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Review Article

Adult Congenital Heart Disease is Really a Heterogenous Specialty: Message from the Colombian Adult Congenital Heart Disease Chapter

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Abstract

Congenital heart defects are the most frequent malformation. The prevalence varies among the different geographical regions of the world, due in part to the birth rate, population density and local record-keeping. With increased survival, the congenital heart population has undergone a great change, increasing the prevalence of adults with congenital heart disease.

Surgical and survival successes have turned the pediatric patient with congenital heart disease into an adult postoperative patient. It is estimated that 10%-15% of congenital heart diseases are diagnosed in adulthood in developed countries, and 30% in Latin American countries. Thus, the clinical presentation and natural evolution will be more severe. The adult´s postoperative heart is not a healthy heart, it is a transformed, ¨neo-formed¨, heart. Its original, or native, anatomy has been modified with synthetic repair elements. Postoperative patients have long, asymptomatic "honeymoon" periods; during this time, many discontinue medical follow-up, and most patients are

discharged to general adult cardiology services where the importance of ongoing care of the prior surgical repairs is not taken into account. This patient have really a heterogenous disease, which natural course and clinical presentation may be affected by the concomitant diseases acquired in adulthood. A new profile of cardiovascular patients has emerged with special attention and care needs which require the participation of several specialists to resolve the current problems. This manu-script explains why congenital heart disease in adults is actually a heterogeneous specialty, and why cardiac, and non-cardiac specialties must participate to resolve the health needs of this population. It also describes the levels of specialized training, with special interest in developing the highest training level (level III) in Colombia, and finally explains the formation of the Colombian Adult Congenital Heart Disease Chapter and the initial activities as an initiative spurred by the need to encourage the development of the specialty in Colombia.

Introduction

Congenital heart defects are the most frequent malformation. Generally, they are five times more prevalent than other types of defects, and close to 1.3 million children with congenital heart disease (CHD) are assumed to be born every year, worldwide. In the United States (US), an average of 40,000 children are born with CHD every year; in other words, there is a new case of CHD every 15 minutes, and, of these, 25% require surgical intervention with-in the first year of life, due to the severity of the defect. In Latin America, 54,000 children with CHD are born each year, representing 150 new cases per day. The prevalence varies among the different geographical regions of the world, due in part to the birth rate, popula-tion density and local record-keeping. A recent study published by Liu Y *et al.* shows that the birth prevalence of CHD from 1970 - 2017 progressively increased to a maximum in the 2010 -17 period of 9.410/1,000 [95% confidence interval (CI) 8.602-10.253]. This repre-sented a significant increase compared to the fifteen prior years (P¼ 0.031) [1]. As science advances, with the inclusion of new high-resolution imaging technologies which allow more precise and earlier diagnoses, the number of cases recorded at birth increases. For all types of CHD, the prevalence is generally highest in Asia with almost 10 cases for each 1,000 live births (LBs). It is followed by Oceania (9 -10 cases per 1,000 LBs) and North America (8-9 cases per 1,000 LBs), with similar rates in Europe and South America (8 cases per 1,000 LBs), and, finally, low rates in Africa, due to poor records (3-4 cases per 1,000 LBs) [1].

Discussions

Generalities Regarding the Origin and Development of Pediatric Cardiology

For many years, CHD care has focused on childhood, since it is an important cause of mortality in children under five years of age, especially in developing countries. The specialty tasked with providing all the initial treatment has classically been pediatric cardiology. This is a relatively young medical specialty, having been constituted as a subspecialty of cardiology in the mid-twentieth century.

Various historical events have marked the development of this specialty around the world. Towards the end of the 18th century, Morgagni created the anatomic and physiological bases of CHD. In his book, he

described ventricular septal defects and the single ventricle, confirming Leonardo Da Vinci´s observations from the 15th and 16th centuries. The first book written about "human heart malformations" on record is that of Thomas Peacock in 1858. In this text, he correlated the anatomical and clinical data and pioneered the description of Tetralogy of Fallot.

