Severe primary insulin-growth factor-1 (IGF1) deficiency (SPIGF1D) is a rare cause of growth delay. Diagnostic criteria include age- and sex-dependent low basal IGF1 levels (<2.5th percentile), height ≤ -3SDS, absence of growth hormone (GH) deficiency and of any secondary causes of growth failure.

- Report the growth pattern and pubertal status
- Identify the molecular causes of SPIGF1D
- Describe the growth response after 1-year of recombinant human IGF1 (rhIGF1) treatment in a young patient with ACAN mutation

Thirty patients (M/F:17/13, n=30) with SPIGF1D (historical study cohort) out of 2546 patients referred for growth failure to Paediatric Endocrinology Department of Necker Children’s University Hospital, in Paris between 2004-2009 (Teissier et al, EJE, 2014).

Twenty patients with SPIGF1D (new cohort, n=20) among patients referred with growth retardation between 2016-2020.

- Molecular studies are ongoing, based on a candidate gene approach or next-generation sequencing gene panel
- Genetic analysis reflect the heterogeneous spectrum of the disease
- rhIGF1 is indicated if criteria for treatment in the approved indication are fulfilled and should not be delayed
- Long-term follow-up and genetic investigations are necessary for providing more insights in the SPIGF1D management

This work was in part supported by a grant from IPSEN Pharma