Prirođene srčane bolesti u Hrvatskoj — pregled sadašnjeg stanja i ciljevi

Congenital heart diseases in Croatia — a review of current state and goals

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U ovom broju časopisa prikazan je izvrstan iscrpni pregled najnovijih znanstvenih istraživanja iz područja prirođenih srčanih bolesti, kako u djece, tako i u odraslih. S obzirom na relativno malu prevalenciju ovih bolesnika u općoj populaciji, znanstvena istraživanja u ovom području iznimno su važna. Ona doprinose razvoju dijagnostike i liječenja prirođenih srčanih bolesti, budući da je većina smjernica koje sada imamo još uvijek dominantno temeljena na konsenzusu stručnjaka, a ne na čvrstim znanstvenim podacima[1]. U Hrvatskoj je zdravstvena skrb o prirođenim i korigiranim srčanim greškama u odrasloj dobi u fazi razvoja, što je i razumljivo, obzirom na ne tako davni razvoj kardiokirurške i pedijatrijske skrbi koja je omogućila sve bolje i dulje preživljavanje sve većeg broja pojedinaca. Obzirom na brojne specifičnosti ove populacije i njihove potrebe, u kardiološkom zbrinjavanju ovih bolesnika od pedijatrijske do odrasle dobi potrebno je slijediti europske i svjetske standarde.

Neki aspekti stanja u hrvatskoj pedijatrijskoj kardiologiji

Pedijatrijska kardiologija u Hrvatskoj napreduje sukladno iskoracima razvijenih europskih zemalja, što se može prikazati s nekoliko aspekata objavljenih u literaturi; epidemiološke studije, genetika u pedijatrijskoj kardiologiji, zbrinjavanje boesnika s prirođenim srčanim greškama koji iz dječje prelaze u odraslu dob, razvoj fetalne kardiologije, zbrinjavanje kardiomiopatija i plućne hipertenzije, razvoj intervencijske kardiologije, transplantacije srca i drugo.

Epidemiologija

U Hrvatskoj su učinjene dvije velike epidemiološke studije. U obadvije se koristio upitnik EUROKAT i BWIS, s time da je prva obuhvatila razdoblje od 1995. do 2000. godine samo s populacijskim registrom, a druga je imala uz populacijski i hospitalni registar, pa je učinjena klinička epidemiološka studija. Slijedili smo Clarcovu etiopatogenetsku osnovu.

U prvoj studiji (1995.-2000.) nađena je incidencija prirođenih srčanih grešaka (PSG) od 8‰ (na 276.565 poroda nađeno je 1.126 muške i 1.978 ženske djece s PSG). Rezultati ne odstupaju u općoj niti pojedinačnoj incidenciji od onih objavljenih u literaturi (0,55-1%)[2].

This issue of the journal outlines a detailed overview of the most recent scientific researches in the area of congenital heart diseases, not only in children, but in adults as well. Considering a relatively small prevalence of such patients in the general population, the scientific researches are extremely important in this area. They contribute to the development of diagnostics and treatment of congenital heart diseases, since most of the guidelines that we now have available are still dominantly based on the experts' consensus, not on sound scientific data[1]. In Croatia, the healthcare on congenital and corrected heart defects in adulthood is in the stage of development, which is understandable, considering the cardiac and pediatric care that has developed not so long ago, which, however, enabled better and longer survival of an increasing number of individuals. Considering many specifics of this population and their needs, the European and international standards are to be followed in cardiac management of these patients from pediatric to adult age.

Some aspects of the state in the Croatian pediatric cardiology

Pediatric cardiology in Croatia keeps up with advances of the developed European countries, which may be presented by using several aspects published in the literature; epidemiological studies, genetics in pediatric cardiology, management of patients with congenital heart disease passing from childhood to adulthood, development of fetal cardiology, management of cardiomyopathy and pulmonary hypertension, development of interventional cardiology, heart transplantation etc.

Epidemiology

Two epidemiologic studies have been conducted in Croatia. The both studies applied the questionnaire EUROKAT and BWIS, whereas the first included the period from 1995 to 2000, while the other had a hospital-based registry in addition to the population-based registry, so the clinical epidemiologic study was conducted. We followed the Clark etiopathogenic basis.

The first study (from 1995 to 2000) showed an incidence of congenital heart defects (CHD) of 8‰ (out of 276.565 births, 1.126 male and 1.978 female children were born with CHD). The results deviate in neither general nor individual incidence from those published in the literature (0.55-1%)[2].