The initial papers in America began with the contributions of the Canadian physician Maude Elizabeth Seymour Abbott in 1905, who wrote the chapter on "Congenital Heart Disease" for Dr. Osler´s book *System of Modern Medicine*. In 1936, she published her book Atlas of *Congenital Heart Disease*, which describes a significant number of CHDs, represented in more than 400 cases of different anatomical components. Her book went down in history as it provided the essential elements for the emergence, growth and development of cardiac surgery and pediatric cardiology. She is thought to be the person who placed the study of children with CDH on the international scene. Due to their great contributions, Dr. Abbot, Dr. Taussig, Dr. Nadas and Dr. Keith are recognized as pioneers of modern pediatric cardiology.

In Latin America, according to the contributions of Dr. Fause Attie (1935-2009), former Director General of the Instituto Nacional de Cardiología Ignacio Chávez [Ignacio Chávez National Institute of Cardiology] in Mexico, there are four determining factors in the de-velopment of the specialty [2]:

1.1. Integration of technological development with clinical management of congenital heart disease

1.2. Modification of the natural history: pioneers in congenital heart disease

1.3. Rational grounds for medical-surgical treatment

1.4 Pediatric cardiology: A constantly evolving specialty

Regarding Dr. Fause Attie´s remarks, I would like to concentrate on the fourth point. Based on my expert opinion, I highlight pediatric cardiology´s constant evolution. New subspecialties have emerged in pediatric cardiology (hemodynamics, electrophysiology, echocardiography, intensive care, and cardiac rehabilitation, among others). The integration of new high-definition imaging technologies which allow the highly pre-cise study of congenital malformations beginning in early embryonic stages led to the development of fetal echocar-diography and the emergence of fetal cardiology as a CHD-integrated specialty. There is constant development in this field, not just from a diagnostic but also a therapeutic perspec-tive, modifying the natu-ral history of the disease [3,4]. Other specialists have been linked to the field such as gynecologists, perina-tologists, neonatologists, intensive care pediatri-cians, anesthesiologists, pediatric cardiovascular surgeons, geneticists and pediatric cardiologist echocardiographists, all working as a team.

In summary, prenatal diagnosis has advanced greatly; today, the sensitivity of an extensive cardiac echocar-diography examination is 89.6% [5,6]. Concurrently over the last 50 years, there have been great advances in the development of pediatric cardiology. With the newly incorporated subspecialties, the post-surgical results have improved dramatically. Consequently, the current survival rate is more than 85% in almost all countries in the world [7].

Congenital Heart Disease is Really a Heterogenous Disease

As more patients with CHDs of varying complexity reach adulthood, we learn more about the evolution of congenital heart disease. Today we understand why the same CHD re-paired in childhood does not evolve in the same way in two adults. The answer lies in the heterogeneity of the defect. The same CHD has different degrees of severity and therefore a different clinical presentation in childhood (for example, pulmonary atresia with an intact septum may have different degrees of right ventricular and pulmonary branch hypoplasia, be associated or not with various degrees of tricuspid regurgitation, have or not have sinusoids, and have or not have other associated defects, among others). The therapeutic management and initial approach tend to vary according to each particular case, from needing palliative procedures prior to repair (systemic-pulmonary fistula), to undergoing first intention repair. Consequently, the natural evolution and clinical presentation will be different in two adults, even though we are dealing with the same CHD. The natural course and clinical presentation will also be affected by the concomitant diseases acquired in adulthood, as well as whether or not target organs have been affected (for example, liver dysfunction in Fontan, kidney failure), which adds severity to the clinical presentation and prognosis [7].

The New Adult Cardiovascular Patient (Neo-Formed Hearts)

With increased survival, the congenital heart population has undergone a great change, increasing the prevalence of adults with CHD (ACHDs). A new profile of cardiovascular patients has emerged with special attention and care needs which require the participation of several specialists to resolve the current problems, analyzing each particular case. This population should never be discharged from cardiology. The concept of cure through a surgical intervention should be eliminated. This is only accepted for simple CHD cases according to the 2001 Bethesda classification [8].

Surgical and survival successes have turned the pediatric patient with CHD into an adult postoperative CHD patient. The adult´s postoperative heart is not a healthy heart, it is a transformed, ¨neo-formed¨, heart. Its original, or native, anatomy has been modified with synthetic CHD repair elements (patches, valves, intra or extra-cardiac tubes, stents) which modify not only its primary anatomy but its whole physiology [9].

