

Disease

Ghosal hematodiaphyseal dysplasia^{1, 2, 3}

Gene

TBXAS1

Inheritance	AR
Current Treatments	NSAIDs, corticosteroids, tocilizumab
Evidence for Treatment	Expert opinion
Non-Molecular Testing Availability	Yes
Non-Molecular Test	IL-6 level after 3 weeks of age ³
Genes Included in Disease Frequency	TBXAS1
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=tbxas1&viewType=Card

References

¹ <https://omim.org/entry/231095>

² <https://pmc.ncbi.nlm.nih.gov/articles/PMC10082374/>

³ <https://pubmed.ncbi.nlm.nih.gov/41162285/>