

Disease

CHARGE syndrome^{1, 2}

Gene

CHD7

Inheritance	AD
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile ¹
Disease Frequency (per 100,000)	1.72 ⁵
Genes Included in Disease Frequency	JAK3, IL2RG, LCK, NHEJ1, FOXN1, PNP, PRKDC, ORAI1, IL7R, PTPRC, ADA, RAG1, RAG2, DCLRE1C, CD3D, CD3E, CD247, AK2, CORO1A, CHD7, CIITA, RFXANK, RFX5, RFXAP, DOCK8, DOCK2, LIG4, MTHFD1, NBN, SLC46A1, PGM3, RMRP, STIM1, TTC7A, ZAP70, RAC2, PAX1, LAT, NUDCD3
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=chd7&term=&cntry=&state=&city=&dist=

References

- ¹ <https://pubmed.ncbi.nlm.nih.gov/29159871/>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/18505430>
- ³ <https://pubmed.ncbi.nlm.nih.gov/29168326/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/28160409/>
- ⁵ www.mdpi.com/2409-515X/3/2/15/pdf