

## Disease

# Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia<sup>1, 2, 3</sup>

## Gene

### AIRE

|                                                 |                                                                                                                                                                                                                                               |
|-------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR and AD                                                                                                                                                                                                                                     |
| <b>Current Treatments</b>                       | prednisone, prednisolone, azathioprine, 6-mercaptopurine, mycophenolate mofetil, rituximab, oral swish/swallow suspension of amphotericin B, calcium, recombinant PTH, cyclosporine ophthalmic solution, mTOR inhibitor                       |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                                            |
| <b>Disease Frequency (per 100,000)</b>          | 0.805 <sup>4, 5</sup>                                                                                                                                                                                                                         |
| <b>Genes Included in Disease Frequency</b>      | AIRE                                                                                                                                                                                                                                          |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=AIRE+Deficiency&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=AIRE+Deficiency&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

## References

- <sup>1</sup> <https://omim.org/entry/240300>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/32557834/>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/30565240/>
- <sup>4</sup> <https://pubmed.ncbi.nlm.nih.gov/34217342/>
- <sup>5</sup> <https://pubmed.ncbi.nlm.nih.gov/34122451>