



**GLANZMANN
THROMBASTHENIA**

**AN INHERITED
BLEEDING DISORDER**

**AN INFORMATION
BOOKLET**

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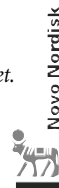


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Introduction

Glanzmann Thrombasthenia is a rare hereditary disease. Learning that you have this disease is not easy. Learning your child has it can be even harder. Feelings of insecurity and frustration are common. What's more, given the rarity of the disease, there is very little written information available to people who are affected.

Our goal is to create a booklet that will be useful to people who each day have to face a health problem whose complications can be mild or serious, depending on the severity of the deficiency.

Glanzmann Thrombasthenia is a disease of blood clotting. It affects the ability of the blood platelets to gather around the site of a broken blood vessel. The platelets are sometimes present in normal quantity but are unable to work normally. To be precise, Glanzmann Thrombasthenia is caused by a deficiency of a protein on the surface of platelets, called *Glycoprotein IIb/IIIa*. This protein is needed so that platelets aggregate around an injury to a blood vessel. Because of the deficiency, platelets fail to form a plug to stop the bleeding.

Discovery of the disease

Glanzmann Thrombasthenia was discovered in Berne, Switzerland in 1918 by a pediatrician named Glanzmann. The children affected by the disease all came from a tiny village, called Le Valais, situated high in the Swiss Alps. In this village there were frequent marriages between close relatives.

How the Disease is Inherited

Glanzmann Thrombasthenia is an inherited bleeding disorder. It is passed on from parent to child at the time of conception. The bleeding problem is caused by an abnormal gene.

Each cell of the body contains structures called chromosomes. A chromosome is a long chain of chemicals known as DNA. This DNA is arranged into about 30,000 units called genes. These genes determine such things as the colour of a person's eyes. In the case of Glanzmann Thrombasthenia, one of the genes on a particular chromosome has a defect.

The defective gene in Glanzmann Thrombasthenia is on a chromosome that does not decide the sex of the child. This means that Glanzmann Thrombasthenia can affect females as well as males. In this way, it is unlike other bleeding disorders such as Factor VIII Deficiency, also called hemophilia A, in which the defective gene is sex-linked – and therefore only males are severely affected.

A *carrier* is a person who carries the defective gene but is not affected by the disease. In order for a person to inherit Glanzmann Thrombasthenia, both parents must be carriers. In such a case, the baby inherits two defective genes, one from the mother and the other from the father.

If a person inherits the defective gene from only one of the parents, he/she will be a carrier, and will usually show no signs of the disease.

The three illustrations below show how Glanzmann Thrombasthenia can be passed on.

Figure 1 shows what can happen when a carrier of Glanzmann Thrombasthenia has children with another carrier. There is a 1-in-4 chance that a child will have Glanzmann Thrombasthenia, a 1-in-2 chance that a child will be a carrier and a 1-in-4 chance that a child will be normal.

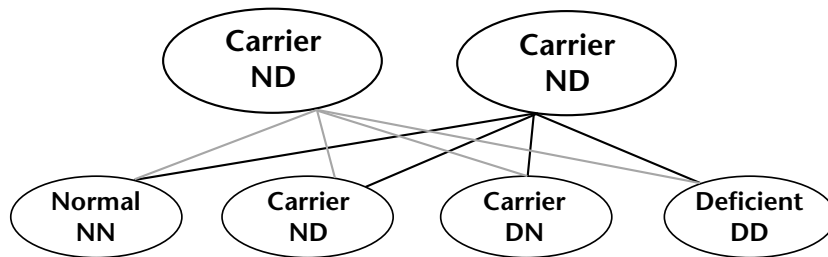


Figure 1

Figure 2 shows what can happen when someone with Glanzmann Thrombasthenia has children with a non-carrier. All the children will be carriers, but none of them will have the disease.

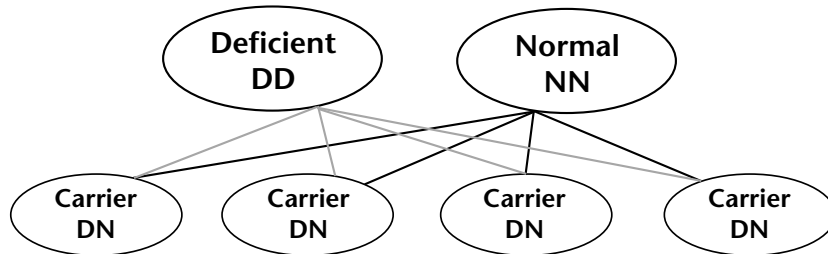


Figure 2

Figure 3 shows what can happen when someone with Glanzmann Thrombasthenia has children with a carrier. There is a 1-in-2 chance that a child will be a carrier. There is also a 1-in-2 chance that a child will have Glanzmann Thrombasthenia.

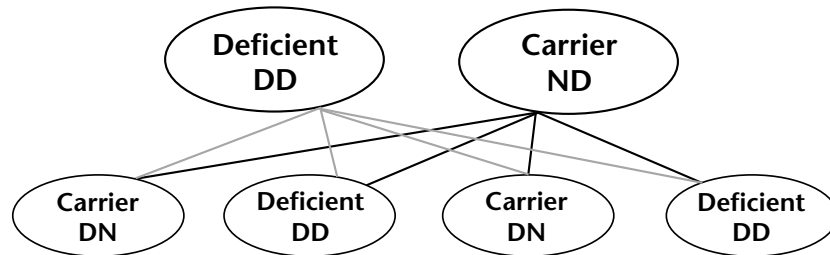


Figure 3

Severity

Glanzmann Thrombasthenia has three categories of severity, depending on the importance of the platelet deficiency in Glycoprotein IIb/IIIa.

