

Disease

Adenine phosphoribosyltransferase deficiency¹

Gene

APRT

Inheritance	AR
Current Treatments	Allopurinol or Febuxostat, Low purine diet and ample fluid intake
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	adenine phosphoribosyltransferase enzyme activity ²
Disease Frequency (per 100,000)	1.5 ³
Genes Included in Disease Frequency	APRT
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=aprt&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK100238/#aprt-def.Summary>

² <https://www.ncbi.nlm.nih.gov/books/NBK100238/>

³ <https://www.ncbi.nlm.nih.gov/m/pubmed/27994857/>