Druga je studija uz populacijski imala i hospitalni registar PSG, pa je uz populacijsku statistiku učinjena i klinička epidemiološka studija (ishod liječenja u djece s PSG u spomenutom razdoblju). Rezultati te studije objavljeni su u recentnoj domaćoj i međunarodnoj literaturi[3-5]. U analizi ishoda koristili smo se ABC scorom (*Aristotle Basic Complexity Score*), a u analizi kardiokirurške trijaže (domaći vs. inozemni kardiokirurzi) procjenom kardiokirurškog rizika (*risk adjustment for congenital cardiac surgery-1 method*). Od 1. listopada 2002. do 1. listopada 2007. god. u Hrvatskoj je rođeno 205.917 djece, od čega je 1.480 djece imalo PSG (7,2‰). Najčešće postavljena dijagnoza je ventrikulski septalni defekt (VSD) (34,6%). Na kardiokirurško liječenje upućeno je 430 djece. Na njima je učinjeno 556 operacija, od čega 202 u Hrvatskoj, ostatak u inozemstvu. Ukupni mortalitet nakon kardiokirurškog liječenja iznosio je 4-5% i u Hrvatskoj i u inozemstu (znak dobre procjene kod trijaže za kardiokirurški rizik). Razlike u mortalitetu i morbiditetu nakon kardiokirurških operacija izvedenih u Hrvatskoj i onih u inozemstvu uočavaju se nakon analize ishoda liječenja prema dogovorenim metodologijama.

Pedijatrijska kardijalna kirurgija u kliničkoj epidemiološkoj studiji

Od 29. listopada 2010. godine ravnopravni smo članovi baze podataka EACTS (*European Association for Cardiothoracic Surgery*) gdje smo putem registra unijeli podatke za Hrvatsku od 2008. do 2011. godine. Na osnovu prikaza tih podataka, jasno je vidljiv dinamički razvoj kardijalne kirurgije u području PSG, tako da se stalno povećava broj operiranih bolesnika u zemlji, a smanjuje broj operiranih u inozemnim centrima. U dubokoj depresiji kardijalne kirugije prije 2008. godine broj operirane djece pao je na ispod 50 tijekom jedne godine, a od 2008. do 2011. god. raste broj operacija u zemlji u odnosu na inozemstvo kako slijedi; godine 2008., 2009., 2010., 2011. — broj operacija u zemlji 55, 78, 121, 123, a operacije u inozemnim centrima 78, 55, 61, 70. Vidljivo je da se u zemlji radi dvostruko više operacija nego u inozemstvu u najnovije vrijeme, dok je prije 2008. godine taj odnos bio obrnut.[6]

Genetika

Genetski čimbenici se smatraju glavnim razlogom nastanka PSG, a teratogeni čimbenici danas su manje zastupljeni. Analiziraju se tri razine: aneuploidija, delecijski sindromi i mutacije. Na razini aneuploidije svoj djeci s fenotipskim obilježjima poznatih sindroma (Down, Turner, Edward, Patau) radi se kariogram[7], a ima i drugih trisomija koje se sporadično dijagnosticiraju. Delecijski sindromi rutinski se dokazuju FISH metodom, kao što je DiGeorgeov sindrom (delecija na kratkom kraku kromosoma -22q11.2) i Williams-Beurenov sindrom (delecija na kromosomu[7], regija za elastin)[8,9]. Delecija na kratkom kraku kromosoma 22 analizira se ne samo u bolesnika koji imaju fenotipska obilježja sindroma srce-lice već i kod drugih konotrunkusnih anomalija kao što je tetralogija Fallot, *double outlet right ventricle*, subaortni VSD, Taussig-Bingova anomalija, aortopulmonalni prozor, interupcija luka aorte tip B i C, *truncus arteriosus* (sva 4 tipa po van Praaghu) i pulmonalna atrezija. Iako još nemamo mogućnosti u zemlji dijagnosticirati mutacije koje su uzrokom PSG ili kardiomiopatija, brojne su greške egzaktno dijagnosticirane zahvaljujući suradnji s inozemnim labora-

The second study had a CHD hospital-based registry in addition to the population-based registry, so in addition to the population-based statistics, a clinical epidemiologic study was conducted (an outcome of treatment of children with CHD in the above mentioned period). The results of the study were published in the recent local and international literature[3-5]. While analyzing the outcome, we applied the ABC score (*Aristotle Basic Complexity Score*), and while analyzing the cardiac-surgical triage (local vs. international cardiac surgeons) we applied the assessment of the cardiac surgery risk (risk adjustment for congenital cardiac surgery-1 method). From 1st October 2002 to 1st October 2007, some 205.917 children were born, of whom 1.480 children had CHD (7,2‰). The most common diagnosis that was made was ventricular septal defects (VSD) (34.6%). Some 430 children were referred to cardiac surgery. They underwent 566 surgeries, of which 202 surgeries were performed in Croatia and the rest of them abroad. Total mortality after the cardiac surgery was 4 to 5% both in Croatia and abroad (a sign of good assessment for the cardiac surgery risk). The differences in mortality and morbidity after the completion of cardiac surgeries performed in Croatia from those performed abroad can be seen after completing the analysis of the outcome of the treatment according to the agreed methodologies.