Postoperative ACHDs have long, asymptomatic "honeymoon" periods; during this time, many discontinue medical follow-up. This stage frequently coincides with the transition from adolescence to young adulthood. Most patients are discharged to general adult cardiology services where the importance of ongoing care of the prior surgical repairs is not taken into account; often, the CHD repair elements deteriorate and show natural wear and tear. For medium and high complexity CHDs, even under the best circumstances and with an initial surgical repair in childhood or adolescence, the residuals and sequelae of the necessary CHD repair will continue to evolve throughout time. This will lead to hemodynamic changes which will produce clinical repercussions, functional class deterioration, and, ultimately, an inevitable relapse. During the asymptomatic period, which may even last for decades, losses to follow-up reach over 70% [10].

The ACHD population is derived from two main groups (Figure 1).

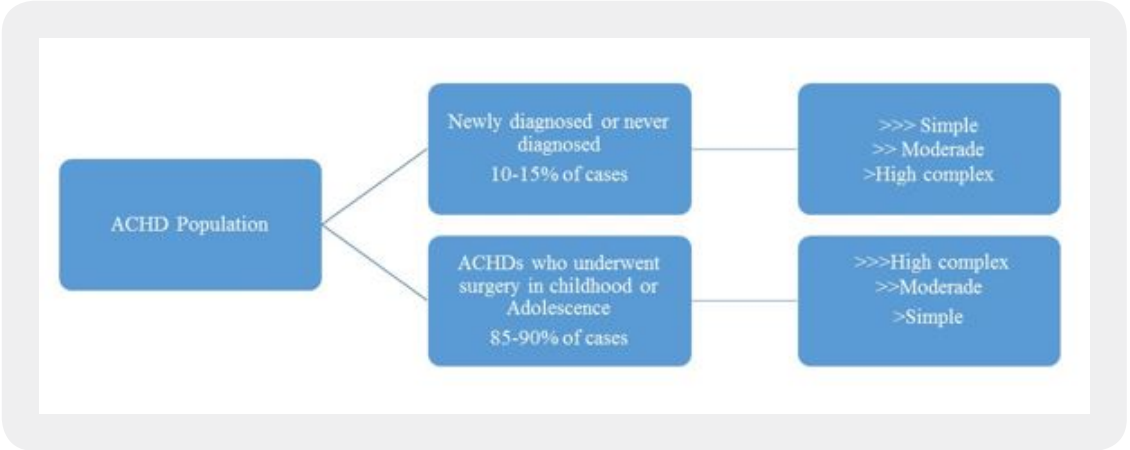

Figure 1: *Current ACHD population from two main groups*

Generally, it is accepted that up to 10% or at the most 15% of CHDs with are diagnosed in adulthood. This is accepted for developed countries, with most of these being low complexity congenital defects. For developing countries, the scenario is different, with more cases of adults not having been diagnosed early and not having been treated adequately, and the diagnosis of complex defects is not uncommon. Thus, the clinical presentation and natural evolution will be more severe.

A recent study by Márquez González *et al.* in a high complexity hospital in Mexico showed that of 3,483 CHD cases, adults represented almost 30% of the newly diagnosed cases and another 30% were undertreated (diagnosed and treated in childhood, but, for various reasons, did not continue or finish treatment for their CHD [11]).

The New Adult Congenital Heart Disease Specialty: General, Expert and Specialist Levels

The complexity of CHDs demands a high level of understanding of the defects. Given the heterogeneity of ACHDs, knowing how to approach each particular case and make appropriate decisions is a challenge. Previously, little was known about the natural evolution of CHDs in adults. Complex defects were almost nonexistent; if they were not treated in childhood, they simply did not survive. Most ACHDs had simple defects, and there was a certain degree of unconcern for this population. Efforts were aimed at developing pediatric cardiology units, especially in developing countries. However, due to the rapid growth in ACHD prevalence in developed countries, this situation has reversed, with more adults than children with CHD today. Consequently, and of necessity, the adult congenital heart disease specialty has been created worldwide.