- **Type 1 (Severe):** A level less than 5% of normal
- **Type II (Less severe):** A level between 5% and 20% of normal
- **Type III (Least severe):** A variant of Thrombasthenia with levels of more than 50% of normal, but with major abnormalities in the way platelets aggregate.

Symptoms

Children with Glanzmann Thrombasthenia show signs of bleeding during their first year of life. This could be bruising just under the skin, called purpura, or bleeding from mucous membranes, especially in the mouth and nose. Mucosal bleeding tends to be the most serious.

Purpura is very frequent. It often has no apparent cause. Fortunately, it is not dangerous and does not cause the child pain.

Mucosal bleeding into the mouth and nose (with the child's first teeth, or after the child bites his/her tongue or cheek) are the hardest bleeds to control.

Very rarely, a deeper, more important hemorrhage can occur, sometimes in the gastro-intestinal tract. Brain hemorrhages can occur, but they are extremely rare in people with Glanzmann Thrombasthenia.

Bleeding can be caused by a trauma or even by a seemingly harmless event like sneezing, crying, coughing, the eruption of a tooth or a common cold. The oozing can last many hours ... and even days.

Treatment

Fast and appropriate action is the key to successful treatment. These are some of the ways to stop bleeding.

- Prolonged pressure at the site of a cut on the skin or in the mouth.
- Packing in the nose in the event of a nose bleed (epistaxis).
- Amicar™ syrup or Cyklokapron™ tablets to stop clots from being broken down.
- Intravenous transfusion of platelets in the case of a severe bleed.
The transfusion of platelets is an effective treatment for bleeding in patients with Glanzmann Thrombasthenia. However, its use is often limited by the appearance of antibodies that destroy the transfused platelets. It is very important that there be a rigorous selection of appropriate platelets starting with the first transfusions. This will help delay the body's natural defense system from rejecting platelet transfusions that it sees as foreign.
- Recombinant Factor VIIa (Niasase™).

Prevention

Here are some tips that can help prevent bleeds from happening.

- Never take aspirin (ASA) or any medication containing aspirin; use alternate medications recommended by your treatment centre.
- Get vaccinations against influenza and against hepatitis A and hepatitis B – viruses that can still, very rarely, be transmitted by blood transfusions.
- Wear a helmet when riding a bicycle, skiing or playing sports.
- Prevent dental problems and gingivitis. See the dentist every six months. Your centre can recommend a dentist who is familiar with bleeding disorders.
- Keep a small child's nails cut short so they do not accidentally cause cuts in the skin.
- Protect a child from insect bites as these can cause bleeding.

Problems Specific to Women

Menorrhagia

Menorrhagia is the major bleeding problem for women after the age of puberty. The use of oral contraceptives can regularize the menstrual cycles and reduce the bleeding. Cyklokapron or Amicar can be given at the same time. These two antifibrinolytic drugs act by slowing down the body's own destruction of clots that have formed.

Hemorrhages can be especially severe at the time of a girl's first menstruation. Hormonal therapy is sometimes recommended before the first menstruation to avoid serious bleeding.

In certain cases, despite these treatments, transfusions are still needed. Most women who need transfusions have Type I Glanzmann Thrombasthenia.

Bleeding in pregnancy and childbirth

Because Glanzmann Thrombasthenia is so rare, there is very little documentation about bleeding in pregnancy and at the time of childbirth.

A woman with Glanzmann Thrombasthenia who is expecting a child should be followed in a treatment centre that has experience with such patients.

Evolution of the Disease and Prognosis

While the bleeding problem is lifelong, with modern medical care, Glanzmann Thrombasthenia is associated with a low death rate from hemorrhage.

For a woman, the problems associated with Glanzmann Thrombasthenia diminish with time, as she goes through certain stages of life – notably, childhood, puberty and her childbearing years.

The Comprehensive Care Team

As the name suggests, a comprehensive care team provides most of the medical services required by a child or adult with an inherited bleeding disorder. The team is composed of several professionals, including:

- a medical director, usually a hematologist
- a nurse coordinator
- a physiotherapist and
- a social worker.

The team works closely with other specialists—a surgeon, an orthopedist, a rheumatologist, a dentist, a geneticist and a psychiatrist, among others. The purpose of this multidisciplinary team is to ensure the well being of the patient and, in the case of a child, of the parents as well.

For More Information

You can obtain a list of Hemophilia Treatment Centres by contacting the National Office of the Canadian Hemophilia Society:

**National Office
Canadian Hemophilia Society
625 President-Kennedy Avenue
Suite 1210
Montreal, Quebec H3A 1K2
Tel.: (514) 848-0503
Toll-free: 1-800-668-2686
E-mail: chs@hemophilia.ca
Web site: www.hemophilia.ca**

This brochure provides general information only. The CHS does NOT practice medicine and does not suggest specific treatments. In all cases, we suggest that you speak with a doctor before you begin any treatment.

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Novo Nordisk

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We're all related by blood.

