



**Diagnosis: Barth Syndrome ICD# E78.71**

**This patient has been diagnosed with Barth syndrome** and should be triaged as soon as possible upon arrival in the emergency department, even if he does not appear to be ill. Decompensation and death may occur rapidly. Early and aggressive intervention may reduce complications and stop further deterioration or death.

**Barth syndrome** is a rare genetic, life-threatening disorder caused by a mutation in the *tafazzin* gene (TAZ, also called G4.5), resulting in an inborn error of phospholipid metabolism.

**Cardinal Characteristics of Barth Syndrome**

- *Cardiomyopathy*
- *Neutropenia*
- *Low Muscle Mass and Muscle Weakness*
- *Exercise Intolerance*
- *Growth Delay*

**Additional Clinical Problems of Barth Syndrome**

- *Heart failure*
- *Sepsis*
- *Arrhythmia*
- *Thrombosis*
- *Hypoglycemia*

Date:	
Patient Name:	First Last MI
Date of Birth:	
Street Address:	
City, State, Postal Code:	
Phone:	
Corresponding Documents	Medications, Allergies, Healthcare Providers, My Health History

## Common Presentation and Acute Complications

### Cardiac Dysfunction

- Cardiomyopathy and heart failure
- Life-threatening arrhythmias, even when heart function is normal (PVCs and PACs may not be benign)
- Cardiac decompensation can be triggered by intercurrent illness and stress. In the setting of clinical changes, a recent normal echocardiogram should not be considered reassuring.
- If patient has received a heart transplant, seek advice from transplant team.

### Infection/ Neutropenia and Sepsis

- Neutropenia with an ANC approaching zero is common. Consequently, a normal ANC is not necessarily reassuring and may be a sign of a life-threatening infection (relative neutrophilia).
- Normal body temperature often runs below 98.6F/37C. Therefore, mild fever may signify infection. Infection should be considered if temperature is > 100.4F/38C
- Rectal temperatures are contraindicated due to high risk of neutropenia.

### Nutritional and Metabolic Issues

- Limited tolerance for fasting with potential for rapid metabolic decompensation
- Increased risk of hypoglycemia
- Increased risk of electrolyte imbalances, particularly when experiencing diarrhea and/or vomiting

## Notes

## Recommended Tests for Urgent Evaluation

- D-stick/blood glucose
- Complete Blood Count with differential
- Comprehensive Metabolic Panel
- Venous Blood Gas
- C-reactive protein (CRP)
- B-type natriuretic peptide (BNP)/ N-terminal pro b-type natriuretic peptide (NT-proBNP)
- Electrocardiogram (EKG)
- Echocardiogram
- Chest X-ray

## Management (Even if the patient does not appear unwell)

- Place onto continuous telemetry and frequent vital signs
- Obtain labs/place stat peripheral IV if oral feeding is not tolerated/inadequate or there are signs of dehydration,
  - IV fluid boluses should be done with care, and in the context of any existing cardiac dysfunction
- Treat hypoglycemia
  - Patients are prone to hypoglycemia particularly after a long fast and episodic vomiting and diarrhea. Correct hypoglycemia with age and weight appropriate protocol in administering IV bolus of glucose solution and glucose drip
- Consider admission.
  - <Insert first name>'s physicians contacts are provided below. Please contact for further care management instructions or before discharging him from the ED.
- Consider sepsis evaluation

## Notes

### **BARTH SYNDROME FOUNDATION**

## Admission

Most patients who present in the ED of hospital will require admission. Use caution when discharging the patient to go home. Please consult with these providers who are most familiar with the patient's condition as needed.

## Notes

## Additional Resources

**Physician to Physician Consultation:** The Barth Syndrome Foundation maintains a list of multidisciplinary sub-specialists with expertise in the disease who have agreed to speak DIRECTLY with peer physicians during times of crisis. To obtain a list of these physicians contact The Barth Syndrome Foundation.

**Barth Syndrome Foundation Website:** Additional information about Barth syndrome is available for clinicians on the Barth Syndrome Foundation website [www.barthsyndrome.org](http://www.barthsyndrome.org)

### **BARTH SYNDROME FOUNDATION**

2005 Palmer Avenue #1033, Larchmont, New York 10538, Phone (914) 303-6323, Fax (518) 213-4061 [www.barthsyndrome.org](http://www.barthsyndrome.org)

## My Healthcare Providers

<b>My Healthcare Providers</b>		
Area of Specialty/ Subspecialty		
Name		
Address		
Telephone Number		
Has reviewed this document	Yes	No
Area of Specialty/ Subspecialty		
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Has reviewed this document	Yes	No

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