

Hospital Management Plan

Date:
Patient Name:
DOB:
NHS No:
Address:

Diagnosis: Barth Syndrome

Problems:

Medications:

Barth Syndrome

- **Please read carefully. Meticulous treatment is important as there is a high risk of serious complications.**
- **If the instructions do not make sense or a problem is not addressed you must discuss your concerns with the consultant on call.**

Background

Barth Syndrome is a rare inherited inborn error of metabolism characterised by cardiomyopathy, neutropenia, delayed motor development, chronic fatigue, exercise intolerance and growth delay. Patients are also prone to hypoglycaemia particularly after a long fast and episodic diarrhoea. It is X-linked and mostly affects males. Whilst Barth syndrome has no definitive cure, early and aggressive intervention may reduce complications and stop further deterioration or death.

Major clinical problems associated with Barth Syndrome include

- Cardiac dysfunction
- Arrhythmias
- Life threatening bacterial infections due to neutropenia and neutrophil dysfunction

Admission

Most patients who present to hospital will require admission. Only allow home if you and the family are entirely happy and you have discussed the problems with the consultant on call. Please discuss with the National Barth Syndrome Service if there are any queries. The patient/family must have a clear management plan and know when to return if they are discharged.

Initial plan and management in hospital

Make a careful clinical assessment including blood pressure and temperature, even if the patient does not appear unwell.

Neutropenia

- If the patient has a temperature of >38C consider bacterial infection
- Do a full blood count, U&Es, pH and gases, blood glucose, blood culture and CRP
- If no obvious focus, consider a septic screen including viral PCR throat swab and Chest Xray
- Please note if the patient is usually neutropenic, a normal neutrophil count may not necessarily be reassuring as this may be a limited response to a serious infection
- Rectal temperatures are contraindicated due to high risk of neutropenia.
- Start IV antibiotics - decisions should be based primarily on the clinical status.
- Fluids
 - Can the patient tolerate oral fluids?
 - If the patient is relatively well - may be treated orally but assess very carefully.
 - If the patient is obviously unwell –must be treated with intravenous antibiotics
 - If there is any doubt at all, put up an intravenous line.

Cardiac

Admissions of Barth patients with cardiac illness need to be discussed with the cardiology team of the Hospital and the Barth team at Bristol. If patient has received a heart transplant, seek advice from the patient's transplant cardiologist.

Cardiac decompensation can be triggered by intercurrent illness and stress. A recent normal echo should not be considered reassuring in the setting of clinical changes.

In an emergency, ABC guidelines (resuscitation protocols) should be followed.

Investigations:

Consider need for ECG and ECHO and cardiology consultant review.

Progress and Going Home

If not improving or if deteriorating, please escalate care and discuss with consultant. Please consult the Barth syndrome service as needed.

Only allow home if you, the patient and the family are entirely happy and you have discussed the problems with the consultant on call. There must be a clear management plan and clear advice about when to return.

Barth Syndrome Team email :

Dr G Pierre
Dr E Chronopoulou
Dr Guido Pielas
Dr Adam Gassas
Hayley Smith

Barth/Metabolic Consultant
Barth/Metabolic Consultant
Cardiac Consultant (Barth)
Haematology Consultant
Barth Specialist Nurse



barthsyndromeservice@uhbristol.nhs.uk

Hayley.smith@uhbristol.nhs.uk
07795 507294

Barth Secretary

0117 3428102