Diagnosis of Severe Primary IGF-I Deficiency

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Clinical Presentation of Primary IGFD

Severe primary IGF-I deficiency (SPIGFD) generally presents as classical **Laron syndrome** but non-classical cases with mild or moderate phenotypes should also be considered.

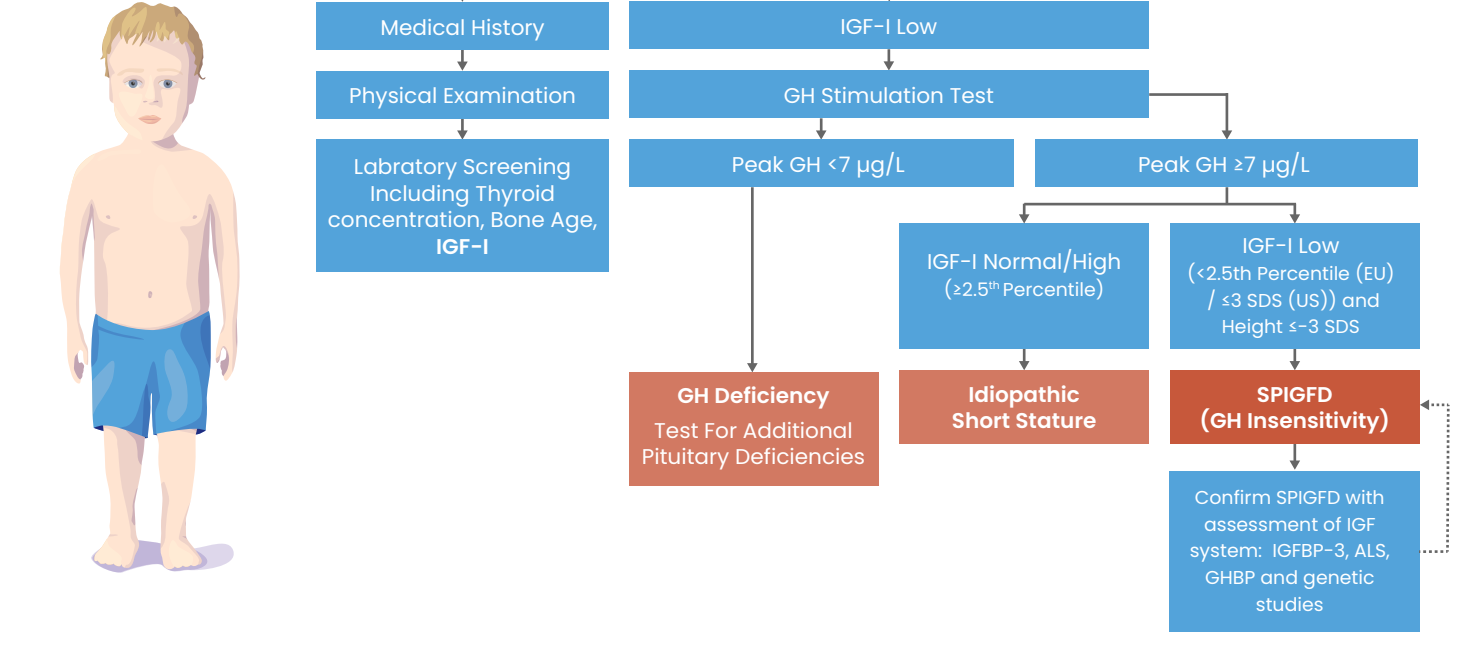
Clinical Features

- **Proportional Short Stature**
- **Frontal Bossing**
- **Underdeveloped Muscles**
- **Delayed Puberty**
- **Midfacial Hypoplasia**
- **Truncal Obesity**
- **High-pitched Voice**
- **Acromicria**
- **Small Genitalia**
- **Hypoglycaemia**

Endocrine Assessment of a Patient with Short Stature

An endocrine investigation should be conducted to assess the GH-IGF-I axis.

Diagnosis of SPIGFD requires **severe short stature, low serum IGF-I and normal or increased growth hormone secretion** as well as **lack of other pathology** in the general work-up of short stature.



Severe Primary IGF-I Deficiency (SPIGFD) Definition

- Height ≤-3 SDS
- Basal IGF-I ≤-3 SDS (FDA) or IGF-I <2.5th percentile ~-2 SDS (EMA)
- Normal or elevated growth hormone concentration
- Exclude secondary and acquired IGFD

An early and correct diagnosis is essential to allow children to achieve their full growth potential with appropriate treatment

ALS, acid labile subunit; GH, growth hormone; GHBP, growth hormone binding protein; IGF-I, insulin-like growth factor-I; IGFBP-3, IGF-I binding protein-3; SDS, standard deviation score

Developed by International experts: Prof. Philippe Backeljauw (Cincinnati Children's Hospital Medical Center, USA) and Prof. Peter Bang (Linköping University, Sweden).

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