

Monitoring your PK deficiency over time



When you have pyruvate kinase (PK) deficiency, knowing which tests and follow-up assessments are needed can help you and your doctor identify health trends early on. The schedule below was developed with input from the **International Guidelines for the Diagnosis and Management of Pyruvate Kinase Deficiency** and leading PK deficiency specialists* to show recommended tests and their frequency for people age 18 and older. If you only see your hematologist once a year, another member of your healthcare team may perform these tests. In the tables below, choose the first description that applies to you. The information in that column can be used in conversations with your doctor about your monitoring schedule.

*The Monitoring Guidelines were written by a Steering Committee of leading physicians convened by Agios, and the physicians were compensated by Agios for their time.

	Regularly transfused within the last year	Not or minimally transfused within the last year
Blood tests		
Iron levels (ferritin)	At least every 1 to 3 months	Yearly
Bilirubin	Every visit	Every visit
Complete blood count (CBC)		
Reticulocytes (new red blood cells)		
Vitamin D levels	Yearly	Yearly
Diabetes screening	Fasting glucose testing, oral glucose tolerance testing, or fructosamine measurements are recommended, as hemoglobin A1c measurements are unreliable.	
Kidney function • Creatine, phosphorous, magnesium, and albuminuria	The frequency of monitoring depends on your risk factors. Make sure to talk to your doctor about what they may be based on your clinical situation.	

Urine tests		
Kidney function • Urine protein: creatinine ratio	The frequency of monitoring depends on your risk factors. Make sure to talk to your doctor about what they may be based on your clinical situation.	

	Regularly transfused within the last year	Not or minimally transfused within the last year
Heart, liver, and bone scans		
Heart and liver MRI If you are on chelation therapy, liver MRI should be done every year.	Liver MRI: Yearly Heart MRI: • Every year if concentration of iron on the liver is high or chelation therapy is not working • Every 2 years if concentration of iron is within expected range with chelation therapy	Liver MRI: Every 5 years Heart MRI: Scans can be performed less often than every 12 months, depending on the concentration of iron levels in your liver.
<i>Children, especially young children, may not be able to stay still for an MRI without sedation. Talk to your doctor about the risks that sedation can pose versus the risk of not being able to conduct these tests.</i>		
Echocardiograph (echo) with tricuspid regurgitation jet (TRJ)	Every 1 to 5 years, depending on risk factors. Make sure to talk to your doctor about what they may be based on your clinical situation.	
Bone density test (DEXA scan)	The frequency of scans depends on your initial results and risk factors. Make sure to talk to your doctor about what they may be based on your clinical situation.	

Lab tests: Normal ranges for adults [†]			
Ferritin	Females (18-39 years): 10 to 120 ng/mL	Females (40+ years): 12 to 263 ng/mL	Males (18+ years): 20 to 250 ng/mL
Total bilirubin	Less than 1.2 mg/dL		
Reticulocytes	0.5% to 2.5%		
Vitamin D levels	Some experts recommend levels of 20 to 40 ng/mL, while others recommend 30 to 50 ng/mL		
DEXA scan	T-score of -1 or higher		

[†]Different labs may have different definitions of “typical” or may use different measurements. Talk to your doctor about the meaning of your results.

Key Terms

CBC: A complete blood count assesses many elements to get a more complete picture of the health of your blood. A CBC typically includes, among other measures, hemoglobin, hematocrit, and white blood cell levels

Chelation therapy: Medicine that binds to iron in your bloodstream so it can be eliminated via the digestive or urinary tract

HIV: A blood-borne virus that can cause acquired immunodeficiency syndrome (AIDS)

Hepatitis: Blood-borne viruses that can cause the liver to become inflamed

Transfusion: The process of putting blood into the bloodstream by intravenous (IV, meaning through the veins) infusion

If you are the caregiver of someone under the age of 18, suggested monitoring may vary. Talk to your loved one’s doctor to find out more.

Follow-up tests

Based on what your blood tests show, you may need further assessments.

Results	Background
High iron levels may require additional blood tests to assess the function of these organs: <ul style="list-style-type: none">• Thyroid• Pituitary• Pancreas	Iron overload can damage the endocrine system (the body)
You may need an abdominal ultrasound if you have: <ul style="list-style-type: none">• High bilirubin levels• Worsening jaundice or yellow eyes• New or worsening abdominal symptoms	High levels of bilirubin can cause gallstones or gallbladder disease Worsening jaundice or abdominal pain, nausea, and vomiting can indicate that you have gallstones or some form of gallbladder disease
You may need additional blood tests to discover the cause of a low reticulocyte count (also referred to as reticulocytopenia [ruh-TIK-you-lo-sigh-toe-PEA-knee-yuh])	A parvovirus infection can cause the body to stop producing new red blood cells (an aplastic crisis). The virus usually causes fever and a rash on the face A parvovirus infection can only happen once and is more likely to occur in childhood
Your doctor may ask you to make diet or lifestyle changes if you have low vitamin D levels and then retest you to see if your levels have increased	People with PK deficiency are at greater risk of osteopenia and osteoporosis, so it's important to take steps to ensure healthy bones
Your doctor will perform a visual exam if you have back pain or unexplained, painful swelling, and may need to perform other tests	Because PK deficiency can lower the number of red blood cells you have, your body may try to make red blood cells in places where it shouldn't, like around the spine. This is called extramedullary hematopoiesis (extra-MED-you-lerry-HEE-mah-toe-PO-ee-sis)

Key Terms

Hormones: Chemicals that travel throughout your body via the bloodstream. They help regulate growth and development, metabolism (changing food into energy), mood, sexual function, and reproduction. Hormones are part of the endocrine system

Jaundice: Yellowing of the skin caused by high levels of bilirubin in the body

Scleral icterus: Yellowing of the whites of the eyes caused by high levels of bilirubin in the body

Managing PK deficiency can impact your quality of life. Take time out to talk to someone on your healthcare team every year about how you're feeling. If you have feelings of worry or sadness that don't go away, be sure to notify your healthcare team.



If you would like an Agios Clinical Nurse Educator to walk you through this monitoring guide and help you prepare for your next doctor's appointment, enroll in myAgios Patient Support Services: <https://pkdeficiency.myagios.com/>