

# Two Cases of Costello Syndrome and Literatures Review

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**【Objective】** To investigate the clinical features of HRAS-associated Costello Syndrome.

**【Method】** Characteristics of clinical data and gene mutation of two cases Costello Syndrome in Department of Pediatrics of Sun Yat-Sen Memorial Hospital, Sun Yat-Sen University were retrospectively analyzed. The related literature was searched by using search terms “Costello” or “HRAS” Syndrome” .

**【Result】** Both patients were presented with mental retardation, growth retardation,postnatal feeding difficulties and characteristic facial appearance,and some cases also exhibit cardiovascular, skeletal muscle or central nervous system abnormalities.Two casea carried the same HRAS gene mutation site Exon2 c.34g>a P. (Gly12Ser).

**【Conclusion】** Costello syndrome is a rare multisystem disorder accompanied by tumor predisposition.It is vital to comfirm the diagnosis through the identifcation of a specifc germline mutation in the HRAS .High forehead、 curly hair, thick lips , slack skins and HRas gene sequencing are helpful to the diagnosis. In addition,early intervention treatment and tumor monitoring are necessary.



Fig.1 Typical facial features of the patient 1: curly hair, broad forehead, low-set ears, wide nasal bridge, full lips and large mouth, and deep palmar creases as well as loose, soft skin.



Fig.2] (a-d) Typical facial features of the patient 2: sparse, curly hair, broad forehead, low-set ears, wide nasal bridge, full lips and large mouth, and deep palmar creases as well as increased pigmentation.



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