Condition Insight Report (CIR)

Thalassaemia

Version 1.1
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Created in collaboration with United Kingdom Thalassemia Society

Overview

What is the condition usually called/ any abbreviations used?

- Transfusion Dependent Thalassaemia
- Thalassaemia Major
- Cooley's Anaemia
- Mediterranean Anaemia
- Erythroblastic anaemia
- Microcytemia (beta)

Brief overview of the condition

Thalassaemia is an inherited blood disorder that lowers, alters or stops the production of haemoglobin. There are several types of thalassaemia depending on the severity of the condition. The most severe for is beta thalassaemia major, followed by beta thalassaemia intermediate & haemoglobin H disease. Thalassaemia Minor/trait/carrier is the mildest form of the condition and usually is asymptomatic.

Beta Thalassaemia Major is a progressive condition in which symptoms and treatment burden increase as patients grow older. Not only can people with thalassaemia develop tumours due to extra medullary hemopoiesis, they are also at risk of developing allo-antibodies and suffer from reactions from repeated blood transfusions which can cause a variety of life-style changes (affecting the ability to walk, shower, or live independently.)

Additionally, due to excess iron accumulated during regular blood transfusions over time, patients also develop other multi-organ damage and failure. As iron is not excreted or used to produce haemoglobin, it deposits in vital organs of the body leading to people with thalassaemia developing other secondary conditions which can impact on daily life.

What is the generally preferred term for someone with this condition?

Person living with thalassaemia

Presenting Symptoms / Co-occurring conditions

Generally, a person with thalassaemia can look healthy and their difficulties with daily living and mobility may not be apparent. It is therefore vital to gain an understanding of their condition by encouraging an open and frank discussion and looking beyond the appearance.

For instance, people with thalassaemia can develop:

1. **Liver and Gall Bladder Diseases**
2. **Endocrine and Metabolic Dysfunction**
3. **Cardiac Dysfunction**
4. **Pulmonary Care**
5. **Thrombosis/ Thrombotic Events**
6. **Spleen**
7. **Pain Syndrome**
8. **Dental Issues**
9. **Chelator Side Effects and Toxicity**
10. **Rheumatology**
11. **Psychological**

Other symptoms people are likely to experience are:

- Fatigue / exhaustion
- Breathing restrictions
- Palpitations
- Headaches
- Reduced concentration
- Insomnia

Due to iron overload in the pancreas, some patients often suffer from **diabetes mellitus** and may struggle to consume the right diet. Increasingly, the prevalence of pancreatic insufficiency is on the rise amongst the older patients, which can cause severe weight loss and malnutrition as they are unable to absorb nutrients. This in turn can exacerbate symptoms of their secondary conditions and decrease their overall health and quality of life.

Patients with thalassaemia are also prone to developing **bone disease** (osteopenia/ osteoporosis syndrome) due to bone marrow expansion and as a result, are more susceptible to falling and fracturing bones. This is not visible; however, it can result in patients becoming frail and anxious about managing their daily lives. Many patients may also require bone replacement surgery over their lifetimes and have difficulty with their mobility.

Iron overload is a silent killer. There are not many visible symptoms for people who don't specialise in haemoglobinopathies. The impact of iron deposition in every organ and its implications on their functioning can be fatal. Iron overload can also cause hormonal and fertility issues. Most men with thalassaemia have low testosterone levels and thus require treatment with testosterone injections every three months. Low testosterone can cause a myriad of problems such as decreased muscle mass/ strength, decreased body hair, swelling/tenderness of the breast tissue, increased fatigue, hot flashes, sleep disturbances, impotency and issues fathering children. Not only does this cause physical complications it can also result in psychological issues such as memory/ concentration loss, depression, lack of self-esteem, body issues and puts pressure on personal relationships. Patients can be very embarrassed to talk about these issues. In women, menstrual cycles are disrupted and irregular, affecting fertility. Patients may find this hard to express and openly discuss, and may not have come to terms with what this means for their future life choices.

