# Beta Thalassemia

## Mechanism of Disease

Beta thalassemia is a rare, inherited blood disorder characterized by low levels of hemoglobin, which resides in red blood cells and carries oxygen throughout the body.  

### How It Develops

Most beta thalassemia cases are caused by a mutation in the beta (HBB) gene, which provides instructions for making beta-globin, a protein subunit of the hemoglobin protein that carries oxygen to cells.  

### Key Statistics

- **Globally**: 1.5% (50-50 million people)  
- **In the United States**: Less than 200 people

## Diagnosis and Symptoms

Beta thalassemia is usually inherited as an autosomal recessive manner, which means both copies of the HBB gene in each cell have mutations.  

### Beta Thalassemia Affects Men and Women Equally

Both mutations in the HBB gene can result in either:  

- **Beta-thalassemia major**  
- **Beta-thalassemia intermedia**  

### Potential Symptoms

- Anemia  
- Bone and muscle abnormalities  
- Liver and heart problems  
- Respiratory problems  
- Seizures  
- Growth delay  

### Beta-Thalassemia Major

- Bone and muscle abnormalities  
- Heart and liver problems  
- Respiratory problems  
- Seizures  
- Growth delay

With proper disease management, patients can enjoy a near-normal lifestyle, and can experience normal physical and emotional development from childhood to adulthood, including parenthood.

## Treatment and Management

### Complimentary Treatments & Management

- **Splenomegaly**: Splenomegaly may occur due to an enlarged spleen, which may cause severe anemia by destroying too many red blood cells.

- **Iron Overload**: Iron overload is most often managed by blood transfusions, which provide replacement red blood cells to patients living with anemia due to beta thalassemia who are of major or intermedia severity. Advances in red blood cell transfusions have further prolonged the lives of patients with beta-thalassemia minor.

### Splenectomy

- **Indications**:  
  - In patients with severe anemia  
  - To reduce the risk of splenic abnormalities  

### Beta-Thalassemia Major and Intermedia

- **Impact on Quality of Life**:  
  - Patients with beta-thalassemia major and intermedia may require more transfusions as they age.

### Prognosis

In more advanced stages, heart and liver problems such as anasarca (generalized edema), peripheral neuropathy (nervous system abnormalities), and impaired liver function can occur. Beta-thalassemia major patients can also experience heart rhythm abnormalities and liver fibrosis.

Some individuals with beta-thalassemia major and intermedia may may be associated with a severe beta thalassemia and can impact a patient's survival.  

### Table: Beta Thalassemia Types

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Beta-thalassemia major</strong></td>
<td>An almost complete lack of beta-globin, presenting as severe anemia.</td>
</tr>
<tr>
<td><strong>Beta-thalassemia intermedia</strong></td>
<td>Causes mild to moderate anemia and may need regular blood transfusions.</td>
</tr>
<tr>
<td><strong>Beta-thalassemia minor</strong></td>
<td>Causes asymptomatic anemia and may require occasional blood transfusions.</td>
</tr>
</tbody>
</table>

### Challenges

- **Anemia**: Significant reductions in red blood cell counts and hemoglobin levels.  
- **Bone and Muscle Abnormalities**: Abnormalities of the spleen, liver, heart, and muscle tissues.  
- **Liver and Heart Problems**: Abnormalities associated with liver enzymes and cardiac function.  
- **Respiratory Problems**: Respiratory difficulties and breathing issues.  
- **Seizures**: Seizures and neurological complications.  
- **Growth Delay**: Stunted growth and development.

### Beta Thalassemia Treatment

- **Blood Transfusions**: Regular blood transfusions can help increase red blood cell counts and oxygen levels.  
- **Chelating Agents**: Used to reduce iron overload and its associated complications.  
- **Folic Acid Supplementation**: Helps with the production of red blood cells and oxygen levels.  
- **Hepatic and Cardiac Monitoring**: Monitoring for signs of liver and heart abnormalities.

### Beta Thalassemia Minor

- **Symptoms**: Asymptomatic, with mild anemia that may not require transfusions.

### Beta Thalassemia Intermedia

- **Symptoms**: Causes mild to moderate anemia and may need regular blood transfusions.

### Beta Thalassemia Major

- **Symptoms**: Causes severe anemia and may require regular blood transfusions.

### Genetic Counseling

Genetic counseling is a comprehensive process that includes education, assessment, and planning for individuals and families affected by genetic conditions. It involves assessing the risks of inheriting or passing on a genetic disorder, discussing options for prenatal testing, and providing guidance on managing the condition. 

### Disease Management

- **BLOOD TESTS, INCLUDING A COMPLETE BLOOD COUNT AND SPECIAL HEMOGLOBIN TESTS**:  
  - Hemoglobin levels indicate the amount of oxygen carried in red blood cells.

### Monitoring

- **Bone Marrow Transplantation**: Transplanting healthy stem cells from a donor can replace damaged bone marrow and improve blood cell production.

### Splenomegaly

- **Splenectomy**: Surgical removal of the spleen may be recommended to reduce the risk of complications and improve blood cell production.

### Intravenous Iron Infusions

- **Iron Overload Management**: Intravenous iron infusions can help manage iron overload and its associated complications in individuals with beta-thalassemia.

### Iron Chelation Therapy

- **Chelating Agents**: Used to reduce iron overload and prevent complications such as organ damage.

### Other Treatments

- **Folic Acid Supplementation**: Helps with the production of red blood cells and oxygen levels.

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