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A novel FKR...related muscular dystrophy founder mutation in South African Afrikaner patients with a phenotype suggestive of a dystrophinopathy.

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Author information

Abstract

BACKGROUND: Fukutin-related protein (FKRP) muscular dystrophy is an autosomal recessive disorder caused by mutations in the FKR... gene. The condition is often misdiagnosed as a dystrophinopathy. A previously unreported mutation, c.1100T>C in exon 4 of FKR..., had been identified in homozygous form in two white South African (SA) Afrikaner patients clinically diagnosed with a dystrophinopathy.

OBJECTIVES: To investigate whether the c.1100T>C mutation and the common European FKR... mutation c.826C>A are present in other patients of Afrikaner origin with suspected dystrophinopathy, and whether a founder haplotype exists.

METHODS: The c.1100T>C mutation was initially tested for using an amplification refractory mutation system technique in 45 white SA Afrikaner patients who had tested negative using multiplex ligation probe amplification screening for exonic deletions/duplications in the dystrophin gene. Sequencing analysis was used to confirm the c.1100T>C mutation and screen for the c.826C>A mutation. Two cohorts (each numbering 100) of Afrikaans and other white controls were screened for the c.1100T>C and c.826C>A mutations, respectively.

RESULTS: Of the 45 patients, 8 patients (17.8%) were homozygous for c.1100T>C, 2 (4.4%) were compound heterozygotes for c.1100T>C and c.826C>A, and 1 (2.2%) was heterozygous for c.1100T>C with a second unidentified mutation. The c.1100T>C mutation was found in 1/100 controls, but no heterozygotes for the c.826C>A mutation were identified. Linked marker analysis for c.1100T>C showed a common haplotype, suggesting a probable founder mutation in the SA Afrikaner population.

CONCLUSION: FKR... mutations may be relatively common in Afrikaners, and screening should be considered in patients who have a suggestive phenotype and test negative for a dystrophinopathy.

This test will be useful for offering diagnostic, carrier and prenatal testing for affected individuals and their families. As FKRP muscular dystrophy is autosomal recessive in inheritance, the implications of a positive diagnosis in a family differ significantly from those of an X-linked dystrophinopathy.

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