemorrhage (from berry aneurysm), infective endocarditis, aortic rupture/dissection, premature coronary and cerebral artery disease, and associated heart defects. (1,2,3.4.5.6)

C. Clinical signs

Due to medical, surgical, and technological evolutions over the past decades, >90% of individuals who are born with CHD, now survive into adulthood. As a result, the prevalence of CHD in the community has increased and now by far exceeds the number of children with CHD.

In congenital heart defects, anatomical changes cause functional changes in the heart and circulation, which further cause new anatomical changes and vice versa. They are interdependent, dynamic and constantly progressing, from the beginning to the end of life. Depending on the severity of the hemodynamic disorders, a number of defects are fatal before birth or immediately after birth. A significantly higher number of anomalies allow the development of the child, but with disorders (developmental delay, feeding dyspnoea, heart failure, etc.) that require correction in the earliest childhood or during adolescence. There are also defects with mild hemodynamic disorders and without them, which remain undiagnosed in childhood, and for quite a long time in adults until the appearance of problems due to complications of the defect, e.g., pulmonary hypertension and heart failure in the IV or V decade of life of a patient with a moderately large left-right shunt.

ACHD have an anamnestic long asymptomatic period, followed by a feeling of fatigue, shortness of breath on exertion, palpitations, arrhythmias, respiratory infections, and bacterial endocarditis.

Thanks to advances in early detection, medical and/or surgical treatment of defects and interventional cardiology, palliative/total correction of even complex congenital heart defects is performed at an earlier age, which allows the patient to live with fewer symptoms or without them. In the population of adults with congenital heart defects, more and more patients are operated on instead of patients with advanced functional and/or pathoanatomical disorders that prevent surgical intervention (except for transplantation). The condition of the pulmonary vascular bed and pulmonary hypertension play a decisive role in determining the operability of the defect, on which the clinical manifestations, course and prognosis of many defects also depend.

Pulmonary hypertension occurs due to increased blood flow and/or resistance in the pulmonary circulation. Increased pulmonary flow is given by defects with the left-right shunt. Increased pulmonary vascular resistance is most often the result of histopathological, obstructive changes in the small muscular arteries and pulmonary arterioles that occur in patients with pulmonary hypertension and/or increased pulmonary blood flow. Assessment of pulmonary hypertension and the condition of the pulmonary vascular bed is most often performed by catheterization of the heart. In addition to pressures, the magnitudes of blood flow and vascular resistance in the pulmonary and systemic bloodstream are determined and their relative ratios are calculated. In advanced cases, pulmonary angiography and lung biopsy are required.

Cyanosis: Central-type cyanosis in adults with congenital heart defect is a symptom and sign of hypoxemia due to the right-left shunt. Congenital cyanotic heart defects are less common, and cyanosis occurs much more often due to left-right shunt reversal in unoperated patients with initially cyanotic defect (Eisenmenger's syndrome due to a large interventricular septal defect, atrioventricular septal defect or open arterial canal).

Erythrocytosis is a physiological response to hypoxemia. Increased erythrocyte mass (haematocrit) and total blood volume are features of polycythaemia in patients with right-left shunt. Polycythaemia itself has side effects: thrombosis, embolism, bleeding. Thromboembolic complications and symptoms such as headache, dizziness, fatigue, numbness of the fingers, are caused by increased blood viscosity due to high haematocrit ($Hct > 60\%$). Bleeding

is a consequence of haemostasis disorders, due to impaired platelet function and abnormalities in the coagulation and fibrinolytic system.

Infective endocarditis is a common complication in patients with congenital heart defects, and routine antibiotic prevention is mandatory in all non-operated patients, as well as in most patients after defect correction. Patients are especially at risk after the implantation of valves (artificial, heterograft or homograft) and “conduit”, so in these cases, parenteral administration of a combination of antibiotics is recommended. (1,2,9,11,12,13,14,15,17)

D. Diagnostic tools

The diagnosis is made by clinical examination, ECG, X-ray of the lungs and heart, biomarkers, echocardiography, Cardiac magnetic resonance imaging (CMRI), Computed tomography (CT), cardiopulmonary exercise testing (CPET) and cardiac catheterization. **Echocardiography** remains the first-line investigation and continues to evolve, with improved functional assessment using 3D echocardiography. **Cardiovascular Magnetic Resonance Imaging (CMRI)** has become an essential facility in the specialist unit. It enables 3D anatomical reconstruction, which is not restricted by body size or acoustic windows and has rapidly improving spatial and temporal resolution. **Cardiovascular CT** has high spatial resolution and rapid acquisition time; it is particularly relevant for imaging the great vessels, coronary arteries, and collateral arteries, and for parenchymal lung disease. **Cardiopulmonary exercise testing** has an important role in the CHD population, in which quality of life and functional capacity are key measures of the success of intervention. Cardiac catheterization is mainly reserved for resolution of specific anatomical and physiological questions, or for intervention. **Different classes of biomarkers** have been reported to be associated with adverse events in the CHD population, including: neurohormones peptides [B-type natriuretic peptide (BNP) and N-terminal-pro-BNP (NT-pro-BNP)], markers of myocardial injury (high-sensitivity troponins) or inflammation marker (high-sensitivity C-reactive protein). (1,2,6,7,10,17)

D. Therapeutic consideration

Heart Failure: The development of heart failure is a common problem affecting 20 to 50% of the ACHD population and is a main cause of death.