Pediatric cardiac surgery in clinical epidemiologic study

Since 29 October 2010, we have been the equal members of the EACTS database (*European Association for Cardiothoracic Surgery*) where we entered the data for Croatia through the registry during the period from 2008 to 2011. The analysis of these data clearly shows the dynamic development of cardiac surgery in the area of CHD, so that a number of patients operated on in the country are continuously rising and a number of patients operated on abroad are declining. During the deep depression in cardiac surgery before 2008, a number of operated children had fallen to below 50 in one year, while from 2008 to 2011 we recorded a growing number of surgeries in the country compared to surgeries performed abroad in the way as follows: Year 2008, 2009, 2010, 2011 — the number of surgeries in the country was 55, 78, 121, 123, while the surgeries in international surgical centers was 78, 55, 61, 70. It is evident that twice more surgeries have been performed in the country than abroad recently, whereas this ratio was reversed before 2008.[6]

Genetics

Genetic factors are considered to be the main reason for occurrence of CHD, while teratogenic factors are now less common. Three levels have been analyzed: aneuploidy, deletion syndromes and mutations. At the level of aneuploidy, all the children with phenotypic characteristics of known syndromes (Down, Turner, Edward, Patau) undergo cariogram[7], and there are some other trisomies that are sporadically diagnosed. Deletion syndromes are routinely proved by the FISH method, such as DiGeorge syndrome (deletion of the short arm of chromosome -22q11.2) and Williams Beuren syndrome (deletion on chromosome[7], the region of elastin)[8,9]. Deletion of the short arm of chromosome 22 is analyzed not only in patients who have phenotypic features of heart syndrome-face, but also for other conotruncal anomalies such as tetralogy of Fallot, *double outlet right ventricle*, subaortal VSD, Taussig-Bing anomaly, aortopulmonary window, interruption of the aortic arch type B and C, *truncus arteriosus* (all 4 types by van Praagh) and pulmonary atresia. Although we are still unable to diagnose mutations causing CHD or cardiomyopathies in our country, many defects

torijima (Mowat-Willsonov sindrom, Coffin-Lowrijev sindrom, Noonanov sindrom, Barthov sindrom)[10,11].

Fetalna kardiologija

Fetalna kardiologija organizirana je u Hrvatskoj prema smjernicama AEPC (*Association of European Paediatric Cardiology*)[12]. Prema tim uputama Fetalna kardiološka ambulanta smatra se onom koja pregleda godišnje 250-300 trudnoća. U Fetalnoj kardiološkoj ambulanti Referentnog centra za pedijatrijsku kardiologiju pregleda se godišnje oko 600 trudnoća na okolnost moguće prenatalne dijagnoze: morfološke promjene (PSG), hemodinamske promjene (kardiomiopatije) ili funkcionalne promjene (aritmije). Senzitivnost pretraga prelazi 95%, a započeta je i intrauterina terapija aritmija[13]. Pregledava se i prati velik broj trudnica s gestacijskim dijabetesom na okolnost moguće dijabetičke fetalne kardiomiopatije i trudnica sa sustavnim bolestima zbog mogućnosti prenošenja anti-Ro-SSA i anti-La-SSB protutijela s konsekutivnim razvojem kompletnog atrioventrikulskog bloka. Kod tih se pacijenata provodi i prevencija AV-bloka kortikosteroidima[14]. Na ovom području napisan je i udžbenik iz fetalne kardiologije[15] i redovito se održava tečaj trajnog medicinskog usavršavanja. Fetalna kardiologija znatno je unaprijedila ukupni odnos prema srčanim bolestima u djece na prevenciji, intrauterinom liječenju, etičko-psihološkom pristupu roditeljima, blagovremenom transportu djeteta i u cijelosti adekvatnijem pristupu PSG.