Fluctuations

Due to the progressive nature of the condition and the treatment that individuals require, there can be significant variations between each day for many. They are also at a higher risk of recurrent infections due to their weakened immune system. Their functional ability and fluctuation in presentation can also be impacted by having blood transfusions – before and after a transfusion they can present very differently. This should be explored.

- **Treatment regimes** (such as blood transfusions, bisphosphonates, examinations/ consultations)

  On the lead up to blood transfusions what additional symptoms do they have? How long do these symptoms present in the same way post a transfusion? How often are transfusions?

- Fatigue

- **Recurrent infections and or inflammation**
  How often are these? How long do they generally last, or can they give an example of the last time they had one how long this took and whether this was similar to what they would normally experience? What or where are the infections/inflammation? How do they manage these?

Reliability

What specific areas should be covered to ensure a complete, reflective report?

SAFETY	TIMELY	ACCEPTABLE STANDARD	REPEATEDLY
Do they have any symptoms which could cause a safety consideration?	For any activities where restriction is reported how long does it take them to complete these activities? Has how long it takes them changed over time?	How have they adapted to completing tasks over time – is this different to what might be considered 'normal'?	Are they able to repeat a task as often as required? Is this the same every day?
When an individual has an infection or becomes anaemic, it can result in an exacerbation of their symptoms, causing everyday tasks to be become extremely complex and challenging.	The treatment burden of daily medications combined with symptoms of bone and neuropathic pain can cause the individual to become fatigued on a daily basis which can impact how long it takes an individual to complete a task.	Adapting to complete tasks is likely given the nature of the condition. They might not consider the way they complete a task is unusual if they have lived with this for some time. Please make sure to explore how they are completing tasks. People with thalassaemia have been conditioned to live and manage their daily pain and their "normal" day would be the day they can move and tolerate the pain, which for someone without the condition could mean that they are unable to move.	Patients can suffer from extreme fatigue, exhaustion, breathlessness, palpitations, bone pain (due to the bone marrow going into overdrive), headaches, lack of concentration, cognition disturbances, low mood, anxiety, depression and insomnia. This can all impact on their ability to complete a task in the same way as often as required. You need to explore what occurs most often for them, or the differences for them.

What input/ treatment may they receive?

- **Audiology**- iron chelation toxicity
- **Blood work-** can be as often as weekly for anticoagulation, iron chelation toxicity (neutropenia, kidney & liver failure), urine tests as well as full blood counts, cross matching for transfusion etc
- **Cardiac (ECGs, Echo Cardiograms, MRI T2*)**
- **Dermatology** (with regards to iron chelation and blood transfusion reactions, biopsies)
- **Endocrinology-** Diabetes control, hormones management, bone health
- **Gastroenterology**- Stomach, gall bladder, pancreas
- **Gynaecology**- Hormone replacement treatment and management
- **Immunology**- blood transfusion and iron chelation related allergies
- **Liver / Hepatology/ Pancreatic** (MRI, biopsies)
- **Nephrology**- iron overload and iron chelation toxicity can result in kidney failure and eventually the need for dialysis treatment.
- **Neurology**- nerve damage, neuropathy, paralysis due extra medullary haematopoiesis, diabetes, cannulation damages, seizures
- **Ophthalmology**- iron chelation toxicity and diabetes related complications
- **Physiotherapy** – sessions to help with muscle and bone pain
- **Surgical consultations** (for kidney and gall stones)

Medical Reports from Consultant Haematologist, Clinical Nurse Specialist or General Practitioner

Care plans/ carer diaries

What kind of medical evidence is likely to best convey the impact of this health condition or disability on an individual?

Physiotherapy reports

DEXA and MRI reports

REMEMBER: Absence of evidence does not mean absence of a restriction.

Sensitivities

What areas might they find difficult to mention or perhaps understate the impact of?

it is important to speak to an involved advocate/ family member/ carer to get a clearer picture.

