Duplication of 10q24 locus: broadening the clinical and radiological spectrum.

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Résumé en anglais	Split-hand-split-foot malformation (SHFM) is a rare condition that occurs in 1 in 8500-25,000 newborns and accounts for 15% of all limb reduction defects. SHFM is heterogeneous and can be isolated, associated with other malformations, or syndromic. The mode of inheritance is mostly autosomal dominant with incomplete penetrance, but can be X-linked or autosomal recessive. Seven loci are currently known: SHFM1 at 7q21.2q22.1 (DLX5 gene), SHFM2 at Xq26, SHFM3 at 10q24q25, SHFM4 at 3q27 (TP63 gene), SHFM5 at 2q31 and SHFM6 as a result of variants in WNT10B (chromosome 12q13). Duplications at 17p13.3 are seen in SHFM when isolated or associated with long bone deficiency. Tandem genomic duplications at chromosome 10q24 involving at least the DACTYLIN gene are associated with SHFM3. No point variant in any of the genes residing within the region has been identified so far, but duplication of exon 1 of the BTRC gene may explain the phenotype, with likely complex alterations of gene regulation mechanisms that would impair limb morphogenesis. We report on 32 new index cases identified by array-CGH and/or by qPCR, including some prenatal ones, leading to termination for the most severe. Twenty-two cases were presenting with SHFM and 7 with monodactyly only. Three had an overlapping phenotype. Additional findings were identified in 5 (renal dysplasia, cutis aplasia, hypogonadism and agenesis of corpus callosum with hydrocephalus). We present their clinical and radiological findings and review the literature on this rearrangement that seems to be one of the most frequent cause of SHFM.

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