Rx-Genes
Treatment Guidance at the Point of Care

**Disease**

# Gastrointestinal defects and immunodeficiency syndrome

**1, 2**

**Gene**

## TTC7A

| | |
|---|---|
| **Inheritance** | AR |
| **Current Treatments** | leflunomide, immunoglobulin replacement, hematopoietic stem cell transplantation (HSCT) - bone marrow transplant |
| **Evidence for Treatment** | expert opinion |
| **Non-Molecular Testing Availability** | yes |
| **Non-Molecular Test** | T and B Lymphocyte and Natural Killer Cell Profile, immunoglobulin levels[1] |
| **Disease Frequency (per 100,000)** | 1.72[3] |
| **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=ttc7a&term=&cntry=&state=&city=&dist= |

**References**


1 https://pubmed.ncbi.nlm.nih.gov/29921470/

2 https://www.ncbi.nlm.nih.gov/pubmed/31743734

3 www.mdpi.com/2409-515X/3/2/15/pdf