

Disease

Pyruvate kinase deficiency^{1, 2, 3}

Gene

PKLR

Inheritance	AR
Current Treatments	Mitapivat. red cell transfusion
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	complete blood count and red cell pyruvate kinase activity ⁵
Disease Frequency (per 100,000)	3 ⁶
Genes Included in Disease Frequency	PKLR
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Pyruvate+Kinase+Deficiency&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31483964>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/30681718>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32043619/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/32702739/>
- ⁵ <https://pubmed.ncbi.nlm.nih.gov/30681718/>
- ⁶ <https://www.ncbi.nlm.nih.gov/pubmed/30358897>