Congenital heart disease research paper sample

Science, Genetics

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Congenital Heart Valve Disease

The congenital heart disease refers to the defects in the functioning and structure of the heart that occur during conception, and is said to take place somewhere between the 3rd and 10th week of pregnancy. According to research, the prevalence of Congenital Heart Disease may vary from about 4 to 10 for every 1, 000 live births. It was noted that the frequency of the disease increased as more adults are diagnosed to be suffering from critical CHD from 1985 until 2000, where there are almost equal cases in the number of adults and children who are detected to have a severe congenital heart abnormality (Marelli et al, 2007). Interestingly, while the majority of infants who are born with the Congenital Heart Disease, does not suffer from other forms of inborn abnormality, in a recent study, it was revealed that certain abnormalities also occur in about 20 to 40 percent of the CHD cases. It also claimed that about 30% of children who suffer from the abnormality of the chromosomes are diagnosed with Congenital Heart Disease (Richards and Garg, 2010). In addition to that, it was discovered that 50% of patients who are born with Trisomy 21 or Down Syndrome are likely to suffer from the CHD, that may range from defects of the atrial and ventricular septal to that

of atrioventricular canal lesion types. In relation to genetic factors, studies of human DNA revealed that the mutation of Fibrillin 1 (FBN1) was the result of the Marfan syndrome (Richards and Garg, 2010). The Marfan syndrome is a hereditary defect that upsets the body’s connective tissues, a typical indication is a progressive dilation of the aortic root, with a predisposition to dissection, dislocated lens, and abnormal skeletal conditions (Richards and Garg, 2010). On the other hand, in cases when a patient suffers from the Trisomy 13, or Patau Syndrome, the frequency of the CHD increases up to 80% (Richards and Garg, 2010). The Trisomy 13 is the medical term coined for genetic disorders where an individual is born, not with two, but three genetic material of Chromose 13 (National, 2015).

The recent breakthroughs in medical science provides a better understanding of congenital heart disease, one among the most common causes of death among infants and children. The advancement in genetic technology and the accomplishments of the international scientific community on the Human Genome Project, results for the determination of a single gene defect that is now associated with syndromes link to congenital heart diseases. There is an increasing evidence suggesting the involvement of the presence of a single gene mutation in a broad spectrum of genes in the functioning of the cardiovascular system. The Molecular Cardiology and Neuromuscular Insititute in New Jersey claimed that discrete mutations in certain nuclear transcription factors which are the proteins that play an important regulatory function during cardiovascular development and morphogenesis, has the possibility to effect Pleiotropic cardiac malformations (Garcia-Marin, 209). The earliest transcription factors that are

expressed in the developing heart and are vital in the activation of cardiac-specific genes are the GATA4, Nkx2. 5, HAND2, TFAP2, and Tbx5, however, the genetic mutations of these transcription factors may result in severe cardiac defects. For instance, the GATA4, NKX2-5, HAND2, TGAP2B and TBX5 are linked to septal abnormalities, conduction defects, right ventricular hypoplasia, patent ductus arteriosus in Char syndrome, and Holt-Oram syndrome, respectively (Marin-Garcia, 2009). These emphasize the critical effect of disruption of the normal and early stages of development and biological processes to that of the progression of congenital heart disease.

The Transcription Factors

The GATA4 is a transcription factor that is vital for proper embryonic and cardiac development. In a study conducted among 100 patients who are diagnosed of CHD, 19 forms of genetic mutations were observed in 1 that is linked to Atrial Septal abnormalities, 2 associated with Tetralogy of Fallot and 3 with an intense association with Ventricular Septal Defect (Matapally et al, 2015).

The Transcription Factor TBX5 is another Cardiac transcription factor, the main function of which is to regulate the cardiac development and the mutations in the genes that are responsible for its functioning are the key elements that causes CHD. For example, the mutations in TBX5 that is responsible for encoding the T-box transcription factor is defined as the main cause of Holt-Oram syndrome, which is an autosomal and leading abnormality that chiefly disturbs the normal functioning of the heart and that of the upper limbs. After the identification of the DNA binding elements and the typical targets, it was established through a functional analysis of the

TBX5 that the majority of the genetic mutations leads in TBX5 proteins that are defective in DNA binding, transcriptional activity and/-or protein-protein interactions (Ghosh et al, 014). There are other studies that substantiate the link between TBX5 and the development of CHDs; recently, there are about 70 genetic mutations that have been detected among patients suffering from CHD (Ghosh et al, 2014). Interesting to note is the relationship or interaction of TBX5 with NKX2. 5 and GATA4, other transcription factors that also play critical roles in cardiac development. Genetic mapping revealed the occurrence of mutations in the genes encoding these proteins in patients with CHD showing the overlapping clinical structures (Ghosh et al, 2014). Along with this, other TBX5 have been recognized, and among these interacting proteins that are important in heart development is the MEF2C. MEF2C is an interacting factors that associates with TBX5 and is needed during the early stages of heart development (Ghosh et al, 2014).

The transcription factor AP2 (Tfap2) is a key element in the epidermal and migratory cell development of the neural crest in human and other vertebrate embryos. They are the group of genes that are first released in the ectoderm and they determine the results within the epidermis/crest by the utilizing direct and indirect means (Hoffman et al, 2007). Accordingly, the Tfap2 family originated from one genetic ancestry that go through the process of gene duplication to distribute to the other 5 members among living vertebrates. Other than being an ancestral gene that affects the gain of novel roles for different subfamily members in patterning the epidermis, the Tfap2 gene family is vital in cardiac morphogenesis (Kusuma et al, 2011).

In summary, recent advances in medical technology allowed for the ability of scholars trace the effects of genetics in the development of congenital heart disease. It is interesting how genetic mapping provided a means to identify the genes that are involved in the main development of the cardiac structure and function. Although the current approaches that link chromosomal and genetic factors has not fully achieved the objective of fully elucidating the relationship between CHD and genetics, science has greatly made a step towards better understanding the disease and the factors that affects its development.

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