Arrhythmias: The entire spectrum of arrhythmias may be encountered in ACHD patients. Supraventricular arrhythmias: atrioventricular reentrant

tachycardia (AVRT), intraatrial reentrant tachycardia (IART), ectopic atrial tachycardia (EAT) and atrial fibrillation (AF). Ventricular arrhythmias: sustained ventricular tachycardia (SVT) & sudden cardiac death (SCD). Bradycardia: sinus node dysfunction (SND) and AV block. **Sudden cardiac death (SCD)** related to ventricular arrhythmia is of concern (7–26% of all deaths in adults). Although the incidence in the CHD population at large is relatively low (<0.1% per year), some defects place patients at higher risk, with occasional disease-specific substrates and risk factors. Identifying patients at risk for SCD remains a challenge. **Pulmonary hypertension** is an important prognostic factor in patients with CHD, requiring particular attention in pregnancy or prior to reparative cardiac or other major surgery. Sudden cardiac death and risk stratification. **Cardiac surgery** in ACHD patients deserves special attention. Even small operations can carry a high risk. The need for personalized risk assessment, understanding the specific anatomy and haemodynamics, experience with CHD surgery or redo surgery, and special requirements in intensive care units are factors that determine short- and long-term outcomes. **Catheter interventions:** The most frequent percutaneous interventions are closure of shunt lesions (in particular secundum-type ASD, rarely VSD, and persistent arterial duct), fistula, or unusual collaterals; balloon dilatation of the pulmonary valve and valved grafts; balloon dilatation and/or stenting of narrowed great vessels [e.g. (re-)coarctation of the aorta (CoA) and pulmonary arterial stenosis]; and transcatheter pulmonary valve implantation (TPVI). **Infective endocarditis:** The risk of infective endocarditis in ACHD patients is higher than in the general population, with marked variation between lesions. **Antithrombotic treatment:** It is known that patients with ACHD are at increased risk of thromboembolic events. **Management of cyanotic patients:** cyanosis caused by R–L shunt due to an anatomical communication between the systemic and pulmonary circulation at the atrial, ventricular, or arterial level. Cyanotic patients are complex and must be followed by an ACHD specialist. Multimodality imaging is key for adequate assessment of overall anatomy and ventricular and valvular function, and quantification of blood flow, including perfusion distribution. Special structural and organizational healthcare requirements are necessary to meet the needs of ACHD patients. (1,2,3,9,10,11)

Future directions

The main unmet needs in adult congenital heart disease are: 1. Increase in the number of randomized controlled trials. 2. Advanced care of heart failure in ACHD (Mechanical assist devices, transplant in complex CHD). 3. Cardiac resynchronization therapy in complex ACHD. 4. Leadless pacing. 5. Primary prevention of sudden cardiac death in patients with systemic right ventricle or single ventricular physiology. 6. Targeted therapies for pulmonary hypertension in Fontan patients. 7. RCTs for novel therapeutic agents in pulmonary hypertension associated with CHD. 8. Drug therapies in patients with failing systemic right ventricle or single ventricle. 9. Direct oral anticoagulants in ACHD patients. 10. Implementation of AI for better assessing systemic right ventricular or single ventricle failure. 11. Development of validated prognostic models. 12. Omics-based personalized healthcare. There is a need for specialist ACHD centres worldwide. Staff requirements for specialist ACHD centres include adult/paediatric cardiologist with ACHD certification, ACHD imaging specialist (certified in TTE/TOE, CMR, CCT), congenital interventional cardiologist, CHD surgeon, anaesthesiologist with CHD experience and expertise, specialist nurse, invasive electrophysiologist with ACHD experience, pulmonary vascular disease expert, clinical geneticists, psychologist, social worker and palliative care team. (3,1,2)

Conclusion

ACHD represent a challenge for clinicians. Their early recognition and follow-up in adolescence will contribute to better care of these patients. The vast majority of patients survive into adulthood and their profile in terms of comorbidities has changed. Organization of tertiary and nontertiary care, collaboration at national and international level, randomized controlled trials and implementation of novelties, such as research based biobanking, e-health and artificial intelligence should all be employed to meet their healthcare needs. Importantly, the care for ACHD patients is a lifelong process and also requires advance care planning strategies.

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UROĐENE SRČANE BOLESTI ODRASLIH - NOVE SMJERNICE I PERSPEKTIVE KLINIČKE NJEGE

Apstrakt

Trenutna prevalencija urođenih srčanih mana (USM, eng. CHD) u svijetu je ~9 na 1000 novorođenčadi, sa značajnim geografskim varijacijama. Najnovija svjetska saznanja u posljednjih 50 godina o njihovom porijeklu, dijagnozi i terapiji doprinijela su njezi. Budući da danas ima sve više odraslih pacijenata s urođenim srčanim manama čak i u poodmakloj dobi, uključujući duboku starost, pojam “grown-up CHD” više se ne čini prikladnim i stoga je zamijenjen pojmom “adult CHD” (ACHD) prema smjernicama ESC-a objavljenim 2020. godine. Zbog medicinskog, hirurškog i tehnološkog napretka u posljednjim decenijama, >90% pojedinaca koji su rođeni s USM sada preživljavaju i u odraslu dob. ACHD predstavlja izazov za kliničare. Uprkos optimalnom medicinskom i hirurškom liječenju, mnogi će doživjeti progresivno smanjenje kardiopulmonalne funkcije što dovodi do uznapredovale srčane insuficijencije. Teška ventrikularna disfunkcija i/ili plućna hipertenzija možda neće biti podložne korektivnim intervencijama. Njihovo rano prepoznavanje i praćenje u adolescenciji doprinijet će boljoj skrbi za ove pacijente. Važno je istaknuti da je briga o pacijentima s ACHD-om doživotni proces i zahtijeva unaprijed planirane strategije njege.

Ključne riječi: urođena srčana mana kod odraslih, zatajenje srca, plućna hipertenzija, infektivni endokarditis, iznenadna srčana smrt