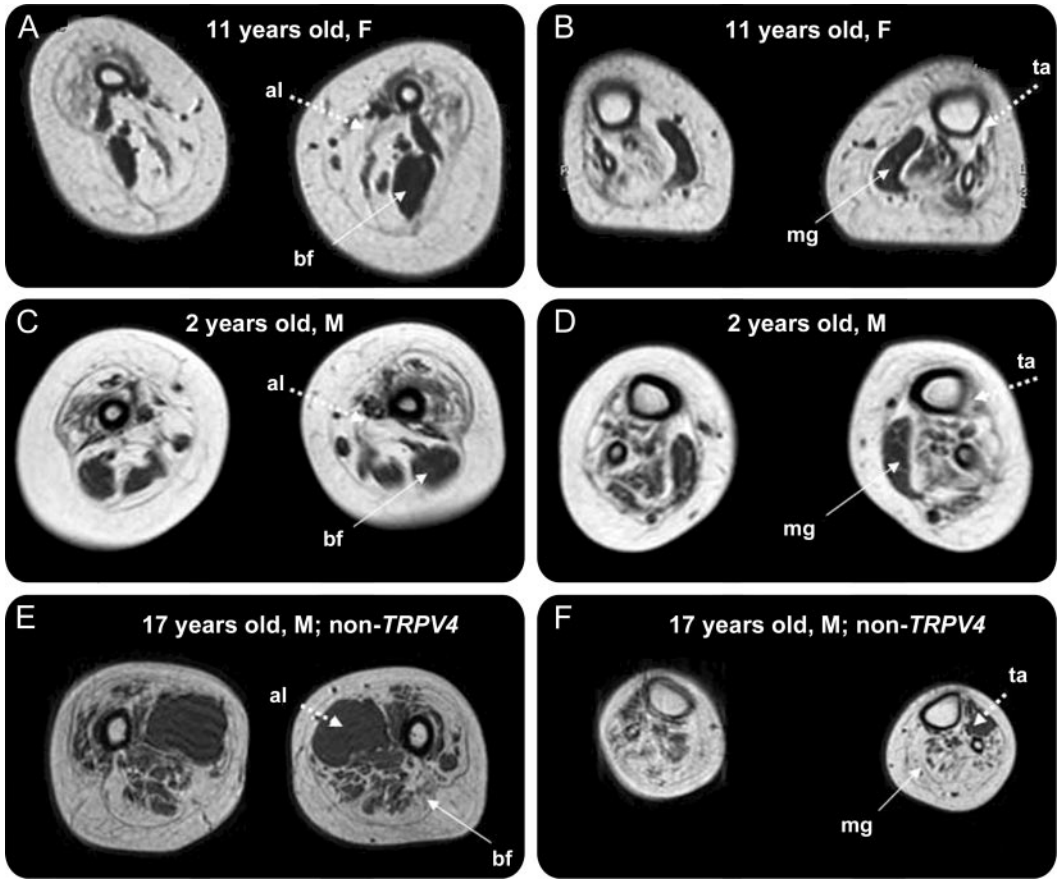
Muscle MRI in *TRPV4*-related congenital distal SMA

Figure **Transverse T1-weighted MRI of thighs and calf muscles in distal spinal muscular atrophy (dSMA) patients harboring mutations in transient receptor potential vanilloid 4 (*TRPV4*)**



Mutations in the *TRPV4* gene[1] are associated with autosomal dominant congenital dSMA, scapuloperoneal SMA, hereditary motor-sensory neuropathy 2C, and orthopedic clinical conditions ranging from spondylomethaphyseal dysplasia to lethal neonatal metatropic dysplasia. In 2 sporadic patients (A–D) with dSMA harboring de novo mutations in *TRPV4*, an extensive fatty atrophy preserving biceps femoris (bf) and medial gastrocnemius (mg) was present at muscle MRI. This pattern is different from non-*TRPV4* patients[2] (E–F) where the medial compartment at thighs (with hypertrophy of adductor longus [al]) and anterior muscles at calf level (tibialis anterior [ta]) are spared.

A 2-year-old boy and an 11-year-old girl showing marked weakness in proximal and distal muscles, atrophy of distal legs, and clubfoot were investigated for congenital spinal muscular atrophy (SMA) as suggested by EMG and muscle biopsy. Both children, who had normal *SMN1* gene testing, harbored mutations (p.P97R; p.R232C) in *TRPV4*.[1] MRI of muscle showed similar severe changes preserving biceps femoris in the lateral compartment of the thighs and medial gastrocnemius in the posteromedial calves (figure). The pattern of myoimaging, unrelated to disease duration, is not seen in other congenital distal SMA[2] and might facilitate appropriate molecular analyses.

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