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Myotonic dystrophy type 1 (DM1) is an autosomal dominant multisystemic disorder caused by the expansion of an unstable CTG trinucleotide repeat at chromosome region 19q13.3.1 The number of CTG repeats is unstable in the abnormal range and usually increases in size in successive generations, in association with anticipation.2,3 A decrease in the CTG repeat size during transmission from parents to child can also occur in about 6.4% of transmissions, most frequently during paternal transmissions (10%).4 In the French-Canadian DM1 population, intergenerational contractions occur in about 7.4% of transmissions, all in cases of paternal transmission. We report here 2 French-Canadian DM1 families with paternal transmission, both originating from the Saguenay-Lac St-Jean, in which all affected children display CTG contractions. Although it was already reported that intergenerational contractions could be observed in several sibs in a same family, it was not noted whether this occurred in all affected sibs.4 Overall, these observations support the existence of a paternal factor that prevents CTG expansion.

Case reports. The pedigree of the 2 families and Southern blot analysis are shown in the figure.

Family A. No clinical information was available for I-1. II-4 was diagnosed with DM1 at age 38. He had the classic manifestations of the disease with distal muscle weakness and wasting, myotonia, and cataracts, with a Muscular Impairment Rating Scale (MIRS) of 3.5 Molecular analysis by Southern blot revealed a CTG repeat size of about 500. His son and daughter were evaluated at age 32 and 36. The son...
had myotonia and hypersomnia but no muscle weakness, whereas the daughter was clinically asymptomatic, with an MIRS of 1. Molecular analysis revealed a CTG repeat size of 360 and 260 repeats for the son and the daughter. The brother of II-4, II-5 was diagnosed with DM1 at age 48. He had muscle weakness and wasting, myotonia, a first-degree heart block, and cataracts, which had been extracted at age 30. The MIRS was 4. Molecular analysis revealed a CTG repeat size of 630. His son and daughter were diagnosed with DM1 at age 25 and 27. Both were clinically asymptomatic, with a MIRS of 1. Molecular analysis revealed a CTG repeat size of 260 repeats for both children. A new evaluation of the daughter at age 42 revealed that she was still clinically asymptomatic.

**Family B.** I-1 was diagnosed with DM1 at age 50. He had the classic manifestations of the disease with distal muscle weakness and wasting, myotonia, cataracts, and an MIRS of 3. Molecular analysis revealed a CTG repeat size of 500. I-1 had 4 affected children who were diagnosed with DM1 at age 30, 34, 35, and 36. All were clinically asymptomatic with an MIRS of 1. Molecular analysis revealed a CTG repeat size of 250 for all 4 children. Subjects II-5 and II-6 both have 2 affected children. All were clinically asymptomatic with a CTG repeat size of 210 at age 10 (III-9), 5 (III-10), 7 (III-12), and 5 (III-13).

**Discussion.** We report 2 DM1 families with paternal transmission in which a CTG repeat size contraction was observed in all affected descendants. In family A, the contraction was observed in all affected children from 2 branches and, in family B, a contraction was observed in all affected children and has remained stable in the next generation. There is only one reported case in which a CTG repeat size contraction in one child was associated with a concomitant expansion in the brother, during paternal transmission.6 In this latter study, however, a possible bias may exist because of the weak differences in the size of the CTG repeats between the father and the children (833 for the father and 500, 667, and 1,000 for the children) and because the molecular analysis was performed at different times.

This observation raises the possibility that, in cases of paternal transmission, when a contraction is observed in a sibling, intergenerational contractions could be observed in all affected sibs, in different branches of the same family, and could remain stable across further generations. Because this is of major importance for genetic counseling, this needs to be confirmed in a large number of similar families.

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