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Infantile systemic hyalinosis

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Description

A 6-year-old dystrophic girl presented to the pediatric clinic's emergency department with infected wounds and ulcerated tumors on her ears and buttocks. Her parents recounted a history of progressive joint contractures since the age of 4 months, alongside the emergence of growing papules on her face (Figure 1A) and ears, plus a severe gingival hyperplasia. With time, subcutaneous nodules and tumors appeared on her hands, feet, back, and buttocks. The progression of the girl´s skin lesions intensified, resulting in increased pain and ulcerations (Figure 1B, 1C). External genetic testing unveiled a homozygous mutation in the human anthrax toxin-2 gene, leading to the diagnosis of an infantile systemic hyalinosis. This condition triggers the deposition of hyaline material in the dermis and other tissues, impacting multiple organs except the brain. Intellectual ability is thus not impaired. The clinical manifestation of papulonodular skin lesions, gingival hyperplasia, joint contractures, failure to thrive, and recurrent pulmonary infections strongly suggests an inherited deposition disorder. Regrettably, there is currently no specific treatment available for this devastating disease, and management options remain confined to supportive care.

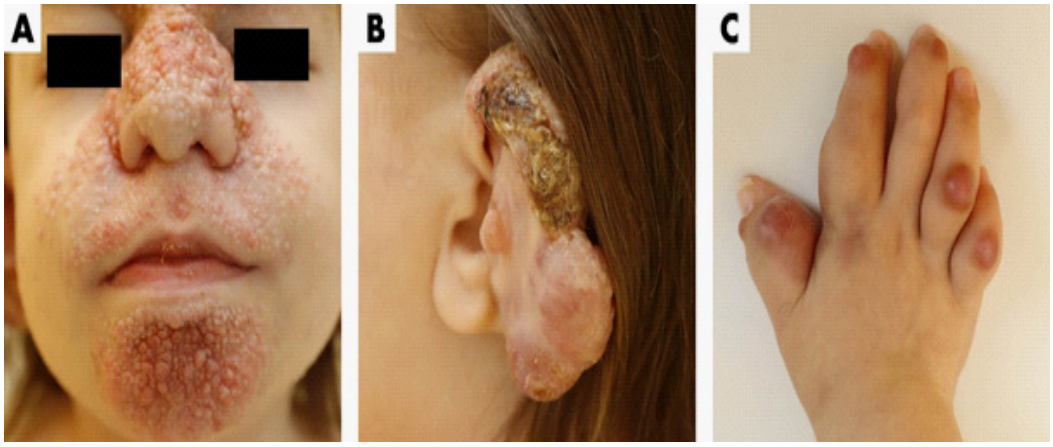

Figure 1: Clinical images of patient.

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