Intervencijska kateterizacija srca

Intervencijska kateterizacija čini u razvijenim europskim centrima oko 50% od ukupnog broja kateterizacija srca, a taj će se broj i relativno povećavati na uštrb smanjenog broja dijagnostičkih kateterizacija, a zbog primjene drugih metoda (npr. magnetska rezonancija, MR). U Kliničkom bolničkom centru Zagreb danas se radi nešto manje od 40% intervencijskih zahvata koji obuhvaćaju cijeli spektar intervencija u pedijatrijskoj kardiologiji, osim ugradnje stentova i nekih specijalnih metoda (npr. transkateterska ugradnja zalistaka na mjesto pulmonalne ili aortne valvule). Od siječnja 1996. do prosinca 2009. god. invazivnoj dijagnostici podvrgnuto je 2.379 djece, 51% muške i 49% ženske, sa srednjom dobi u trenutku invazivne dijagnostike 4,1±3,8 godina (1 dan — 20,5 godina). U tom se razdoblju broj interventnih kateterizacija godišnje povećavao od 20/160 (12,5%) 1996. god. do 60/182 (32,9%) 2007. god. Od toga je bilo 140 atrioseptostomija, 80 dilatacija pulmonalne valvule, 36 dilatacija aortne valvule u veće djece, 9 dilatacija kritične aortne stenoze, 58 dilatacija koarktacije, 133 zatvaranja Bottalieva duktusa (4 PFM spiralom, 68 Cookovom spiralom i 61 Amplatzerovim kišobranom), u 37 bolesnika zatvoren je atrijski septalni defekt (ASD) tip II Amplatzerovim ili Cardio-seal kišobranom, u 37 bolesnika učinjena je biopsija miokarda[16].

Prirođene srčane bolesti u odraslih

U svijetu su, već neko vrijeme, prirođene srčane bolesti prepoznate kao rastuće područje kardiologije koje postaje predmet sve većeg broja istraživanja. Za to je svakako zaslužan veliki napredak u pedijatrijskoj kardiologiji i kardijalnoj kirurgiji PSG koji je omogućio sve većem broju djece sa srčanim bolestima, posebno onih s kompleksnim greškama, preživljavanje i prijelaz u odraslu dob.

were accurately diagnosed owing to collaboration with foreign laboratories (Mowat-Willson syndrome, Coffin-Lowry syndrome, Noonan syndrome, Barth syndrome)[10,11].

Fetal cardiology

Fetal cardiology has been organized in Croatia according to the AEPC (*Association of European Paediatric Cardiology*) guidelines[12]. According to such instructions, the Fetal Cardiac outpatient department is considered the one which examines 250-300 pregnancies a year. Some 600 pregnancies are examined in the Fetal cardiac outpatient department of the Referral center for pediatric cardiology for potential prenatal diagnosis on an annual basis: morphological changes (CHD), hemodynamic changes (cardiomyopathies) or functional changes (arrhythmias). The test sensitivity exceeds 95%, while the whole intrauterine treatment of arrhythmias started[13]. A large number of pregnant women with gestational diabetes are examined and followed up for potential fetal cardiomyopathy and pregnant women with systemic diseases are examined and followed up for potential transmission of anti-Ro-SSA and anti-La-SSB antibodies with consecutive development of the complete atrioventricular block. In these patients, the prevention of AV-block by titrating corticosteroids is conducted[14]. In this area, a textbook in fetal cardiology[15] was written and a course in lifelong medical training is held on a regular basis. Fetal cardiology has significantly and completely improved a position towards heart diseases in children with regard to prevention, intrauterine treatment, ethical and psychological approach to parents, timely transportation of a child and more appropriate approach to CHD as a whole.

Interventional cardiac catheterization

Interventional catheterization accounts for 50% in developed European centers of the total number of cardiac catheterizations, and this number will relatively rise at the expense of a reduced number of diagnostic catheterizations, as a result of applying some other methods (e.g. magnetic resonance imaging, MRI). Today, less than 40% of the interventional procedures is performed at the University Hospital Centre Zagreb, including a full range of interventions in pediatric cardiology, except for stent implantation and some special methods (e.g. transcatheter valve implantation at the place of pulmonary or aortic valve). From January 1996 to December 2009, some 2,379 children, of whom 51% of male and 49% of female children underwent the invasive diagnostics, at a mean age 4,1±3,8 (ranging from 1 day — 20.5 years of age) at the time of invasive diagnostics. At that time, the number of catheter interventions annually rose from 20/160 (12.5%) in 1996 to 60/182 (32.9%) in 2007. Of these, there were 140 atrioseptostomias, 80 dilatations of pulmonary valve, 36 aortic valve dilations in older children, 9 dilations of critical aortic stenosis, 58 dilatations of coarctation, 133 ductus Bottali closure (4 with PFM coils, 68 with Cook coils and 61 with Amplatzer Duct occluder), atrial septal defect (ASD) type II was closed in 37 patients by Amplatzer Duct occluder or Cardioseal occluder and myocardial biopsy was performed in 37 patients[16].

