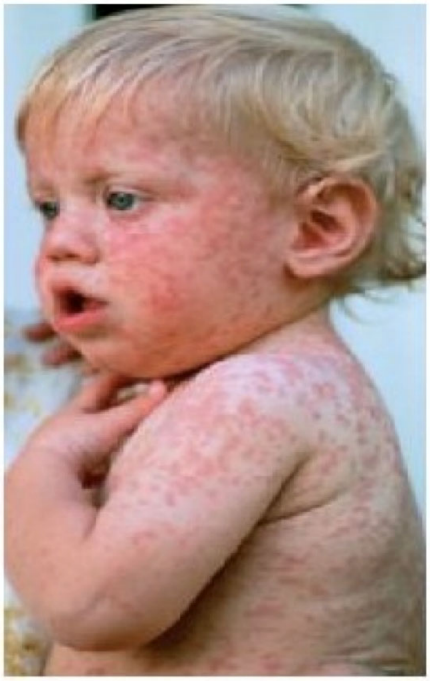
BIOLOGY

Investigating Disease: The Search for Better Health

RUBELLA

PART A



(Image: Rubella Symptoms. Cited

What is Rubella?

Rubella, commonly known as German measles or 3-day measles, is a fairly mild infectious, respiratory, viral disease that is a result of the rubella virus *(Centers for Disease Control and Prevention, 1995)*. Rubella is a common childhood disease that can also affect young adults, and like measles often occurs in epidemics *(Rubella. Cited 18/05/10)*. Humans are the only source of this disease, and can transmit it from one person to another. Rubella is transmitted through airborne droplets or direct contact with discharges from the nose or throat of an infected person *(Communicable disease factsheets, 2003)*. The symptoms of this disease include swollen glands, joint pains, low fever and a fine red rash. Although it is relatively mild in most instances, rubella can have serious complications for pregnant women and may cause miscarriages or serve birth defects in the infected woman's child *(Rubella – Definition. Cited 18/05/10).*

Historical Development

Rubella has been recognised for over two centuries. Originally described in Germany by two physicians, De Bregan in 1752 and by Orlow in 1758, Rubella was regarded as a cross between the measles and scarlet fever *(Resource Packages: German Measles (Rubella). 2008)*.

In 1814, George de Maton was the first person to make a clear distinction between rubella and the scarlet fever, with further clarification coming in 1866 when Henry Veale, an English Royal Artillery surgeon, introduced the term "rubella" from the Latin word meaning "Little Red" *(Rubella -Its Origination & History. Cited 26/05/10)*.

During the past 50 years there have been major advances in our understanding of the disease and its prevention *(Resource Packages: German Measles (Rubella). 2008)*.

In 1914, Alfred Fabian Hess, an American physician, theorised that rubella was caused by a virus, based on experiments with monkeys. In 1938, Hiro and Tosaka confirmed this by passing the disease on to healthy children using a filtered nasal washing from an infected person. In 1940, there was a widespread epidemic of rubella in Australia. Subsequently in 1941, at the annual meeting of the Ophthalmological Society Of Australia, ophthalmologist Norman McAllister Gregg, drew attention to the association between rubella contracted during the first trimester of pregnancy and congenital cataracts in infants born to those mothers. Gregg notified 78 cases of congenital cataracts in infants and found out that 68 of the 78 cases contracted the disease through their mother, who had caught the rubella virus in the early stages of pregnancy *(Resource Packages: German Measles (Rubella). 2008)*. Gregg also described a variety of problems now known as Congenital Rubella Syndrome and noticed that the earlier the mother was infected, the worse the damage was to the fetus. His work stimulated laboratory research scientists to eventually isolate the rubella virus, two decades after his first observations *(Rubella. Cited 26/05/2010)*.

This occurred in 1962, when the rubella virus was isolated in a tissue culture by two separate groups led by physicians Parkman and Weller *(Rubella. Cited 26/05/2010)*. This led to the precise laboratory diagnosis of the disease, and allowed for the next major advance in our understanding of the disease, the development of the rubella vaccines *(Resource Packages: German Measles (Rubella). 2008)*.

Efforts to develop a vaccine for rubella were accelerated between the years of 1962 to 1965 when a worldwide Rubella epidemic broke out *(Resource Packages: German Measles (Rubella). 2008)*. What started in Europe, quickly spread to the USA. Throughout 1964 to 1965, the USA had an estimated 12.5 million rubella cases that resulted in approximately 30,000 stillbirths and 20,000 malformed infants. This epidemic and the availability of the viral culture stimulated research into the development of a vaccine. By 1969, a live, attenuated virus vaccine was available for use and in the early 1970s, a triple vaccine containing attenuated measles, mumps and rubella (MMR) viruses was introduced to prevent rubella *(Rubella. Cited 26/05/2010)*.

