
SPOT DIAGNOSIS (IMAGE GALLERY)



CONGENITAL SKELETAL DEFECT

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A term male child appropriate for gestational age with birth weight of 2.6kg born to a primigravida unregistered 22 years old mother by normal vaginal delivery was found to have the short limbs and shortened toe-like fingers and rudimentary grape like toes. (Figure 1).

What is the diagnosis?

Grebe`s syndrome. It is a rare autosomal recessive genetic disorder caused by mutation in CDMP1 gene. Children with Grebe syndrome have considerable physical limitations and normal intelligence.

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