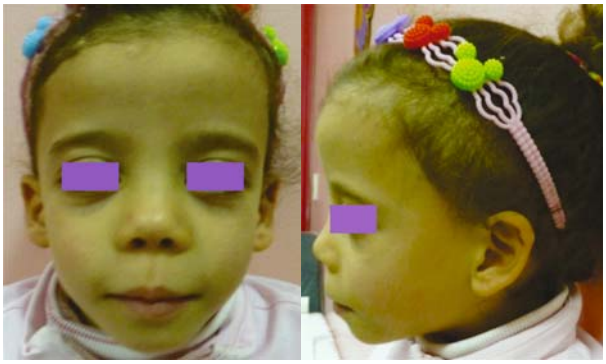
3M Syndrome: A Rare Cause of Short Stature

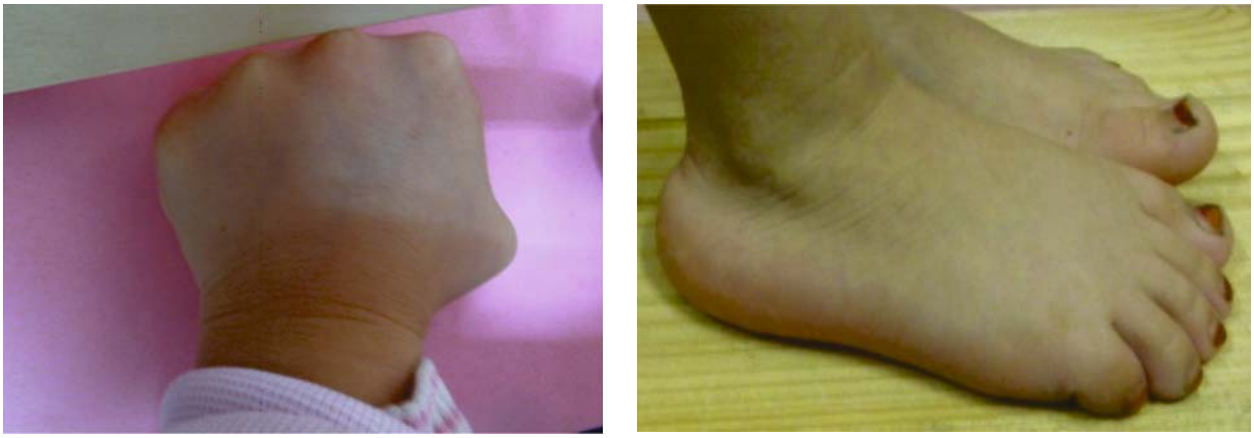
The Miller-McKusick-Malvaux (3M) syndrome (OHIM #273750) is a rare autosomal recessive disease characterized by severe intrauterine and postnatal growth retardation with dysmorphic facial features called gloomy face, and skeletal abnormalities.

The characteristic features, present at birth are: a relatively large head, dolichocephaly, frontal bumps (***Fig*. 1 a** and **b**), a triangular face, a pointed chin (***Fig*. 1 a**), an up-turned nose, full lips, provided eyebrows, a long philtrum, and malar hypoplasia (***Fig*. 1 a**). Skeletal abnormalities include short neck, prominent trapezius, square shoulders, short thorax, pectus excavatum, hyperlordosis,very flexible joints, short fifth finger (***Fig*. 2 a**) and short and prominent heels (***Fig*. 2 b**) which are almost pathognomonic of the syndrome.



(a) *(b)*

Fig. 1. *Dysomorphic features in a child with 3M syndrome. (a) Triangle-shaped face with frontal bumps, hypoplastic midface, provided eyebrows, an upturned nose with a fleshy tip, long philtrum, full lips and a pointed chin; (b) Frontal bumps, upturned nose and long phitru.*



(a) *(b)*

Fig. 2 *Short fifth finger (a), and short prominent heels (b) in a child with 3M syndrome.*

Differential diagnoses include Silver-Russel Syndrome, Dubowitz Syndrome, MULIBREY Dwarfism and Fetal Alcohol Syndrome. There is no specific treatment for 3M syndrome. However, the use of recombinant human GH for the treatment of short stature has been suggested.

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