

Disease

C1QB associated C1q deficiency^{1, 2, 3}

Gene

C1QB

Inheritance	AR
Current Treatments	Plasma infusion or exchange, Bone marrow transplantation Hematopoietic Stem Cell Transplantation (HSCT)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	classical pathway and alternative pathway of the complement system functional activity tests ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=c1q&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/29113537>

² <https://omim.org/entry/613652>

³ <https://pubmed.ncbi.nlm.nih.gov/33000368/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/27717414/>