

Disease

Growth hormone insensitivity with immune dysregulation 1, autosomal recessive^{1, 2}

Gene

STAT5B

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant and recombinant human IGF1
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	STAT5 tyrosine phosphorylation, IGFBP-3 and IGF1 ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=stat5b&viewType=Table

References

¹ <https://pubmed.ncbi.nlm.nih.gov/36265659/>

² <https://pubmed.ncbi.nlm.nih.gov/26703237/>