Congenital heart defects in adults

Congenital heart diseases are recognized as a growing area of cardiology which has become the subject of an increasing number of investigations in the world throughout a certain period of time. This is achieved due to a great advancement in pediatric cardiology and cardiac surgery of CHD which enabled survival and transition to adulthood for ever greater number of children with heart diseases, especially those with complex defects.

Sadašnje stanje u Hrvatskoj i problemi

U Hrvatskoj nemamo cjelovite epidemiološke podatke o broju odraslih bolesnika s PSG niti o broju njihovih hospitalizacija ili mortalitetu. Zbog nepostojanja sustavne organizacije skrbi za ove bolesnike, mnogi od njih izgubljeni su u praćenju nakon što izađu iz pedijatrijske dobi te je nepoznat njihov daljnji klinički tijek i broj. Ako pokušamo ekstrapolirati europske i svjetske epidemiološke podatke na Hrvatsku, dobijemo rezultate navedene u **Tablici 1**.[20]

Current state in Croatia and problems

In Croatia, we have no comprehensive epidemiological data on the number of adult patients with CHD or on the number of their hospitalizations or mortality. As a consequence of a lack of a systematic organization of care for these patients, many of them are no more followed up after they turn to age when they outgrow the pediatric care and their further clinical course and the number remains unknown. If we try to extrapolate the European and global epidemiological data to Croatia, we obtain the results listed in **Table 1**.[20]

Table 1. *Estimated number of patients with adult congenital heart disease.*

Country	Population	No of ACHD centres	Severity of congenital heart disease			
			Severe	Moderate	Mild	Total
USA	280 000 000		117 000	302 000	368 000	787 000
France	64 000 000	4	26 676	68 856	83 904	179 436
Germany	82 000 000	5	34 164	88 154	107 456	229 744
Italy	58 000 000	5	23 400	81 540	99 360	204 300
Belgium	10 000 000	4	4 178	11 174	13 616	28 968
TheNetherlands	16 000 000	7	6 669	17 214	20 976	44 859
UK	61 000 000	12	25 536	65 836	80 224	171 596
Croatia	4 500 000	?	1873	5002	6096	12 971

Nagli porast broja ovih bolesnika postao je jedan od važnijih javnozdravstvenih problema koji zahtjeva brzi razvoj službe za cjelovitu skrb bolesnika s prirođenim srčanim bolestima. Postojanje registra odraslih bolesnika s prirođenim srčanim bolestima preduvjet je uvida u pravo stanje na razini cijele države, usmjerene organizacije zdravstvene zaštite te specifičnih znanstvenih istraživanja koje se odnose na ovu populaciju u Hrvatskoj.

Organizacija zdravstvene skrbi ovih bolesnika u Hrvatskoj je u začecima. Trenutno ne postoji veći tercijarni centar za prirođene srčane bolesti u odraslih. O bolesnicima skrbe pojedinačni kardiolozi u raznim ustanovama, ali vrlo često i pedijatrijski kardiolozi. Velik broj bolesnika s kompleksnim prirođenim srčanim bolestima trenutno se zbrinjavaju u četiri klinička bolnička centra u Hrvatskoj, koje kao krovne ustanove objedinjuju najveći dio potrebnih službi. U KBC Zagreb osim toga ovi bolesnici zbrinjavaju se u okviru referentnih centara Ministarstva zdravstva Republike Hrvatske. Uključene službe u prvom redu su pedijatrijski kardiolozi, kardiolozi internisti te kardijalni kiruzi i kardijalni anesteziolozi s iskustvom u ovoj problematici. Nadalje, sve ostale službe potrebne za sveobuhvatno zbrinjavanje ovih bolesnika su pulmolozi, gastroenterolozi, ginekolozi, endokrinolozi i dr.

Kardiolozi se najčešće susreću s bolesnicima koji su imali jednu, a često i više kardiokirurških operacija ili zahvata, najčešće u djetinjstvu. Kod nekih od njih učinjena je kompletna korekcija srčane greške, dok je kod ostalih učinjena djelomična ili samo palijativna korekcija. Mnogi od njih imaju razne probleme i komplikacije navedenih zahvata, ali i prirodnog tijeka bolesti (cijanoza, endokarditisi, aritmije, srčano popuštanje, potrebe za reoperacijom, transplantacijom itd).

