

# GLANZMANN THROMBASTHENIA

AN INHERITED  
BLEEDING DISORDER

AN INFORMATION  
BOOKLET



Canadian Hemophilia Society  
Help Stop the Bleeding



Canadian Association of  
Nurses in Hemophilia Care  
Association canadienne des  
infirmières et infirmiers en hémophilie

Second Edition

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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 and completely normal in situations like this. Our goal is to provide a booklet that will be useful to people who each day have to face this health problem. The complications of Glanzmann thrombasthenia can be variable. Firstly, it depends on whether the person is male or female, and secondly, complications can vary over time. In fact, signs of the disease tend to diminish around adolescence, though they do not disappear completely.

Glanzmann thrombasthenia is a blood clotting disease. It affects the ability of blood platelets to gather around the site of a broken blood vessel. 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 Édouard 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 Glanzmann thrombasthenia is genetically inherited

Glanzmann thrombasthenia is an inherited bleeding disorder. It is passed on from parent to child at 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.

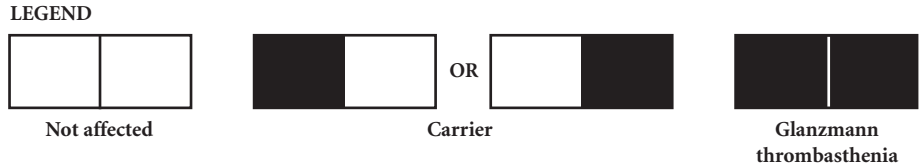
The defective gene in Glanzmann thrombasthenia is on a type of chromosome that does not decide the sex of the child. This means that Glanzmann thrombasthenia can affect females as well as males. In this way, it is unlike other bleeding disorders such as factor VIII deficiency, also called hemophilia A or classic hemophilia, in which the defective gene is sex-linked – and therefore primarily affects males.

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 child inherits two defective genes, one from the mother and one from the father.

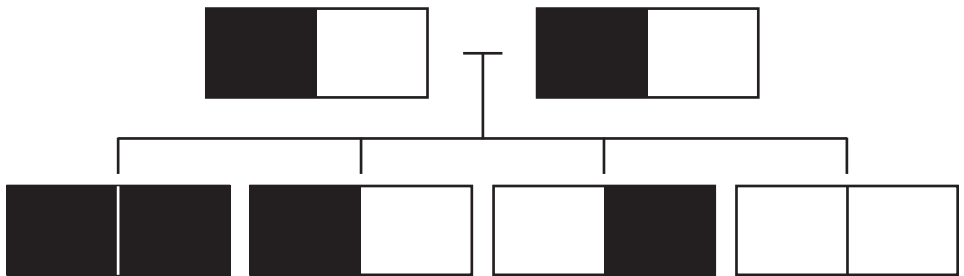
If a child inherits the defective gene from only one parent, he/she will be a carrier, and will usually show no signs of the disease.

A screening test is available for parents to determine whether or not a fetus is affected by the defective gene. However, at present, the test is difficult to obtain. Speak with the team at your bleeding disorder treatment centre, who can help you plan your next pregnancy.

The following four figures illustrate how Glanzmann thrombasthenia can be passed on.

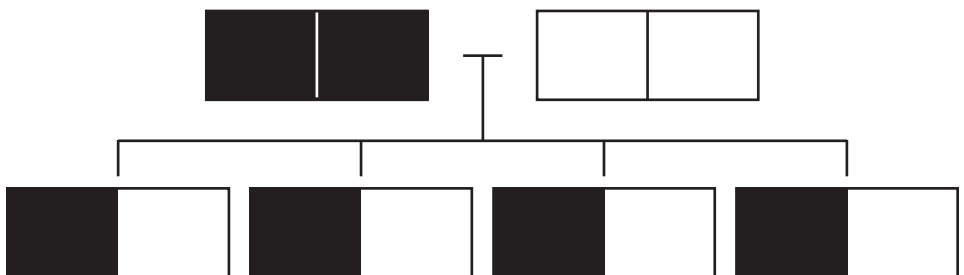


**Figure 1** shows what can happen when a carrier of Glanzmann thrombasthenia has a child with another carrier. There is a 1-in-4 chance that a child will have Glanzmann thrombasthenia, a 1-in-2 chance that a child will be a carrier, and a 1-in-4 chance that the child will not have the disease.



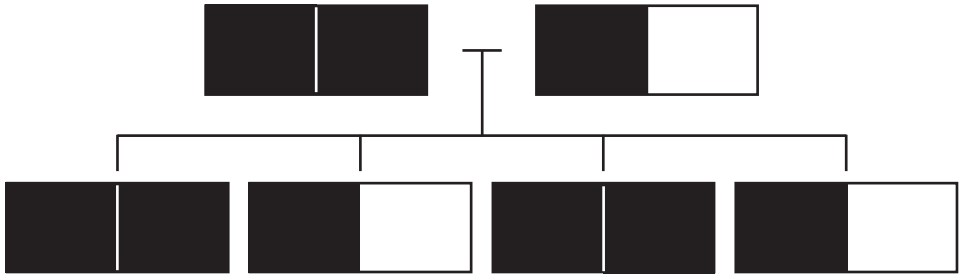
**Figure 1**

**Figure 2** shows what can happen when someone with Glanzmann thrombasthenia has a child with a non-carrier. The child will be a carrier, but will not have the disease.



