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## ECHO

### Autosomal dominant Weill-Marchesani and Marfan syndromes are two sides of the same coin



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A genetic study has suggested that autosomal dominant Weill-Marchesani syndrome and Marfan syndrome are allelic diseases.

Two large families with autosomal dominant Weill-Marchesani syndrome were studied. They were the same families whose condition was previously shown to be linked to chromosome 15q21.1, to the fibrillin-1 gene locus. Fibrillin-1 mutations cause Marfan syndrome, and the two syndromes are clinically similar.

In family 1 Weill-Marchesani syndrome was consistent with linkage to chromosome 15q21.1 and chromosome 19p13.3–p13.2; in family 2 linkage to chromosome 19 was excluded for all six affected members. A deletion in exon 41 of the fibrillin-1 gene was apparent in family 1, and sequence analysis showed heterozygosity for a 24 nucleotide in frame deletion, which segregated with affected family members but was not present in 186 controls of European origin. No mutation was identified in family 2, maybe because of a low rate of mutation, as in Marfan syndrome.

Affected members of both families had their DNA analysed for genetic linkage to 19p13.3–p13.2 and PCR products were sequenced for the fibrillin-1 gene for one family member initially, and other members as necessary.

Weill-Marchesani syndrome can show autosomal dominant or autosomal recessive inheritance, though the clinical features of each are identical. The investigators had already shown that autosomal recessive Weill-Marchesani syndrome mapped to chromosome 19q13.3–13.2 in two large families of Lebanese and Saudi origin. Optical features of the syndrome are microsphaerophakia, with dislocation of lenses, severe short sight, and glaucoma.

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## Autosomal dominant Weill-Marchesani and Marfan syndromes are two sides of the same coin

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