A sudden increase in the number of these patients has become one of the most important public health problems requiring a rapid development of a service for the complete management of patients with congenital heart diseases. The existence of a registry of adult patients with congenital heart diseases is a prerequisite of an insight into the real situation at the entire level of the country, a specialized health care organization and specific scientific researches that relate to this population in Croatia.

The organization of the healthcare of such patients has just been initiated. At the moment, there is no larger tertiary center for congenital heart diseases in adults. Individual cardiologists and frequently pediatric cardiologists manage the patients in various institutions. Many patients with complex congenital heart diseases are currently treated at four clinical hospital centers in Croatia, that as the umbrella institutions encompass most of the services required. In addition to the University Hospital Centre Zagreb, these patients are managed within the referral centers of the Ministry of Health of the Republic of Croatia. The services mainly involve pediatric cardiologists, cardiologists internists, cardiac surgeons and cardiac anesthesiologists with experience in this field. Besides, all other services required for comprehensive management of these patients are pulmonologists, gastroenterologists, gynecologists, endocrinologists etc.

Cardiologists often face patients who underwent one and often several cardiac surgeries or interventions, usually in childhood. Some of them underwent complete correction of the heart defect, whereas the others underwent partial or only palliative surgery. Many of them have many different problems and complications as a result of the above treatments and interventions, and also of a natural course of the disease (cyanosis, endocarditis, arrhythmias, heart failure,

Povremeno se pojavljuju i bolesnici kojima je prirođena srčana greška otkrivena tek u odrasloj dobi (ASD, Ebsteinova anomalija, kongenitalno korigirana transpozicija velikih krvnih žila, koarktacija aorte, bikuspidni aortni zalistak itd). Bolesnici sa srednje teškim i kompleksnim greškama trebali bi biti trajno usmjereno praćeni u tercijarnim specijaliziranim centrima.

U Hrvatskoj udio operativnih zahvata prirođenih i korigiranih srčanih grešaka u odraslih bolesnika (nisu uključeni ASD tip II niti operacije bikuspidne valvule) je mali u odnosu na ukupan broj kardiokirurških zahvata. Primjerice, na Klinici za kardijalnu kirurgiju Kliničkog bolničkog centra Zagreb izvede se godišnje petnaestak operacija u odraslih bolesnika s prirođenim srčanim bolestima. Najčešće su to operativni zahvati: ASD tipa sinus venosus, anomalni utoci plućnih vena, ASD tip I, zamjena pulmonalne valvule i sl.

Osim kardiokirurških zahvata, u Hrvatskoj se uspješno provode i kardiološki interventni terapijski postupci perkutanog zatvaranja ASD-a tip II i perzistentnog foramena ovale (PFO), u tri ustanove: Klinički bolnički centar Zagreb, Klinici za kardiovaskularne bolesti Magdalena te Klinički bolnički centar Rijeka. Transplantacija srca u bolesnika s prirođenom srčanom bolesti do sada je učinjena u dva navrata i to u KBC Zagreb. Određeni broj bolesnika u terminalnoj fazi bolesti s visokom plućnom vaskularnom rezistencijom liječe se i u Klinici za plućne bolesti Jordanovac i potom upućuju na eventualnu transplantaciju pluća/srce i pluća u inozemstvo.

Osim kardioloških, postoje i drugi medicinski i nemedicinski aspekti te psihosocijalni zahtjevi i potrebe ove većinom mlade populacije koja treba pomoć i skrb stručnjaka iz raznih područja. Vrlo važna područja zbrinjavanja su: nekardijalne operacije, problemi koncepcije, trudnoće, poroda, dojenja itd. Nadalje, ovi mladi ljudi trebaju usmjeravanje i ekspertna mišljenja u raznim životnim situacijama npr. bavljenja sportom, vožnje automobila, odabira zanimanja i sl. U svakodnevnom radu s ovom skupinom bolesnika potrebno je sagledati sve ove aspekte te osigurati sveobuhvatnu multidisciplinarnu i kontinuiranu zdravstvenu zaštitu.

