Leukemia Inhibitory Factor Receptor

**Alternative Names**
LIFR

**OMIM Number**
151443

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
5p13.1

**Description**
There is evidence that mutations in the leukemia inhibitory factor receptor gene cause Stuve-Wiedemann/Schwartz-Jampel type 2 syndrome. The leukemia inhibitory factor is a polyfunctional cytokine that affects the differentiation, survival, and proliferation of a wide variety of cells in the adult and the embryo.

**Molecular Genetics**
The leukemia inhibitory factor receptor gene contains 20 exons and encodes a 1,097-amino acid trans-membrane protein that is composed of 6 different domains: 2 cytokine receptor homology domains, 1 immunoglobulin-like domain, 1 type III fibronectin domain with 3 modules, 1 trans-membrane domain, and 1 cytoplasmic domain.

**Epidemiology in the Arab World**

**Oman**
In three Omani families originating from the United Arab Emirates, all with consanguineous parents, Dagoneau et al. (2004) found that children with Stuve-Wiedemann/Schwartz-Jampel type 2 syndrome had a 1-bp insertion, 653_654insT, in exon 6 of the LIFR gene causing frameshift and a stop 2 codons downstream. The presence of this mutation in other families from the United Arab Emirates lead Dagoneau et al. (2004) to suggest a founder effect in the region [See also: United Arab Emirates > Dagoneau et al. (2004)].

**United Arab Emirates**
Through a study of a series of 19 patients with Stuve-Wiedemann Syndrome/Schwartz-Jampel Syndrome Type 2, Dagoneau et al. (2004) mapped the disease gene to chromosome 5p13.1 at D5S418 and identified null mutations in the leukemia inhibitory factor receptor gene. An identical frameshift insertion (653_654insT) was identified in five Omani [See also: Oman > Dagoneau et al. (2004)] and Yemeni [See also: Yemen > Dagoneau et al. (2004)] consanguineous families from the United Arab Emirates, suggesting founder effect. The authors concluded that Stuve-Wiedemann Syndrome and Schwartz-Jampel Syndrome Type 2 are indeed a single clinically and genetically homogeneous condition due to null mutations in the leukemia inhibitory factor receptor gene.

**Yemen**
In two Yemeni families originating from the United Arab Emirates, both with consanguineous parents, Dagoneau et al. (2004) found that children with Stuve-Wiedemann/Schwartz-Jampel type 2 syndrome had a 1-bp insertion, 653_654insT, in exon 6 of the LIFR gene causing frameshift and a stop 2 codons downstream. The presence of this mutation in other families from the United Arab Emirates lead Dagoneau et al. (2004) to suggest a founder effect in the region [See also: United Arab Emirates > Dagoneau et al. (2004)].

**References**

Contributors
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