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Three-year clinical investigation of a Chinese child with craniometaphyseal dysplasia caused by a mutated *ANKH* gene: A case report

Three-year clinical investigation of craniometaphyseal dysplasia

Abstract

BACKGROUND

Craniometaphyseal dysplasia (CMD) is a rare genetic disorder. Autosomal dominant CMD (AD-CMD) is caused by mutations in the *ANKH* gene. Affected individuals typically have distinctive facial features including progressive thickening of the craniofacial bones. Treatment for AD-CMD primarily consists of surgical intervention to release compression of the cranial nerves and the brain stem/spinal cord. To alleviate the progression of the clinical course and improve the quality of life in children waiting

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Author: Sha Yu, Wen-xia Chen, Wei Lu, Chao Ch… **Publish Year:** 2020

Novel heterozygous GATA3 and SLC34A3 variants in a 6-year ...

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The patient, a 6-year-old boy, was the first **child** of noncon-sanguineous **Chinese** parents with no history of renal hypo/ **dysplasia**, deafness, or hypoparathyroidism. After a normal pregnancy, he was born by spontaneous vaginal delivery at 38 weeks of gestation. Apgar scores were 10/1 and 10/5. Growth parameters showed birth weight of 3,200 g (in the

Author: Sha Yu, Wen-xia Chen, Wei Lu, Chao Ch… **Publish Year:** 2020

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