Monitor your patients with PK deficiency from the time of diagnosis

This assessment schedule was created in conjunction with leading physicians* and represents core health evaluations that allow healthcare providers to track patient disease progression over time. The patient's physician will determine the actual frequency of necessary assessments based on individualized need for medical care and routine follow-up.

RECOMMENDED SCHEDULE OF ROUTINE ASSESSMENTS						
	Baseline	**Every visit**	**Q1 months**	**Q3 months**	**Q6 months**	**Q12 months**
Medical history	•					
Genotype[1]	•†					
Physical exam	•					•
Height and weight	•	•				
Laboratory Tests[1]						
Complete blood count	•	•‡				•‡
Reticulocyte count	•	•‡				•‡
Bilirubin[§]	•	•‡				•‡
MONITORING FOR SYMPTOMS AND COMPLICATIONS						
Iron Overload Screening[1,2]						
Ferritin						
Patients on chelation therapy	•		•‖	•‖		
Regularly transfused patients (≥6 transfusions within the prior 12 months)	•			•¶	•¶	
Non- and minimally transfused patients (<6 transfusions within the prior 12 months)	•					•
Liver +/- Cardiac MRI[#]						
Regularly transfused patients	•					•
Non- and minimally transfused patients	•	Scan when ferritin >500 ng/mL; frequency of subsequent scans based on initial findings				
Osteopenia and Osteoporosis[2]						
25-hydroxyvitamin D**	•					•
Bone density test (DXA scan)[‡‡]	•	Frequency of subsequent scans based on initial findings				
Viral Screenings[2††]						
HIV	•[§§]					•
Hepatitis B, C	•[§§]					•

*Members of the Agios Steering Committee were compensated for their time.

†To help direct patient monitoring.

‡Perform at every visit or at least annually. Frequency depends on acute stressors and transfusion needs.

§Or comprehensive metabolic panel, as patient health warrants.

‖Perform every 1 to 3 months.

¶Perform every 3 to 6 months.

#First scan when patient can tolerate an unsedated study. Patients with ferritin >500 ng/mL who receive 10+ transfusions a year may require an MRI even if sedation is needed.

**If low, replete and retest in 8 weeks.[2]

††Baseline scan in early adulthood (10 to 19 years).

‡‡For patients who have received a transfusion within the past 12 months.

§§A baseline assessment for patients new to your practice is recommended.

PK=pyruvate kinase; DXA=dual-energy x-ray absorptiometry; HIV=human immunodeficiency virus; MRI=magnetic resonance imaging.

CONDITIONAL TESTS BASED ON FINDINGS

Patient with evidence of iron overload[2]	Vitamin D deficiency	Elevated bilirubin[1,2]	Aplastic crises[1]
Test for endocrinopathies • Thyroid-stimulating hormone • Sex hormones • Fructosamine*	Consider frequency of DXA	Monitor for gallstones and other signs of gallbladder disease; consider abdominal ultrasound	To evaluate the cause of reticulocytopenia, screen for parvovirus B19 serology (including IgM) or PCR

ADDITIONAL POTENTIAL COMPLICATIONS[1,2]

Complications	Symptom	Imaging
Gallstones and other gallbladder complications	• New or worsening abdominal symptoms • Worsening jaundice • Other related symptoms	Abdominal ultrasound
Pulmonary hypertension	Poor cardiac function or other signs	ECHO
Extramedullary hematopoiesis	Back pain, hepatomegaly, or other signs	Image for evidence of paravertebral or hepatic extramedullary hematopoiesis

QUALITY OF LIFE ASSESSMENTS[2,3]

	Baseline	Every visit	Q1 months	Q3 months	Q6 months	Q12 months
Cognition (fogginess)	•					•
Depression/anxiety	•					•
Fatigue	•					•

*Fructosamine, rather than HbA1c, should be used to screen for diabetes mellitus.[2]

IgM=immunoglobulin M; PCR=polymerase chain reaction; ECHO=echocardiogram; HbA1c=hemoglobin A1c.

For more information and additional resources, visit KnowPKDeficiency.com

References: 1. Grace RF, Mark Layton D, Barcellini W. How we manage patients with pyruvate kinase deficiency. *Br J Haematol.* 2019;184(5):721-734. **2.** Al-Samkari H, van Beers EJ, Kuo KHM, et al. The variable manifestations of disease in pyruvate kinase deficiency and their management [published online ahead of print March 12, 2020]. *Haematologica.* 2020;2019:240846. doi:10.3324/haematol.2019.240846. **3.** Grace RF, Cohen J, Egan S, et al. The burden of disease in pyruvate kinase deficiency: patients' perception of the impact on health-related quality of life. *Eur J Haematol.* 2018;101(6):758-765.

Monitoring guidelines include feedback from the Agios Steering Commitee, whose members were compensated for their time.

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