Generally, a person with thalassaemia can look healthy and their difficulties with daily living and mobility may not be apparent. It is therefore vital to gain an understanding of their condition by encouraging an open and frank discussion and looking beyond the appearance.

Thalassaemia patients live with daily pain and as such may try to disguise how they feel as best as possible. As pain is subjective and symptoms are not easily identifiable, it is important to be mindful that every individual can have a different pain threshold and as such can often hide the agony or soreness they feel by smiling or adapting some type of cognitive behavioural therapy coping mechanisms they may have learnt over time.

Individuals with thalassaemia are often underweight and short in stature. Additionally, as puberty is often delayed, they can look very young in appearance. Consequently, they are often mistaken for and spoken to as children which can cause them distress and prevents them disclosing their need to rely on others when they are unwell. They have often spoken about the guilt they feel not being able to care or provide for themselves. Those who live alone may find it difficult to disclose that they could benefit from extra support

Watch a video **here**.
Thalassaemia & me

Customer Care

How is it best to ask about any sensitive topics and what are the common courtesies?

In general

- Introductions to all, of all.
- Ask someone what they would prefer to be called.
- An explanation as to what is being done and why it is being done will be helpful.
- Have patience- the assessment process can be intimidating and embarrassing for people with thalassaemia.
- Be mindful of not being patronising or condescending- treat adults like adults despite how young they may appear.
- A person with thalassaemia may find the word “sufferer” or “thalassaemic” inappropriate.
- Remember that most patients are forced to live with pain and are encouraged to try to avoid taking analgesics or narcotics as it is habit forming and not designed for long term use.
- Most patients living with this chronic condition are treated with medication normally prescribed for the aging population, patients undergoing treatment for cancer etc. This is sometimes helpful but is also not meant to be used on a long term basis as there are side effects which could lead to other problems
- **Ask specific questions and ask supplementary questions and prompts but use clear, literal language and avoid using jargon.**

Functional Impact

A brief summary of the functional impact those living with this condition may experience

Activity 1: Preparing food

Due to the nature of the condition and treatment required, people will often struggle with personal care on worse days and days/weeks before their transfusion. This may present as extreme fatigue, bone pain and breathlessness.
Also consider depression and low mood may be caused secondary to their condition and they may lack motivation to complete activities.

Remember in PIP…

Exploring STAR is imperative for this condition. How do they feel after preparing and cooking a simple meal? What is their level of fatigue/breathlessness and could this be overcome with aids or would assistance be required? Do they need prompting?

Activity 2: Taking nutrition

Patients with thalassaemia can often experience digestive problems due to the side effects of medication. Also, due to iron overload in the pancreas, some patients often suffer from diabetes mellitus and may struggle to consume the right diet. Increasingly, the prevalence of pancreatic insufficiency is on the rise amongst the older patients, which can cause severe weight loss and malnutrition as they are unable to absorb nutrients.

Remember in PIP…

Can they take nutrition to an acceptable standard? Do they need any meal replacements? Can they digest solids? Are they adequately nourished? Do they need prompting to eat for the duration of a meal? Are they at risk of choking? If so, how is this managed?

Activity 3: Managing therapy and monitoring a health condition

Individuals may be required to self-administer intravenous or subcutaneous iron chelation treatments at home and monitor their own condition.

The treatment burden of daily medications combined with symptoms of bone and neuropathic pain can cause the individual to become fatigued on a daily basis. The logistical challenges and pressure of individuals to be responsible for and maintain their own extensive and complex treatment routine directly impacts quality of life.

Remember in PIP…

Consider if they need assistance with medication or therapy that takes place within the home. If so, how long does the therapy take? Do they need any aids or prompts to manage medication?

Functional Impact

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Activity 4: Washing and Bathing

Due to the nature of the condition and treatment required, people will often struggle with personal care on worse days and days/weeks before their transfusion. This may present as extreme fatigue, bone pain and breathlessness.

