

Broad thumbs and broad hallux: the hallmarks for the Rubinstein-Taybi syndrome

Polegares e hálux alargados: as marcas para o diagnóstico da síndrome de Rubinstein-Taybi

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A 30-year-old man presented to our hospital with developmental delay, recurrent respiratory infections and short stature. Examination showed broad thumbs and hallux, beaked nose and big toes. Rubinstein-Taybi syndrome (RTS) was diagnosed based on typical clinical features.

RTS was described by Rubinstein and Taybi in 1963¹. They reported a patient with big toes, craniofacial

abnormalities and mental retardation. The broad thumbs and first toes are the hallmarks for the syndrome. The diagnosis is based on clinical features (Figure). RTS is an autosomal dominant inherited disease that usually occurs as result of a *de novo* mutation and can also be associated to eye abnormalities, hearing loss and cardiac defects^{2,3}.

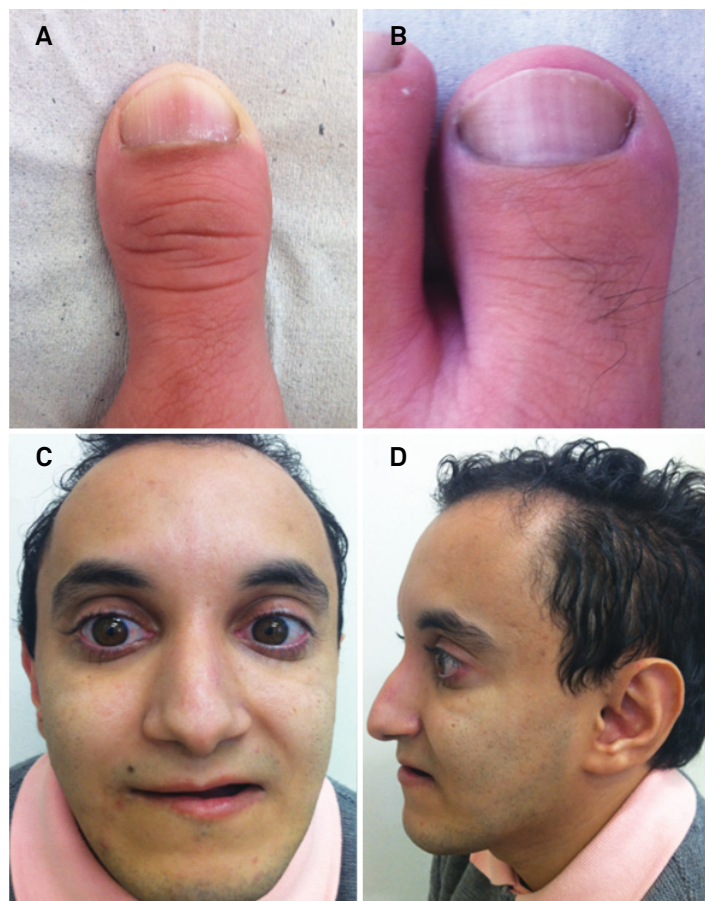


Figure. Note the broad thumbs (A) and broad hallux (B), the typical features described in Rubinstein-Taybi syndrome. Craniofacial abnormalities characterized by downslanting palpebral fissures, arched eyebrows, columella extending below the nares and beaked nose were also seen (C and D).

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Conflict of interest: There is no conflict of interest to declare.

Received 16 June 2013; Received in final form 02 July 2013; Accepted 09 July 2013.

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