

## IMAGES IN DERMATOLOGY

### Ski-Jump Toenails<sup>☆</sup>

### Uñas de pies en «salto de esquí»



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Figure 1

A 9-month-old breastfeeding baby boy was seen at our department with abnormalities of the toenails and congenital bilateral edema of the feet. The paternal grandmother and a paternal female cousin had also presented congenital

lymphedema and abnormalities of the toenails. Examination revealed *ski-jump* koilonychia in the nails of the first toe of both feet and edema of the toes with slight edema of the back of both feet (Fig. 1A and B). The suspected diagnosis was Milroy disease (MD) and this was confirmed by means of a genetic study and presence in the germ line of a heterozygous mutation on p.Gly933Arg (G933R) of the FLT4 gene. This mutation was also detected in the father, a brother, and a paternal female cousin of the patient.

MD is characterized by congenital lymphedema in the lower limbs caused by mutations in the FLT4 gene, which synthesizes the protein of receptor 3 of the vascular endothelial

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growth factor (VEGFR3), responsible for the development of the lymphatic system. The spoon or ski-jump nails are characteristic but only appear in between 10% and 14% of patients, in whom it may be a guiding sign. Heredity is

autosomal dominant with variable expression and incomplete penetrance, estimated at between 85% and 90%. This explains why the disease was expressed phenotypically by our patient but not by his father or brother.