Causative Microbe and its Characteristics

Rubella is caused by the pathogen, rubella virus. The rubella virus is an enveloped single-stranded RNA (ribonucleic acid) virus *(Rubella Virus. Cited 20/05/10)*, from the Togaviridae family and the Rubivirus genus. The rubella virus specialises in infecting humans since Homo Sapiens are the exclusive natural host *(Communicable disease factsheets, 2003)*.

Structure

The rubella virus is a spherical virion that is approximately 50 to 60nm in diameter. The virion contains a nucleoid core composed of multiply copies of the rubella virus capsid protein and a single copy of a viral RNA genome. The virus' core is surrounded by a host-derived lipid bilayer that contains 5 to 6nm long spikes, which project out from the virions' surface, as shown below in the figure 1 *(Lee and D. Scott Bowden)*. These viral spike proteins engineer the steps leading to adhesion, fusion and penetration into the host cells. The rubella virus is composed of three structural proteins: two glycoproteins (E1 and E2) and a single non-glycosylated RNA-associated capsid protein. Through the E1 and E2 proteins, the rubella virus interacts with the surface receptors of the host cells, enabling it to enter the cell. From here the virus is able to replicate in the cytoplasm of the host cell *(VIROLOGY. Cited 23/05/10)*.

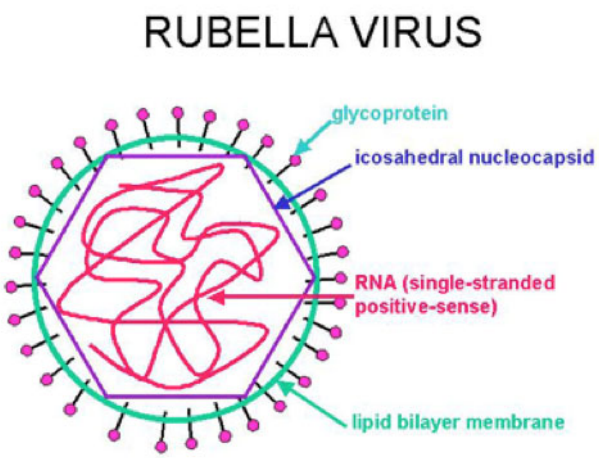


Figure 1: Structure of the rubella virus.

(Image: Structure of rubella virus . Cited 18/05/10)

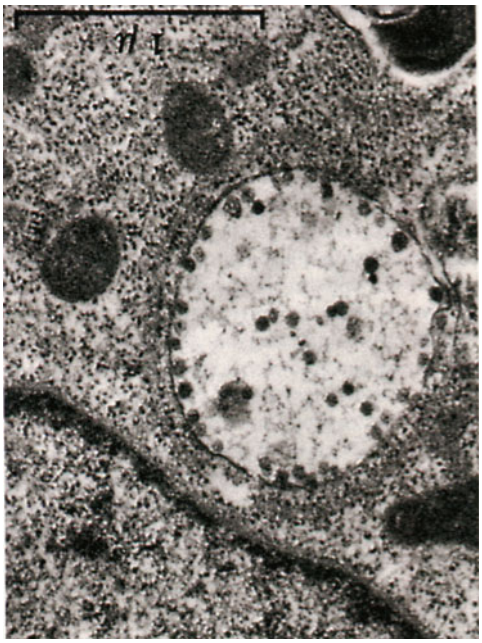


Figure 2: Image of a mature rubella virion.

(Image: Mature rubella virions. Cited 23/05/10)

Transmission Modes

The rubella virus is transmitted through direct or indirect contact with an infected person's bodily fluids, such as mucus, saliva, or water droplets and infects the cells of the respiratory tract. In most cases, rubella is often transmitted when an infected person coughs or sneezes. However, it can also be transmitted when people put their fingers in their mouth or nose after touching an infected surface. But it is also possible for a pregnant woman to transmit the virus to her baby if she becomes infected during pregnancy *(Schoenstadt. 2003)*. A person with rubella can transmit the virus anytime from about seven days prior to the first appearance of the rash to seven days after the recovery of the rash. Even if the rubella symptoms never develop, a person can still spread Rubella if he or she becomes infected with the rubella virus *(Schoenstadt, Rubella Transmission. 2006)*.

How does Rubella transmission occur?

The rubella virus resides in the mucus found in the nose and throat of an infected person. When a person becomes infected with the rubella virus, the virus begins to multiply within the cells that line the back of the throat and nose *(Schoenstadt. 2008)*. When that infected person sneezes or coughs, droplets spray into the air. These infected droplets, in the form of mucus or saliva, can land in other people's noses or throats when they breathe or put their fingers in their nose or mouth after touching an infected surface *(Schoenstadt, Rubella Transmission. 2006)*.

