Acrodysostosis: A case report.

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Abstract:

Acrodysostosis is a rare syndrome characterised by peripheral dysostosis (gross shortening of hands and feet), mental retardation, hypoplasia of nose and hypoplasia of maxilla. An 11 year old female child presented to Civil Hospital Ahmedabad with complaints of short stubby fingers. On detailed general examination, patient had dysmorphic facial features, depressed nasal bridge, upturned nose, gingival hyperplasia and dental malocclusion with skeletal survey suggestive of hypoplastic nasal bone, shortening of metacarpal, metatarsal bone & phalangeal bones and early fusion of epiphysis of distal end of radius and ulna; all pointing towards the diagnosis of acrodysostosis. Any patient presenting with short stature, a short stubby fingers, and acrodysostosis should be suspected. No specific therapy is available.

Key Words: Acrodysostosis, Nasal hypoplasia, Short stubby fingers.

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