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Varying Clinical Presentation of Williams Syndrome: A Case Series

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ABSTRACT Williams syndrome (OMIM 194050) is a rare multisystem genetic disorder with an incidence of 1/ 75000 which usually occurs sporadically caused by the deletion of 26 contiguous genes, including elastin (ELN) (OMIM 130160) on chromosome 7q11.23. The researchers present here three cases of Williams syndrome with cardiac anomalies and varying clinical presentation. In this paper the researchers suggest a defined protocol with more attention while evaluating cardiac anomalies in childhood period, especially when the patient has facial dysmorphism or developmental delay.