In America in December 2012, after a joint petition by the American Board of Pediatrics and the American Board of Internal Medicine, ACHD was formally recognized by the American Board of Medical Specialties as a subspecialty of adult and pediatric cardiology. As such, advanced training is needed to qualify a fellow to sit for the ACHD Board exami-nation and to be an ACHD cardiologist. Advanced ACHD training is for an additional two years after completion of either pediatric cardiology or adult cardiovascular disease training [12].

Currently, three levels of training in ACHD are recognized:

Level I-General or Basic: level of those who have basic adult cardiology training, but have not carried out specific training in CHD. This is the basic training required to become a competent cardiovascular consultant, is required for all cardiovascular fellows, and can be accomplished as part of a standard three-year training program in cardiovascular medicine. This level of training should allow the trainee to develop sufficient knowledge to review and understand consensus-based ACHD care guidelines, to determine which ACHD pa-tients can be managed by a clinician and which patients are best cared for in collaboration with an ACHD specialist. This level of preparation only equips them to care for simple cases.

Level II-Expert or Advanced: level achieved through additional training in ACHD to acquire experience in managing the whole spectrum of patients. Expertise in ACHD is gained by focused experience during training and practice. The training ranges from six months to one year. From a practical perspective, it may be difficult to identify clinicians with expertise in ACHD. Those whose expertise was acquired before the development of formal certification programs and those trained outside the United States who may also have different pathways to achieve ACHD expertise are also considered to be experts.

Level III-Specialist: level of those from a pediatric or adult cardiologist background who have complete training. Requires additional training and experience beyond the cardiovascular fellowship for the trainee to acquire specialized knowledge and experience in performing, interpreting, and training others to perform specific procedures or render advanced specialized care for specific conditions at a high level of skill. In the case of ACHD, Level III training provides the knowledge needed by graduates wishing to make an advanced clinical and/or academic/research commitment to this field and not only become competent in the care of the entire spectrum of ACHD patients, but also participate in teaching about ACHD [13].

The objectives of this specialized training are:

4.1. Management of the transition from adolescence to adulthood

4.2. Recognition of concomitant adult medical conditions

4.3. Knowledge of electrophysiology in ACHD care

4.4. Management of advanced heart failure and determination of transplant candidacy in ACHD patients

4.5. Understanding the unique aspects of caring for cyanotic adults with Eisenmenger syn-drome and pulmonary vascular disease

4.6. Recognition of the importance of palliative care

4.7. Understanding mental health and cognitive outcomes

4.8. Assessing the safety of participation in sports and exercise

4.9. Recognition of women's reproductive health: contraception and pregnancy

4.10. Assessment of sexual function

4.11. Knowledge of the legislative aspects of employment and advocacy

Participation of Parallel Specialties and Non-Cardiac Specialties (Heterogeneity of Care)

The highly variable clinical presentation of adults with the same CHD has taught us that not all cases should be approached and treated in the same way. The appearance of concomitant diseases in adulthood may add severity to the clinical expression of the CHD. The progression and decompensation manifest as heart failure, arrhythmias, syncope, bleeding, pulmonary arterial hypertension, endocarditis, pulmonary thromboembolism and stroke, among others. The CHD itself may cause damage to important organs and systems such as the liver, kidney, central nervous system, and hematopoietic, immunologic, endocrine, respiratory and gastrointestinal systems.

These findings have been included in the most recent 2018 AHA/ACC guidelines for the management of adults with congenital heart disease, to establish the new anatomic and functional classification, taking the functional stages (I-IV) of the New York Heart Association (NYHA) as a reference and combining them with nine clinical variables: hypoxemia; pulmonary hypertension/pulmonary arterial hypertension; hemodynamically significant shunt; venous and arterial stenosis; exercise capacity; and endorgan dysfunction; which, if present, add severity to the adult´s anatomic and functional class [14].

