

Monitoring PK Deficiency

Regular evaluations can help make sure you're getting the right care. Many assessments to monitor PK deficiency are done on a yearly basis, but some need to be done more often based on transfusion frequency, the need for chelation therapy, and discoveries from previous tests.

Know the tests for monitoring your complications



Gallstones

Monitored by ultrasound if there is new or worsening abdominal pain, worsening jaundice, or other related symptoms.



Iron damage to the heart or liver

Monitored by a yearly T2* MRI scan. Patients who receive regular transfusions, or who need chelation therapy, may need to be assessed more frequently.



Osteopenia and osteoporosis

A DXA should be done in early adulthood. Results of the scan determine how often the test should be repeated.



Pulmonary hypertension

An echocardiogram should be done by age 30. Doctors determine if the test needs to be repeated based on what the picture shows.



Extramedullary hematopoiesis

A visual exam is performed regularly, with further testing if there is unexplained swelling, back pain, or symptoms that indicate signs of nerve damage, such as numbness, tingling, burning, or shooting pain.



Blood tests, at least annually, for:

- Degree of anemia (hemoglobin levels)
- Iron overload (ferritin levels)
- Vitamin D levels (to help assess bone health)
- Viruses, such as HIV, and hepatitis A, B, and C (for people who receive transfusions)

If there is evidence of iron overload, your doctor may do additional blood tests to check hormone levels. Iron overload can affect sex hormones or cause changes that contribute to thyroid problems or diabetes.

DID YOU KNOW?

The need and timing of tests varies for everyone. Talk with your doctor about each test to determine a monitoring plan.