



Neutropenia in Barth Syndrome (Chronic, Cyclic or Intermittent)^{i ii}

What problems can Neutropenia cause?

Neutrophils are the main white blood cell for fighting or preventing bacterial or fungal infections. They may be referred to as polymorphonuclear cells (polys or PMNs), white cells with segmented nuclei (segs), or neutrophils in the complete blood cell count (CBC) report. Immature neutrophils are referred to as bands. When someone is neutropenic (an abnormally low level of neutrophils in the blood), the risk of infection increases. The absolute neutrophil count (ANC) is a measure of the total number of neutrophils present in the blood. When the ANC is less than 1,000, the risk of infection increases. Most infections occur in the ears, skin or throat and to a lesser extent, the chest. These infections can be very serious and may require antibiotics to clear infections. When someone with Barth syndrome is neutropenic his defenses are weakened, he is likely to become seriously ill more quickly than someone with a normal neutrophil count.

Tips:

- No rectal temperatures as any break in the skin can lead to an infection.
- If the individual has a temperature > 100.4° F (38° C) or has infectious symptoms, the primary physician or hematologist should be notified. The individual may need to be seen.
- If the individual has a temperature of 100.4° F (38° C) – 100.5° F (38.05° C) > 8 hours or a temperature > 101.5° F (38.61° C), an immediate examination by the physician is warranted.

Some or all of the following studies may be ordered:

- CBC with differential and ANC
- Urinalysis
- Blood, urine, and other appropriate cultures
- C-Reactive Protein
- Echocardiogram if warranted
- The physician may suggest antibiotics (and G-CSF if the ANC is low) for common infections such as otitis media, stomatitis.
- For cuts and scrapes, apply topical antibiotic cream to site and watch for signs of infection such as redness, warm to touch, swelling, and pain. Without neutrophils, not all of these signs may be present and the individual will not be able to fight infection efficiently.
- In addition to standard immunizations, the physician may suggest influenza and pneumococcal vaccines.
- If the individual appears listless without any obvious reason, you should get in touch with your physician immediately.
- Ask the physician about 'Direct Access' for neutropenic children.
 - *This means they can be admitted straight to the ward without going via your GP.*

| Fahrenheit-Celsius conversion table | |
|-------------------------------------|------|
| °F | °C |
| 105 | 40.5 |
| 104 | 40.0 |
| 103 | 39.4 |
| 102 | 38.9 |
| 101 | 38.3 |
| 100 | 37.7 |
| 99 | 37.2 |
| 98 | 36.6 |
| 97 | 36.1 |
| 96 | 35.5 |

Signs and Symptoms of Neutropenia:



- Fever
- Ulcers (Anal or Mouth)
- Rash
- Frequent Infections
- ANC less than 1,000

Daily Routine:

Because most who have Barth syndrome are neutropenic at least sometimes, take care to protect from unnecessary germs and be diligent about personal hygiene and daily routines concerning germs:

- Avoid people who are known to be ill and contagious
- Wash hands frequently with soap and water or with an antimicrobial lotion that can easily be kept on hand at all times
- Do not allow sharing of drinks or food
- Wash fruits and vegetables thoroughly
- Cook meat appropriately
- Brush teeth regularly and thoroughly and have teeth cleaned 2 – 4 times per year
- Replace toothbrush every 6 – 8 weeks
- Keep fingernails short and clean
- Use antibacterial ointment on cuts and scrapes (once they have been cleaned completely) and watch for signs of infection such as redness, swelling and pain.
- Make sure that your friends, family and babysitters understand the special risks of bacterial infection and are careful around your Barth child.

- **Absolute Neutrophil Count (ANC)** This number represents the amount of neutrophils, which are available for defending the body at the time of the blood test. There are two ways to determine this count (percentage equation or absolute numbers). This depends on how the laboratory reports the results. A normal ANC for a non-neutropenic person is generally within the range of 1800-7000.
 ↳ Refer to the [Determining your ANC \(Absolute Neutrophil Count\) document](#)
- **Autosomal recessive:** A particular type of genetic inheritance. In an autosomal recessive condition such as Kostmann syndrome, the child will have the disease only if both parents pass the affected gene on to their child. The addition 'autosomal' refers to the fact that the inheritance is independent of the child's sex.
- **Congenital Neutropenia:** Severe chronic neutropenia beginning at or before birth. It is characterized by persistent, severely diminished neutrophil counts in the bone marrow and blood due to a maturational arrest of early cells in the bone marrow. Subtypes include congenital agranulocytosis (Kostmann syndrome), Shwachman-Diamond syndrome, glycogen storage disease type 1b, and myelokathexis.
- **Complete Blood Count (CBC):** A summary of the numbers of various types of cells present in the blood at the time of the blood draws, same as full blood count (FBC).
- **Cyclic Neutropenia:** Cyclic neutropenia is another inherited type of neutropenia. As the name indicates, in this disease neutrophil counts show a cyclic pattern, with a typical cycle length of 21 days. These cycles vary from patient to patient with some individuals being neutropenic during the whole cycle and others who have low neutrophil counts for only a few days and normal blood counts during the rest of the cycle.
- **Febrile Neutropenia:** Dangerously low level of neutrophils in the blood (usually less than 500 cells per cubic millimeter) accompanied by fever; a condition that indicates that the patient may have a potentially life-threatening infection.
- **Granulocyte:** A subtype of leukocytes, which can be further distinguished into neutrophils, eosinophils and basophils.
- **Granulocyte Colony-Stimulating Factor (G-CSF):** A naturally occurring hormone that stimulates the growth, production, and function of neutrophils. These supplements are administered by injection.
- **Idiopathic Neutropenia:** Severe chronic neutropenia of unknown cause; a broad term for unexplained neutropenia in both adults and children. The condition can be acquired. This is NOT a term that could be associated with Barth syndrome.
- **Neutropenia:** An abnormally low level of neutrophils in the blood.
- **Neutrophils:** A subgroup of granulocytes defending the body against bacteria. Neutrophils are also known as segs, polys or segmented neutrophils.
- **Platelets:** A subgroup of blood cells responsible for clotting, which are also called thrombocytes.
- **Prophylaxis:** Any procedure or treatment used to avoid undesired events; an example of these treatments would be, the use of immunizations, low-dose daily antibiotics, or ear tubes to prevent the development of infections
- **Recombinant Human Granulocyte Colony-Stimulating Factor:** The genetically engineered version of the natural growth factor, (G-CSF).
- **White Blood Cells:** A subgroup of blood cells consisting of monocytes, granulocytes (neutrophils, basophils, and eosinophils), and lymphocytes, which together build the immune system and defend the body against infection
- **White Blood Count (WBC):** The total number of leukocytes (white blood cells) in the blood at the time of the blood test.

ⁱ Severe Chronic International Neutropenia Registry

ⁱⁱ Guidelines for the Neutropenic Patient, Gerald Cox, MD