Thus, we conclude that CHDs are really a heterogenous disease, in which not only the ACHD specialist should participate, but also other non-cardiovascular adult specialties such as: nephrology, pulmonology, neurology, hematology, endocrinology, high-risk obstetrics and gynecology, cardiac pathology, psychology, psychiatry, social work, genetics, gastroenterology, and infectious disease, among others. Other specialties should also actively partic-ipate, such as: hemodynamics, electrophysiology, cardiac rehabilitation, heart failure and intensive care. Finally, other specialties parallel to cardiology should participate, such as cardiovascular diagnostic and interventionist radiology and cardiovascular anesthesiology.

The Creation of the Colombian Adult Congenital Heart Disease Chapter and Its Initial Activities

The chapter was founded in mid-2017 as an initiative spurred by the need to encourage the development of the specialty in Colombia. It seeks to bring together specialists who are active in the management and care of ACHDs to work on academic activities, research, education and professional growth cooperation, to provide specialized care to ACHDs.

It began with six members who were excited to develop the project, supplying ideas and initiatives. By the end of 2018, it already included 20 members from different regions of the country and several cardiology specialties. It has participated in various academic activi-ties at the national and international level, including congresses, symposia, conferences, and webinars with live feeds in several Latin American countries. It has a space on the Sociedad Colombiana de Cardiología´s [Colombian Society of Cardiology´s] web site [15], and has produced more than 40 scholarly works which include articles, book chapters, editorials, conferences, interviews, among others. Of note, the chapter has participated in the most recent American College of Cardiology (ACC) Latin America Conference 2019, the Congreso Interamericano de Cardiología [Interamerican Congress of Cardiology], and the 2nd International Conference on Cardiology & Heart Diseases in Berlin, Germany [16-18].

At the time of this writing, the chapter is composed of 41 members (Figures 2-3) who are seeking to conform a heterogenous team of specialists, become increasingly consolidated as a working group with national cooperation, and be a reference for other countries in Latin America. In order to extend its growth, the 1st National and International Symposium on Adult Congenital Heart Disease was carried out in September 2019 [19]. This event was held for the first time in Colombia, with the participation of several chapter members from the fields of pediatric and adult cardiology, cardiovascular surgery, cardiovascular radiology, elecrophysiology and hemodynamics. International guests from Spain and the US also participated. Conferences with high-level scientific content were presented, and clinically important topics were treated for mutual learning among the specialties. Topics on cardiovascular embryology and heart defects, the initial assessment and approach to ACHD, the transfer process from pediatric cardiology to ACHD care, the approach to and management of arrythmias in ACHD, the hemodynamic study of CHDs, CHD assessment using cardiac resonance and tomography, Fontan circulation, single ventricle and adult complications, pulmonary arterial hypertension in ACHD, congenitally corrected transposition of the great arteries in adults, right ventricle assessment in ACHD, and pregnancy and CHD, along with clinical cases, were presented for discussion and opinion, with a large audience. The activities will continue in the following years, developing necessary and current topics. The chapter is constantly linked with the Interamerican ACHD Council of the Interamerican Society of Cardiology, with many prospects and plans for joint work.

