

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>