A new spastic paraplegia gene.

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A genome-wide scan using high density SNP microarrays identified a single homozygous candidate region exceeding the significant multipoint LOD score of +3 in a large consanguineous Middle Eastern family with autosomal recessive spastic paraplegia. Mutation screening in the coding exons of all candidate genes highlighted one single missense variant affecting a strongly conserved amino-acid that was absent in >500 Caucasian controls. Three additional families were found mutated in this gene. The mapping and subsequent identification of a novel autosomal recessive spastic paraplegia gene demonstrates the extensive genetic heterogeneity of this condition.