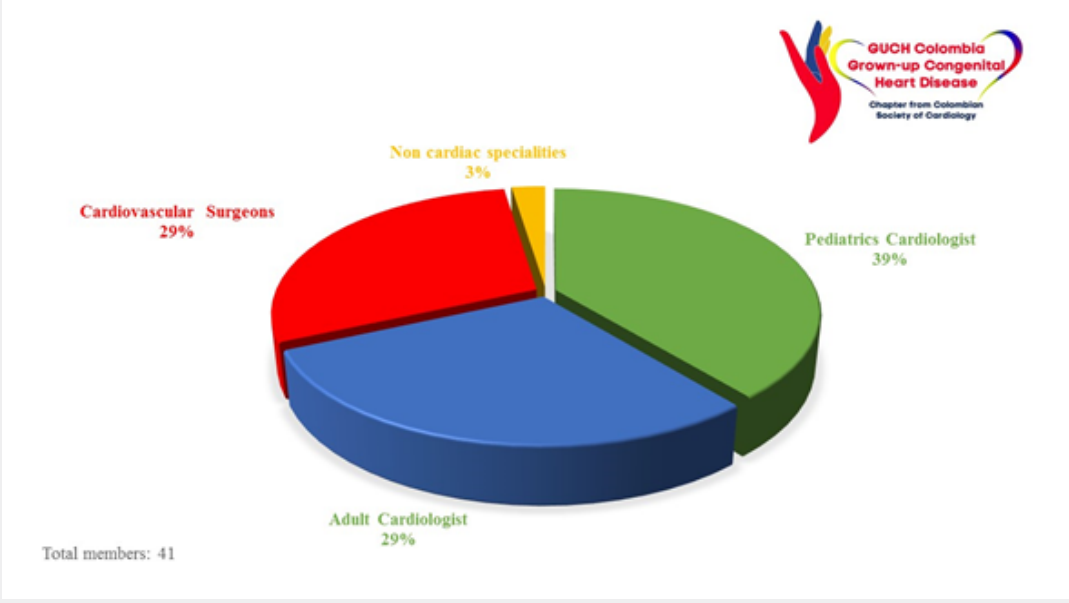

Figure 2: *Who integrate Colombian adult congenital disease chapter at current date*

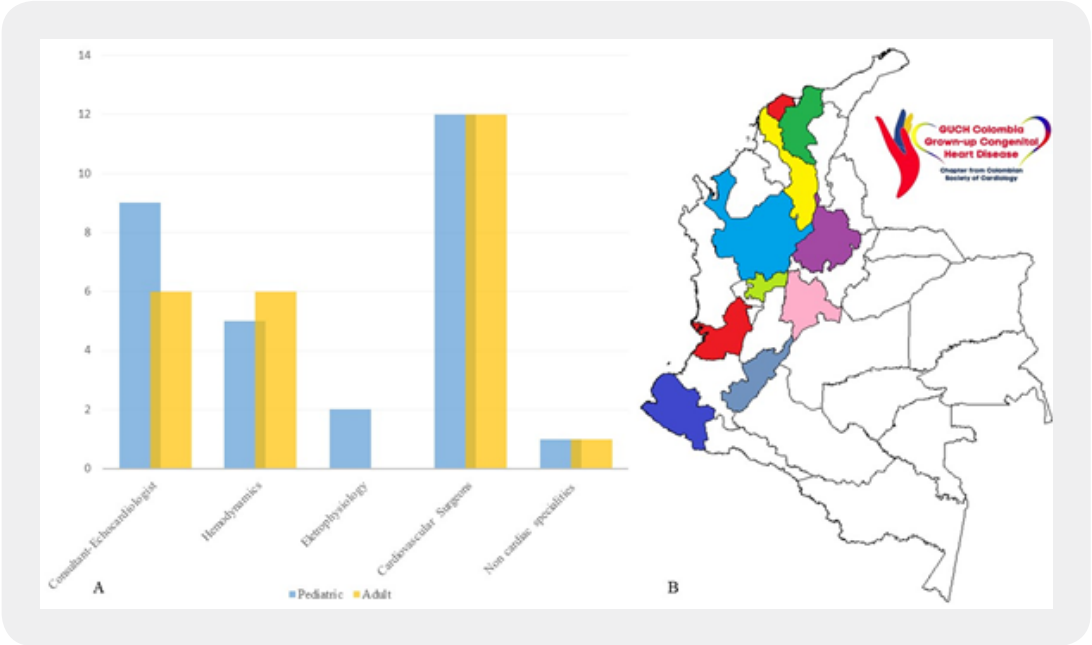

Figure 3: *A. Cardiac and non-cardiac specialties (cardiovascular radiologist) who integrate adult con-genital disease chapter at current date. B. Chapter members around Colombia at current date.*

Conclusions

This manuscript would like to make it clear that adult CHDs are a heterogenous group, in which various adult specialties (both cardiologic and non-cardiologic) need to participate. The various care needs and causes of decompensation in adult CHDs must be identified and adequately treated by several specialists. The first step is to train specialists with Level III training. To begin this path, the Colombian Adult Congenital Heart Disease Chapter was founded, which has had a rapid growth and acceptance in the local and Latin American medical community. This chapter will continue to be active and develop the necessary topics for the care of ACHDs, a population which is growing rapidly throughout the world and especially in Latin America.

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Conflicts of Interests

I have no conflicts of interest to declare.

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