H36 Congenital Heart Defects (CHD) in Children: A Retrospective Review of Autopsies From the Cook County Medical Examiner's Office

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After attending this presentation, attendees will understand the types of CHD commonly encountered in autopsies of children.

This presentation will impact the forensic science community by providing information about the anatomical defects (simple or complex, single or multiple) and the medical history in CHD cases of a medical examiner's pediatric population.

CHDs represent the most common type of birth defect. According to the Centers for Disease Control and Prevention, they affect nearly 1% of births per year in the United States. Anatomically and functionally, CHDs can be broadly classified as simple (involving one anatomic structure) or complex (involving multiple structures) with repairs varying in complexity. While some simple defects, such as atrial or ventricular septal defects, may resolve spontaneously, most CHDs require surgical correction at some point in life, depending on symptoms.

The files of the Cook County Medical Examiner's Office in Chicago were searched for cases of subjects less than 18 years of age involving "congenital heart disease" and "congenital heart defects" as a cause of death, from January 2006 to July 2016. Cases were reviewed for age, sex, race, cause and manner of death, medical history, and autopsy findings.

In this study population, simple defects consisted of Atrial Septal Defects (ASD), Ventricular Septal Defects (VSD), Patent Ductus Arteriosus (PDA), anomalies of the valves, and coarctation of the aorta. Complex defects consisted of Tetralogy Of Fallot (TOF), Hypoplastic Left Heart (HLH), Double Outlet Right Ventricle (DORV), and Transposition of the Great Arteries (TGA).

A total of 64 cases were identified: 35 males and 29 females; 38 African American and 26 Caucasian. Regarding age, 23 were ≤1 month old, 29 ranged between one month - one year, seven ranged between one-ten years old, and five were older than ten years. The cause of death was listed as "congenital heart disease" in all cases but two, where it was undetermined. The manner of death was determined to be natural in all cases but two (undetermined manner).

There were 31 (48%) decedents with a history of CHD prior to autopsy and, of these, 24 underwent a surgical repair. Five cases had a CHD associated with genetic syndromes (mostly Down syndrome).

Regarding the simple defects, coarctation of the aorta was observed in six cases. ASDs were observed in 4 cases as the only defect and in 4 in association with other defects. VSDs were never observed alone; in all six cases they were in association with other defects. PDA was observed alone in 1 case and in association with other malformations in five cases. Valvular stenosis was observed in 1 case alone and was associated with other defects in four cases.

Regarding the complex defects, TOF was observed in four cases. HLH was seen in 11 cases, DORV in seven cases, TGA in five cases, and valvular atresia in ten cases. AVSDs were observed in five cases alone and in association with other defects in six cases. Ebstein's anomaly was seen in six cases: one alone and five associated with other conditions.

This study reveals that, in the cases surveyed, children below one-year-old represented the vast majority of subjects (81%) with CHD. The most common simple defect was ASD (eight cases). The most frequent complex defects weres HLH and ASD (both observed in 11 children). Frequently undiagnosed conditions at autopsy were coarctation of the aorta (four out of six), HLH (five out of 11) and ASD (five out of 11). One case of TOF was not diagnosed prior to autopsy. When diagnosed, HLH, ASD and coarctation of the aorta were commonly surgically treated (4 out of six cases, five out of six cases, one out of two cases, respectively), while TOF was always treated.

It is important to recognize CHD as a potential cause of sudden unexpected death in children. Despite advances in prenatal care and imaging, a significant number of cases of CHD go undiagnosed prior to autopsy (52% in this series). In children, and especially in infants, it is important to carefully assess for the presence of undiagnosed CHD as a potential cause of death, consulting a cardiac pathologist, if possible.

Congenital Heart Defects, Children, Sudden Death