Hemoglobin H Disease

What is Hemoglobin H (Hb H) Disease?

Hb H disease is an inherited anemia found mainly in people of Asian and Southeast Asian descent. It is a form of alpha thalassemia that affects the body's ability to make healthy red blood cells. Red blood cells carry oxygen throughout the body by attaching it to a protein called "hemoglobin." To make normal hemoglobin the body needs 4 alpha genes and 2 beta genes. People with Hb H disease are missing 3 alpha genes. This makes the red blood cells more fragile, causing them to break down more quickly than normal blood cells.

Hemoglobin H – Constant Spring disease is a more severe form of Hb H disease. In addition to having only 1 working alpha gene, these children also have the "Constant Spring" hemoglobin mutation. This makes the hemoglobin even more unstable and these children generally have more severe symptoms.

Generally, people with Hb H disease live long and productive lives. However, there are some complications that you should know about if your child has Hb H disease or Hb H – Constant Spring disease.

What are some of the complications?

Enlarged Spleen (Splenomegaly)

The spleen is an organ that filters damaged red blood cells from the blood. Your child has many damaged red cells, so the spleen must work harder and may become enlarged. This usually causes no symptoms, but sometimes it may cause severe anemia, abdominal pain, fatigue and frequent infections. In these cases the spleen may need to be removed.

Aplastic Crisis

An aplastic crisis occurs when the bone marrow "shuts down" and fails to make red blood cells. Signs of aplastic crisis include dizziness, pale skin, and extreme fatigue. *Should your child experience these symptoms, contact the hematology team immediately.* Your child might need further tests, and possibly a blood transfusion to help the body recover.

Delayed Growth

Your child has lower levels of hemoglobin (anemia) and slightly lower levels of oxygen throughout the body. This may cause your child to reach puberty slightly later than his/her peers, and may affect his/her growth. Your child will see an endocrinologist (growth doctor) if there is a concern.

Osteoporosis (Low Bone Density)

The same anemia that can cause delayed growth can also cause bones to be thinner than normal. It is important your child participates regularly in weight-bearing exercise like jogging and team sports. Your child should also take calcium and vitamin D supplements every day to help build strong, healthy bones.

Gallstones

When red blood cells break down, they release a substance called "bilirubin" into the blood. Children with Hb H disease have higher levels of bilirubin. This bilirubin can build up and form small "stones" in the gallbladder, called gallstones, which can cause abdominal pain. *Contact the hematology team immediately if your child experiences unusual pain in the upper right side of the abdomen.*

Iron Overload

Later in life, patients with Hb H disease are more prone to iron overload, a condition of excess iron in the body that can damage the liver and heart. Unnecessary iron supplements can increase the risk of iron overload. Your child should **not** take iron supplements unless recommended by the hematology team.

Hemolysis (Red Cell Breakdown)

Certain medications, foods and chemicals can sometimes cause red blood cells to break down more quickly, resulting in severe anemia requiring medical attention. Your child should avoid the following:

Pain relievers:	**Antimalarials:**	**Tuberculosis drugs:**
Aspirin	Primaquine	Isoniazid
Phenacetin	Chloroquine	Rifampin
Acetanilide	Hydroxychloroquine	
Antibiotics:	**Sulfa drugs:**	**Other medications:**
Nalidixic acid	Sulfacetamide (eye drops)	Pyrimethamine
Nitrofurantoin	Sulfanilamide	Vitamin K analogs
Furazolidone	Sulfamethoxazole	Phenazopyridine
Chloramphenicol	Sulfapyridine	Quinidine
B-aminosalicylic acid	Sulfasalazine	Gluconate
Ciprofloxacin	Sulfisoxazole	
Doxycycline	Dapsone	
Other products:		
Toluidine blue dye	Trinitrotoluene (TNT)	Fava beans
Methylene blue dye	Naphthalene/mothballs	

Genetic Implications

Hb H is inherited, which means it is passed from parent to child. There is a risk that your child can pass Hb H disease or a more severe form of alpha thalassemia on to his/her own children. It is important that your child and his/her partner seek genetic counseling prior to having children.

When should I call the doctor?

Call the hematology team if your child has any of the following:

- Extreme fatigue
- Pale/yellow skin or yellowish eyes
- Stomach and/or back pain
- Dark or bloody urine

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