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Congenital heart disease and their association with genetic syndromes observed in paediatric cases

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Congenital heart disease is one of the leading causes of morbidity and mortality in children and is one of the most common causes of congenital anomalies. CHD which is a birth defect in the structure of the heart has been frequently observed in association with genetic syndromes. Patients in addition to heart defects, also exhibit extracardiac abnormalities such as facial dysmorphism, gastrointestinal problems, and other developmental problems. Common genetic syndromes such as Down syndrome, DiGeorge syndrome, Noonan syndrome, Edward syndrome, Patau syndrome, Turner syndrome, Klinefelter syndrome and William syndrome have been found to have an association with CHD. The aim of the study is to determine the frequency of paediatric cases with a genetic syndrome and CHD in a hospital based population. Genetic tests such as karyotyping, fluorescence in-situ hybridization (FISH) and DNA mutation analysis were employed to test for the presence of a genetic abnormality in patients suffering from CHD. A total of 204 CHD cases suspected for genetic syndromes were referred for genetic testing during the period of October 2012 to July 2017 of which, 32% of the patients had an association with a genetic syndrome. Down syndrome (14%) followed by DiGeorge syndrome (10%) were found to be strongly associated with CHD. This link portrays the importance of genetic testing in patients with CHD as it can help in the diagnosis, management, and prognosis of their condition.

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