Also consider depression and low mood may be caused secondary to their condition and they may lack motivation to complete activities

Remember in PIP…

It is not just about completing the activity, but how it is completed. How long does it take them to wash their entire body? How do they feel after? How long does it take to recover? Do they require aids or rely on assistance? Are they motivation to wash on the majority of days?

Activity 5: Managing toileting needs and incontinence

If a patient is experiencing bone pain they may have difficulties getting on and off the toilet. Equally, you must always consider co-existing conditions that may impact their ability to complete this activity.

Remember in PIP…

Mobilising to the toilet is not considered within the scope of the activity. Can they sit and stand from the toilet without difficulty? Do they hold onto anything? If so, why? Can they clean themselves? Do they experience any incontinence?

Activity 6: Dressing and undressing

Due to the nature of the condition and treatment required, people will often struggle with personal care on worse days and days/weeks before their transfusion. This may present as extreme fatigue, bone pain and breathlessness.

Also consider depression and low mood may be caused secondary to their condition and they may lack motivation to complete activities

Remember in PIP…

It is not just about completing the activity, but how it is completed. How long does it take them to dress their entire body? How do they feel after? How long does it take to recover? Do they require aids or rely on assistance? Can their symptoms be reduced by sitting down? Are they motivated to dress on the majority of days?

Functional Impact

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Activity 7: Communicating Verbally

Although Thalassemia itself is unlikely to impact this activity, consider other co-existing conditions that might including any potential cognitive or hearing difficulties.

Remember in PIP…

Someone needs to be able to express and understand verbal information. Do they use any aids? Are they effective? Can they understand what is being said? Do they require BSL or do they lip read?

Activity 8: Reading and understanding signs and symbols

Iron chelation toxicity and diabetes related complications secondary to Thalassemia may cause visual disturbances and restrictions that could impact their ability to read.

Remember in PIP…

Do they use any aids to read? Can they read standard size font unaided? Do they have a CVI? Can vision be corrected with spectacles?

Activity 9: Engaging with others face to face

Although Thalassemia itself is unlikely to impact this activity, consider other co-existing conditions that might. Someone may also have a diagnosis of low mood/anxiety secondary to their Thalassemia that impacts their ability to complete this activity.

Remember in PIP…

Who can they engage with? If they struggle with unfamiliar people, who can support? Why is this? Could they engage with the support of anyone familiar? Could they answer the door or speak to a shop keeper alone?

Functional Impact

A brief summary of the functional impact those living with this condition may experience

Activity 10: Budgeting

Although Thalassemia itself is unlikely to impact this activity, consider other co-existing conditions that might. Someone may also have a diagnosis of low mood secondary to their Thalassemia that impacts their motivational ability to complete this activity.

Remember in PIP…

Can they manage simple and complex budgeting decisions unaided? Can they manage their household bills? Can they understand change in a shop? Do they do online banking/shopping?

Activity 11: Planning and following a journey

Iron chelation toxicity and diabetes related complications secondary to Thalassemia may cause visual disturbances and restrictions that could impact their ability to read.

Remember in PIP…

What is their level of visual acuity? Is vision loss bilateral? How do they safely navigate roads and traffic? Do they utilise orientation aids? Do they require supervision? Have they had any accidents out of the home?

Activity 12: Moving Around

Patients with Thalassemia often experience pain, fatigue and breathlessness that will limit their standing and walking tolerance.

Remember in PIP…

STAR is imperative. Try and gather lived examples of walking. How far is it to the local shop? What pace do they walk? How long does this take? Could they repeat this? Does pace reduce? Do they use aids? How do they feel after?

Additional reading or other resources

EXTERNAL

Thalassemia & ME - YouTube

Thalassaemia - NHS (www.nhs.uk)

Thalassemia - Symptoms and causes - Mayo Clinic

VERSION CONTROL

Version	Date	Signed off by	Comments
1.0	01/09/2021	Shah Faisal	New re-banded document
1.1	14/02/2024	Jade Mayfield	Update of video links.