E180splice mutation in the growth hormone receptor gene in a Chilean family with growth hormone insensitivity: A probable common mediterranean ancestor

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Mutations in the GH receptor gene have been identified as the cause of growth hormone insensitivity syndrome (GHIS), a rare autosomal recessive disorder. We studied the clinical and biochemical characteristics and the coding sequence and intron-exon boundaries of the GH receptor gene in a consanguineous family with severe short stature which consisted of two patients, their parents and five siblings. The two adolescents had heights of -4.7 and -5.5 SDS, respectively, with elevated growth hormone associated with low IGF-I, IGFBP-3 and GHBP concentrations. Molecular analysis of the GH receptor gene revealed a mutation in exon 6, present in both patients This mutation, E180 splice, has been previously described in an Ecuadorian cohort, and in one Israeli and six Brazilian patients. We determined the GH receptor haplotypes based on six polymorphic sites in intron 9. Co-segregation of the E180splice mutation with haplotype I was found in this family, compatible with a common Mediterranean a