

Disease

Specific granule deficiency 1^{1, 2}

Gene

CEBPE

Inheritance	AR and AD
Current Treatments	long term antimicrobial prophylaxis with antibiotics
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	complete blood count, peripheral smear, bone marrow aspiration and biopsy ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=cebpe&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/245480>

² <https://pubmed.ncbi.nlm.nih.gov/35726044/>