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 leading PK deficiency specialists and shows 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.

	Chelation therapy	6+ transfusions within the last year and not on chelation therapy	5 or fewer transfusions within the last year and not on chelation therapy
Blood tests			
Iron levels (ferritin)	Every visit	At least every 3 months	Yearly
Bilirubin	Every visit	Every visit	Every visit
Complete blood count (CBC)			
Reticulocytes (new red blood cells)			
Vitamin D levels	Yearly	Yearly	Yearly
HIV	Yearly for people who have had a transfusion	Yearly for people who have had a transfusion	Yearly for people who have had a transfusion
Hepatitis B and C			

	6+ transfusions within the last year	5 or fewer transfusions within the last year
Heart, liver, and bone scans		
Heart and liver MRI	Yearly	These scans should be done yearly if your iron levels are more than 500 ng/mL You may need additional assessments depending on the initial results
	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.	
Bone density test (DXA scan)	Your doctor will schedule an initial scan at about the age of 18 You may need regular scans depending on what this first assessment reveals	

Lab tests: Normal ranges for adults*			
Ferritin	Females (ages 18-39): 10 to 120 ng/mL	Females (ages 40+): 12 to 263 ng/mL	Males (ages 18+): 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		
DXA 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 into the arm

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 for: • Thyroid-stimulating hormone • Sex hormones • Fructosamine	Iron overload can damage the endocrine system (the body system that makes hormones). Hormone levels that are too high or low can cause other health issues In PK deficiency, doctors check for signs of diabetes by looking at fructosamine levels
You may need an abdominal ultrasound if you have: • 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	PK deficiency doesn't cause low vitamin D levels, but low vitamin D levels may impact bone health. People with PK deficiency are at a greater risk for osteopenia and osteoporosis, so it's important to takes 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 lowers the number of red blood cells you have, your body may try to make red blood cells in places where it shouldn't, like in the spine. This is called extramedullary hematopoiesis (extra-MED-you-lerry-HEE-mah-toe-PO-ee-sis)
If you show signs of heart problems, your doctor may perform an echocardiogram	Iron overload can cause pulmonary hypertension. Pulmonary hypertension is a type of high blood pressure that affects the arteries that lead to the heart

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 coping. If you have feelings of worry or sadness that don't go away, be sure to notify your healthcare team.

Visit the **Know PK Deficiency** channel on YouTube to hear people with PK deficiency discuss working with their healthcare team.