

Disease

Severe congenital neutropenia 4^{1, 2}

Gene

G6PC3

Inheritance	AR
Current Treatments	granulocyte colony-stimulating factor (G-CSF), Bone marrow transplant (hematopoietic stem cell transplantation (HSCT))
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	complete blood count, bone marrow aspiration and biopsy ³
Disease Frequency (per 100,000)	0.04 ⁴
Genes Included in Disease Frequency	G6PC3
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Congenital+Neutropenia&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/30028820/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/31157858>
- ³ <https://omim.org/entry/612541>
- ⁴ <https://www.ncbi.nlm.nih.gov/books/NBK285321/>