Common Tests
- Electrocardiogram (ECG/EKG)
- Echocardiogram
- Holter Monitor (an extended wear ECG/EKG)
- Hs-CRP to detect C-reactive protein, a marker of inflammation
- Cardiac biomarkers (BNP or NT-proBNP)
- Comprehensive metabolic panel to evaluate organ function including electrolytes and liver function markers

More Extensive Tests
- Stress tests
  - Metabolic
  - Exercise

Imaging that may be recommended by the doctor
- MR Imaging
- Tilt table test
- PET Imaging

Invasive Tests
- Trans-esophageal echocardiogram
- Electrophysiology study
- Cardiac catheterization
- Heart biopsy

Other Blood Tests
- Lipid profile to examine the amount and types of lipids in the blood
- Complete Blood Count to detect infection and assess the neutrophils
- Blood culture to detect infections in the heart
- Troponin to detect cardiac damage
- CK-MB enzyme to detect cardiac damage

It is ideal for the person with Barth syndrome to be evaluated by a cardiologist with a special interest in heart failure and/or cardiomyopathy at least on a yearly basis.

The cardiologist may also refer the person with Barth syndrome for an evaluation by an electrophysiologist who specializes in heart rhythm abnormalities.

https://www.barthsyndrome.org