

# **Clinical, biochemical and molecular investigations of three Taiwanese children with Laron syndrome**

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摘要

## **Abstract**

Three children of two Taiwanese families were diagnosed with Laron syndrome, two sisters and one boy. Both sets of parents were consanguineous. Clinically, all three presented with the typical craniofacies of Laron syndrome, consisting of prominent forehead and hypoplastic nasal bridge, high-pitched voice, short stature, and central obesity. Biochemically, their levels of serum IGF-I were less than 5 microg/ml before and after an IGF-I generation test, and levels of IGFBP-3 were reduced in all three patients. Sequence analysis of the growth hormone receptor gene revealed that all three carried a homozygous missense D152G mutation in exon 6.