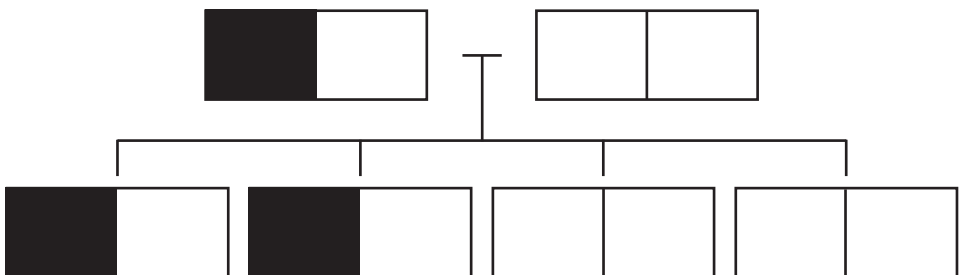
**Figure 2**

**Figure 3** shows what can happen when someone with Glanzmann thrombasthenia has a child with a carrier. There is a 1-in-2 chance that the child will be a carrier, and a 1-in-2 chance that the child will have Glanzmann thrombasthenia.



**Figure 3**

**Figure 4** shows what can happen when a carrier of Glanzmann thrombasthenia has a child with a non-carrier. There is a 1-in-2 chance that the child will be a carrier and a 1-in-2 chance that the child will not have the disease.



**Figure 4**

## Severity

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

- Type I (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.

## Signs and symptoms

The main signs and symptoms related to Glanzmann thrombasthenia are bleeds such as petechiae, ecchymoses (bruises) and mucosal bleeds.

Petechiae are small red spots under the skin. They often develop following localized pressure. In general, they disappear quickly and are of no concern.

Ecchymoses are commonly known as bruises. They tend to draw attention because they cause discolouration and swelling under the skin. However, so long as they do not swell to the size of a golf ball, they are considered harmless and are rarely treated.

Mucosal bleeds are bleeds that involve the mucous membranes of the nose, mouth, intestine, rectum, vagina, and uterus. Mucosal bleeding is often caused by irritation (gastroenteritis, dental cleaning, etc.) and can often be insidious, therefore needs special attention. Treatment is recommended right away at the onset of symptoms.



## Treatment

### At home

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

- Apply direct and prolonged pressure to the wound. The use of KY jelly on the compress helps prevent the gauze from sticking to the wound and from dislodging a newly formed clot when the compress is removed. KY jelly may contain a medicinal ingredient that helps in clot formation. Speak to your physician and nurse about how to treat injuries with first aid.
- Take prescription medication tablets called tranexamic acid (Cyklokapron®), which helps in clot formation.

If these first steps fail to control bleeding after an hour, you must contact the bleeding disorder treatment centre so that a medical assessment can be done.

### At a hospital

In the hospital setting, several procedures can be done such as:

- Blood test to evaluate hemoglobin level, among other things, and if appropriate:
  - Administer a coagulation product called recombinant factor VIIa (NiaStase RT®); in some cases, patients may learn how to manage this treatment at home.
  - Administer intravenous transfusion of blood platelets.

## Problems specific to women

### **Menorrhagia (heavy/prolonged menstruation)**

Menorrhagia – heavy and/or prolonged menstrual bleeding – is the major bleeding problem for women after the arrival of puberty. The use of oral contraceptives or an intrauterine device (Mirena®) can regularize the menstrual cycles and reduce the bleeding. Cyklokapron (tranexamic acid) can be given at the same time. This antifibrinolytic drug acts by slowing down the body's own destruction of blood clots that are formed.

Bleeding can be especially severe during a girl's first menstruation. Hormonal therapy is sometimes recommended before the first menstruation to avoid serious bleeding.

### **Bleeding in pregnancy and childbirth**

Because Glanzmann thrombasthenia is so rare, and there is still little documentation in this regard, we strongly advise that pregnant women be followed at a bleeding disorder treatment centre and by an obstetrician with expertise in the management of care for women with bleeding disorders.

## Sports

Contact sports are not recommended. However, the practice of sports in general and activities such as bicycling and swimming are recommended. It is important to remember to wear the proper protective gear required for different sports. The physiotherapist at your bleeding disorder treatment centre can guide you on how to choose physical activities and sports suitable for your particular circumstances.

## Medications

The use of aspirin and medications that contain ibuprofen (Advil®, Motrin®, etc.) is not recommended. Speak to your pharmacist or bleeding disorder treatment centre about which medicines and natural products you can use..

## Vaccination

Following vaccinations and blood tests, pressure must be applied to the site for at least **15 minutes** to avoid the possibility of major bleeding. Since people with Glanzmann thrombasthenia are more at risk of receiving blood-derived products than in general, vaccination against hepatitis A and hepatitis B is recommended. That said, the blood products available in Canada and many developed countries are extremely safe.

## The comprehensive care team of the inherited bleeding disorder treatment centre

The bleeding disorder treatment centre provides medical care and required services for people with bleeding disorders in order to facilitate management of the disease and to ensure that patients receive safe treatment when particular incidents arise. Consult your nurse for specific information.

The team is generally composed of several professionals, including:

- the medical director, usually a hematologist
- the nurse coordinator
- the physiotherapist
- the social worker and/or psychologist

The team works closely with other specialists – an orthopedic surgeon, rheumatologist, dentist, geneticist and 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.

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## For more information

You can obtain a list of bleeding disorder treatment centres from the Canadian Hemophilia Society Web site ([www.hemophilia.ca](http://www.hemophilia.ca)) or by contacting:

**Canadian Hemophilia Society**  
**Telephone: 514-848-0503**  
**Toll-free: 1-800-668-2686**  
**[chs@hemophilia.ca](mailto:chs@hemophilia.ca)**

*This brochure provides only general information on Glanzmann thrombasthenia. The Canadian Hemophilia Society 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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