

Disease

Otofaciocervical syndrome 2^{1, 2}

Gene

PAX1

Inheritance	AR
Current Treatments	thymus transplantation
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	T and B Lymphocyte and Natural Killer Cell Profile ²
Disease Frequency (per 100,000)	1.72 ³
Genes Included in Disease Frequency	JAK3, IL2RG, LCK, NHEJ1, FOXP1, 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=pax1&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/615560>
- ² <https://pubmed.ncbi.nlm.nih.gov/32111619/>
- ³ www.mdpi.com/2409-515X/3/2/15/pdf