Preporuke organizacije zdravstvene zaštite za bolesnike s prirođenim srčanim bolestima u odrasloj dobi

Prema preporukama iz literature, na svakih 2-10 milijuna stanovnika[21] trebao bi postojati jedan tercijarni centar za bolesnike s prirođenim srčanim bolestima u odrasloj dobi. Kada te podatke prenesemo na Hrvatsku, jedan takav centar trebao bi biti dovoljan. Takav tercijarni centar uključivao bi stručnjake različitih profila tj. kardiologe specijalizirane za prirođene srčane bolesti u odraslih, pedijatre, kardijalne kirurge s iskustvom u kirurgiji prirođenih srčanih grešaka, kardijalne anesteziologe, ginekologe, pulmologe, gastroenterologe, psihologe, genetičare i druge.

S kardiološke strane, za cjelovitu skrb ovih bolesnika potrebna je razvijena neinvazivna dijagnostika, u prvom redu slikovne metode (kompleksna ehokardiografija, MSCT i MR srca) te ostala neinvazivna dijagnostika (spiroergometrija, Holter EKG) uz ostale dijagnostičke metode koje se i inače koriste u kardiologiji. Budući da su aritmije glavni uzrok smrti i pogoršanja kliničkog tijeka kod mnogih kompleksnih prirođenih srčanih bolesti, od posebne je važnosti elektrofiziološka služba. Elektrofiziologija je potrebna, kako za dijagnostiku tako i za liječenje ritmoloških poremećaja u ove skupneed for re-surgery, transplantation, etc.). Patients whose congenital heart disease was detected in adulthood (ASD, Ebstein anomaly, congenitally corrected transposition of the great vessels, coarctation of the aorta, bicuspid aortic valve, etc.) come to hospital from time to time. Patients with moderate and complex defects should be continuously followed up in specialized tertiary centers.

In Croatia, the portion of surgeries of congenital and corrected heart defects in adult patients (not including ASD type II or bicuspid valve surgery) is small compared to the total number of cardiac surgeries. For example, some 15 surgeries are performed for the adult patients with congenital heart diseases on an annual basis in the Department of Cardiac Surgery of the University Hospital Center Zagreb. The most common surgical interventions: sinus venosus type ASD, anomalous pulmonary venous returns, ASD type I, pulmonary valve replacement, etc.

In addition to cardiac surgeries, therapeutic interventional cardiac procedures of percutaneous closure of ASD type II and persistent foramen ovale (PFO) are successfully performed in three institutions in Croatia: University Hospital Centre Zagreb, Clinic for Cardiovascular Diseases Magdalena and University Hospital Centre Rijeka. Heart transplantation in patients with congenital heart disease has so far been performed on two occasions in University Hospital Centre Zagreb. A certain number of patients in the end stage of the disease with high pulmonary vascular resistance are treated in the Clinic for Lung Diseases Jordanovac, and then they may be referred for lung/heart and lungs transplantation abroad.

In addition to cardiac aspects, there are some other medical and non-medical aspects and psychosocial requirements and requirements of such mostly young population needing help and care of professionals specialized in various fields. Some very important areas of management are: non-cardiac surgeries, problems of conception, pregnancy, childbirth, breastfeeding, etc. Furthermore, these young people need guidance and expert opinions in a variety of situations such as doing sports, driving a car, selection of occupation, etc. In our daily work with this group of patients, it is necessary to consider all these aspects and provide a comprehensive multidisciplinary and continuous healthcare.

Recommendations to the healthcare organization for patients with congenital heart diseases in adulthood

According to the recommendations in the literature, per every 2-10 million inhabitants[21] there should be one tertiary center for patients with congenital heart diseases in adulthood. When such data are applied to Croatia, one such center would be sufficient. Such tertiary center would include experts from different fields, that is, cardiologists specializing in congenital heart diseases in adults, pediatrics, cardiac surgeons with experience in surgery of congenital heart defects, cardiac anesthesiologists, gynecologists, pulmonologists, gastroenterologists, psychologists, geneticists etc.

In terms of cardiology, the comprehensive management of these patients requires a developed non-invasive diagnostics, primarily imaging methods (complex echocardiography, MSCT and MRI of heart) and other non-invasive diagnostics (spiroergometry, Holter ECG), along with other diagnostic methods that are commonly used in cardiology. Since arrhythmias are a major cause of death and impairment of the clinical course of many complex congenital heart diseases, electrophysiology unit is particularly important. Electrophysiology unit is required both for the diagnostics and

pine bolesnika. Razvijena invazivna i intervencijska kardiologija također je jedan od važnijih zahtjeva tercijarnog centra. Osim u dijagnostici (gdje značaj postaje manji razvojem novih neinvazivnih metoda) invazivna kardiologija postaje sve važnija u razvoju terapijskih intervencijskih pristupa kao što su: perkutana implantacija valvula, zatvaranje ASD, PFO, perzistentnog duktusa Botalli, dilatacija i ugradnja potpornica kod koarktacija, stenoza raznih provodnika, plućnih arterija i sl.

