William’s syndrome: Case report

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A 7-year old boy was diagnosed with William’s syndrome clinically with the help of some investigations. This child having malocclusion of multiple teeth with dental carries, large forehead, small chin, puffiness around both eye, looks like “elfin facies” with low IQ. He has a systolic murmur (Grade-3/6) in the base of the heart. Echo, Doppler gradient found in the just supra-valvular area, 114mmHg (Supra-valvular Aortic stenosis) & MVP (Mitra valve prolapse). This child also has blockage of naso-lacrimal duct, causing watering of eye. William's syndrome (WS) is a neuro-developmental, multi-system genetic disorder characterized by distinctive personality traits, facial dysmorphisom (“elfin face”) and congenital cardiac defects, of which supra-valvula aortic stenosis is the most common lesion found. It is characterized by congenital heart defects (CHD), Skeletal and renal anomalies, cognitive disorder, social personality disorder and dysmorphic facies. WS is a rare familial multi-system disorder occurring in 1 per 20,000 live births.

Biography

Gobinda Kanti Paul has completed his MD in Cardiology and Post-graduation degree from NICVD under BSMMU in January 2008. He has published more than 10 publications in various index journals. He has passion for non-intervention cardiology specially echocardiography and preventive cardiology. Currently, he is an Assistant Professor/Consultant Cardiology and Residential Physician (RP) in the Mymensingh Medical College and Hospital. He also has a special interest in rheumatic fever and rheumatic heart disease and its prevention.

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