CASE REPORT

INFANTILE SYSTEMIC HYALINOSIS DUE TO HOMOZYGOUS DELETION MUTATION (C.1074DELT) IN ANTHRAX TOXIN RECEPTOR2

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Abstract: *Infantile systemic hyalinosis is an autosomal recessive disorder characterized by hyaline deposits in the papillary dermis and other tissues leading to a progressive and fatal clinical course. The diagnosis is usually based on characteristic clinical findings, followed by confirmation with molecular genetic testing. We report two infants with inherited systemic hyalinosis, one had the diagnosis confirmed by molecular genetic testing identifying the homozygous c.1074delT mutation in the ANTXR2 gene, highlighting the importance of genetic confirmation even in resource-limited settings.*

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