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## Intellectual disability-sparse hairbrachydactyly syndrome

INSFRM

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Intellectual disability-sparse hair-brachydactyly syndrome</u>. ORPHA:3051

Intellectual disability-sparse hair-brachydactyly syndrome is a very rare condition of unknown etiology consisting of short stature, hypotrichosis, brachydactyly with coneshaped epiphyses, epilepsy and severe mental delay. After the initial delineation of this syndrome by Nicolaides and Baraitser in 1993, only five more patients were published in the literature up to now.

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