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Infantile systemic hyalinosis: Report of the first Guyanese case and review of the literature

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Infantile systemic hyalinosis (ISH) is a rare autosomal recessive disorder characterized by diffuse hyaline deposit in the skin, muscles, glands and gastrointestinal tract. Typically, ISH occurs within the first week of the life and is characterized by joint contractures, osteopenia, failure to thrive, gingival hypertrophy, protein-losing enteropathy with intractable diarrhea, thickened skin and frequent infections. We report a patient with ISH confirmed by clinical and histopathologic findings. DNA sequencing of the *CMG2* gene (aka *ANTXR2*) revealed compound heterozygocity for two mutations: c.697G>T in the donor site of exon 8, which affects splicing, and c.858_859delAG (p.Lys286Asnfs) in exon 10, which cause a frameshift with premature termination of the translation. The differential diagnosis was congenital syphilis or Caffey disease. Although there is currently no effective treatment for this disease which is usually fatal by the age of 2 years, identification of the mutations in *CMG2* (also known as *ANTXR2*) makes DNA-based prenatal diagnosis feasible for subsequent pregnancies.

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