Edukacija kardiologa koji će se usko baviti prirođenim srčanim bolestima u odraslih treba biti ciljana. Kurikulum subspecijalizacijskog staža nedovoljan je za usko usmjerene kardiologe iz ovog područja. Usmjerena edukacija potrebna je i za kadrove raznih profila, liječnike drugih specijalnosti te medicinskog osoblja iz područja sestrinstva koji će činiti multidisciplinarni tim.Također je potrebno pružiti primjerenu razinu edukacije općim kardiolozima u županijskim centrima i liječnicima obiteljske medicine koji bi mogli samostalno preuzeti brigu o bolesnicima s jednostavnim PSG. Edukaciju bi trebao provoditi tercijarni centar u kojem se u potpunosti mogu zbrinuti svi oblici prirođenih srčanih bolesti kao i svi oblici komplikacija[22].

S obzirom da je zadnjih godina sve veći broj djece s prirođenim srčanim bolestima operirano bilo u našoj zemlji ili inozemstvu, u idućim godinama se očekuje sve veći broj njih koji postaju odrasli i koji će trebati organiziranu zdravstvenu zaštitu. Upravo je zbog toga potrebno što ranije uspostaviti strukturiranu, hijerarhijsku mrežu skrbi[22] koja bi uključivala tri razine: **1**. liječnike opće medicine, **2**. interniste i opće kardiologe u gradskim i županijskim bolnicama te 3. kardiologe subspecijaliste za prirođene srčane greške u tercijarnim centrima. Isto tako, važno je osvijestiti i same bolesnike te njihove obitelji o potrebi redovitog praćenja u odrasloj dobi u okviru organizirane i usmjerene skrbi ovih bolesnika.

Jedan od budućih ciljeva je i organizacija nacionalnog registra svih bolesnika s navedenim bolestima jer je pored stručnog napretka, potrebno razvijati i znanstvenoistraživačku aktivnost u ovom području, što će također povećati i kvalitetu svakodnevnog rada.

Zaključno, od posebnog značaja za medicinu i kardiologiju u Hrvatskoj je što ranije uspostavljanje adekvatne, cjelovite, multidisciplinarne skrbi za odrasle bolesnike s prirođenim srčanim bolestima, kako bi se uhvatio korak s ostatkom razvijenog svijeta, a sve u svrhu što boljeg zbrinjavanja ove sve veće skupine kompleksnih bolesnika.

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the treatment of rhythmic disorders in this group of patients. Developed invasive and interventional cardiology is also one of the major demands of the tertiary center. Except in diagnostics (which is becoming less significant due to the development of new non-invasive methods), the invasive cardiology is becoming increasingly important in the development of therapeutic intervention approaches such as: percutaneous valve implantation, closure of ASD, PFO, persistent ductus Botalli, dilation and stent implantation for coarctation, stenosis of various conductors, pulmonary arteries and the like.

Education and training of cardiologists who will closely deal with congenital heart diseases in adults should be target education and training. The curriculum of subspecialization service is insufficient for narrow specialization in cardiology of this area. Specialized training is required for personnel engaging in various fields, other specialization physicians and medical staff in the field of nursing that will form a multidisciplinary team. It is also necessary to provide an adequate level of general education and training to cardiologists in the county centers and family physicians, so that they can independently manage the patients with simple CHD. Education and training should be conducted by tertiary center where all forms of congenital heart diseases and all forms of complications can be managed[22].

Since the number of children with congenital heart diseases operated on in our country or abroad has increased in the last few years, we expect an increasing number of them who are becoming adults and will need an organized health care in the years to come. For that reason, it is necessary to establish a structured, hierarchical network of management[22] that would include the three levels: **1**. General practitioners, **2**. Internists and general cardiologists in the city and county hospitals and **3**. Cardiologists — subspecialists for congenital heart defects in tertiary centers. It is also important to make the patients and their families aware of a necessity of regular follow-up in adulthood within the program of organized and specialized management of these patients.

One of the future goals is to organize a national registry of all patients with these diseases because in addition to professional progress, it is also necessary to develop scientific and research activity in this area, which will also increase the quality of daily work.

To conclude, it is of a particular importance for the Croatian medicine and cardiology to establish an adequate, comprehensive and multidisciplinary care for adult patients with congenital heart disease as early as possible, in order to catch up with the rest of the developed world, all with the purpose of better management of this growing group of complex patients.

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