Commentary

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Children Born with a Congenital Heart Defect: Risk Factors and Protective Factors

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Description

With an estimated prevalence of 4 to 50 per 1000 live births, cardiac malformations at birth account for a significant portion of clinically significant birth defects and are an important component of pediatric cardiovascular disease.For instance, it is estimated that between 4 and 10 liveborn infants per 1000 have a cardiac defect, and forty percent of these defects are discovered in the infant's first year of life.However, the actual prevalence may be much higher.This estimate typically excludes, for instance, the most prevalent cardiac defect, the bicuspid aortic valve.A bicuspid aortic valve affects 10 to 20 people in every 1,000 people and is linked to significant morbidity and mortality in later life.Bicuspid aortic valve is highly heritable, both on its own and in combination with other cardiovascular conditions, particularly disorders of the left ventricular outflow tract.The incidence of cardiac malformations approaches 50 per 1000 live births when isolated aneurysms of the atrial septum and persistent left superior vena cava, which each occur in 5 to 10 per 1000 live births, are taken into account.In two separate cohorts of 5000 serial newborns and 5000 serial premature infants in Israel, it has also been demonstrated that the prevalence of ventricular septal defect (VSD) is as high as 5%.A conservative estimate of the prevalence of CHD is 50 per 1000 live births, taking into account the aforementioned factors.

The current state of the art in the treatment of most CHD includes extraordinary diagnostic precision, definitive therapies, and relatively low morbidity and mortality (such as the arterial switch operation for transposition of the great arteries or device closure of intracardiac shunts).As a result of these treatments, an increasing number of people with CHD will live to adulthood and may be able to conceive.Even though CHD diagnosis and treatment have come a long way, our understanding of its causes is still limited, but it has improved in recent years.Some forms of CHD, such as hypoplastic left heart syndrome, are still associated with significant morbidity and mortality, despite the numerous cutting-edge treatments that are currently available for a number of heart defects.It will be possible to define disease risk and gain insight into the pathobiological basis of the congenital heart problem with a better understanding of possible causes.For the following reasons, it is critical for a child's pediatrician to ascertain whether there is an underlying genetic pattern (such as deletions, duplications, or mutations):1) There may be involvement of additional significant organ systems;2) clinical outcome prognostic information may exist; (3) The family ought to be aware of significant genetic risks to reproduction;and (4) there may be additional members of the family for whom genetic testing is necessary.The methods for evaluating infants and children with CHD that are currently available are discussed in the sections that follow.

Prevalence of CHD

The most common congenital malformation, cardiac anomaly is mostly caused by genetic or chromosomal changes.It typically manifests externally within the first year of life and fundamentally depends on the hemodynamic response.Studies of autopsies indicate that mortality is highest at this age.

The majority of infants admitted to the Royal Brompton Hospital in England were neonates, according to a study of infants under the age of one year.

The majority of congenital heart disease patients in our study were infants, followed by neonates.Even though our evaluation looked at patients in the outpatient care unit and looked at hospitalized children at the Royal Brompton Hospital, we notice a difference in when the patients were referred.

Shunt heart defects with pulmonary venocapillary hypertension, such as ventricular septal defect and persistent ductus arteriosus, which were prevalent in our study, accounted for the majority of the diagnosis of congenital heart disease in infants.However, we should keep in mind that many of the children in our study came from rural areas or other states, which may have delayed their access to specialized facilities.

We did not frequently have complex defects like hypoplastic left ventricle and transposition of the great vessels, which were prevalent in other studies and showed up in the first few days of life.We may assume that many of these patients cannot receive specialized care in time or may even die without a diagnosis due to their early death.Considering that the control group is comprised of the same population of children, the delay in weight gain is significant.This difference may have been influenced by the impact on hemodynamics.

Loci and Genes Associated With CHD

Growth delays are common in children with congenital heart disease, pulmonary hypertension, heart failure, and a significant left-to-right shunt.However, there is no linear correlation between the degree of impairment and the degree of hypoxia.

Congenital heart diseases vary widely in frequency and prevalence, according to epidemiological studies.The development of technology and the widespread use of echocardiography have both contributed to an improvement in the process of making a diagnosis and, as a result, an increase in the incidence of some heart defects.Recent research shows that ventricular septal defects occur most frequently, at a frequency of 41.6% in the Samanek and Vorskova 11 study and 15.7% in the NERICP series.

Numerous studies have demonstrated that the beginning of treatment has an impact on a newborn's survival with major congenital heart disease.In duct-dependent lesions, a delay in diagnosis can result in detrimental duct closure and progressive cardiac insufficiency, as well as the risks of hypoxia, acidosis, hemodynamic collapse, brain damage, and multiorgan failure.In such circumstances, emergency surgical intervention for corrective or palliative procedures carries the risk of postoperative cardiac failure and may have an impact on early and long-term morbidity as well as the development of later physical and psychomotor skills.If a prenatal diagnosis is made, the appropriate treatment can be started before the baby is born in a reputable tertiary care facility.Numerous studies have failed to demonstrate a clear benefit of prenatal diagnosis, despite this potential advantage;In the prenatal series, some even showed negative outcomes.These contradictory outcomes could be due to a number of different factors.