The virus can also spread through to bloodstream or lymph system to various other parts of the body that include:

- Joints
- Thymus
- Eyes
- Testes
- Skin
- Tonsils
- Lungs
- Brain
- Spleen

(Schoenstadt, Rubella Transmission. 2006).

After approximately 14 to 21 days after transmission, the symptoms of rubella start to appear. This period between the rubella transmission and the start of the rubella symptoms is known as the "rubella incubation period" *(Schoenstadt, Rubella Transmission. 2006)*. The development of Rubella from transmission to the symptoms is shown below in figure 3.

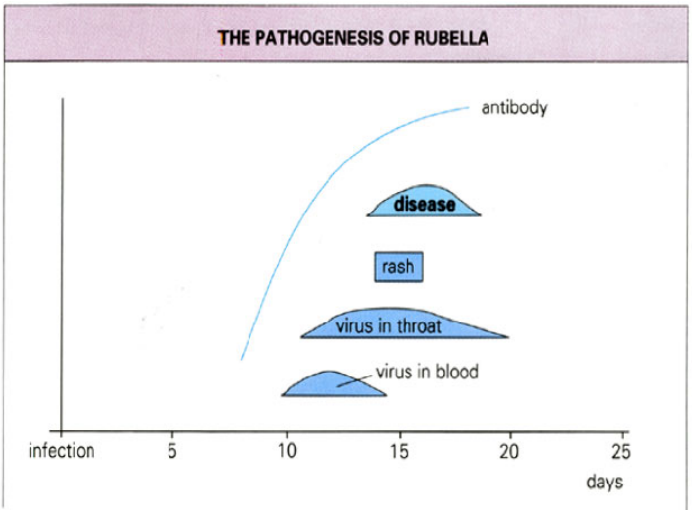


Figure 3: The pathogenesis of rubella
(Image: Pathogenesis of rubella. Cited 18/05/10)

In pregnant women that have been infected with Rubella, the virus can enter the fetus through the placenta, where it can cause birth defects or miscarriages, especially during the first trimester. The organs of the fetus that are developing during this stage of the pregnancy can be seriously affected by the virus, with the most common congenital defects being deafness, cataracts, cardiac defects and Congenital Rubella Syndrome *(Rubella Virus. Cited 20/05/10)*. Infants who have Congenital Rubella Syndrome can transmit the virus to people who have not been immunized through their urine and fluids from their nose and throat *(KidsHealth. Cited 23/05/10)*.

Symptoms

Mild fever, swollen lymph glands, and a red, blotchy rash are some of the possible symptoms of rubella. However, not everyone who becomes infected with the virus will develop symptoms. Because other diseases, such as measles and mumps, can share the same signs as rubella, anyone with possible symptoms should be diagnosed and treated by a doctor *(Schoenstadt, Rubella Symptoms. 2009)*.

Early Rubella Symptoms

The early symptoms of rubella can be the same in both adults and children and usually last three to four days. These symptoms include:

- Mild fever
- Swollen lymph glands. The glands that are most affected by the rubella virus are those located behind the ears and at the back of the neck. The degree to which they become swollen and tender is unique to Rubella *(Golonka. 2008)*.

In adults, other early symptoms can occur from one to seven days before the rubella rash appears. These include:

- Tiredness
- Muscle and body aches
- Joint pain. This symptom is more common in young women, and most often affects the wrists, fingers and knees. It is most common when the rash is present *(Golonka. 2008)*.
- Headache
- Red, water eyes (pink eye)

(Schoenstadt, Rubella Symptoms. 2009)

Other symptoms may include encephalitis, which is the inflammation of the brain and thrombocytopenia, which is the reduction in the number of platelets, which can cause bruising. However, these symptoms are rare.

The Rubella Rash: the classic rubella symptom

The rubella rash is a red, blotchy rash that lasts approximately one to five days. In most cases, the rash lasts three days, which is how Rubella got its other name, "three day measles"*(Schoenstadt, Rubella Symptoms. 2009)*. The rash begins at the hairline of an infected person and then spreads down to the face and upper neck. The rash then gradually moves downward and outward to the limbs, reaching the hands and feet *(Children's Hospital Boston. Cited 23/05/10).* This can be seen in figure 4.

Congenital Rubella

Rubella in a pregnant woman can cause Congenital Rubella Syndrome, with potentially harmful consequences for the developing fetus. Children who are infected with rubella before they are born are at risk of the following:

- Cataracts
- Heart problems
- Mental retardation
- Growth retardation
- Liver, spleen and bone marrow problems.

(KidsHealth. Cited 23/05/10)

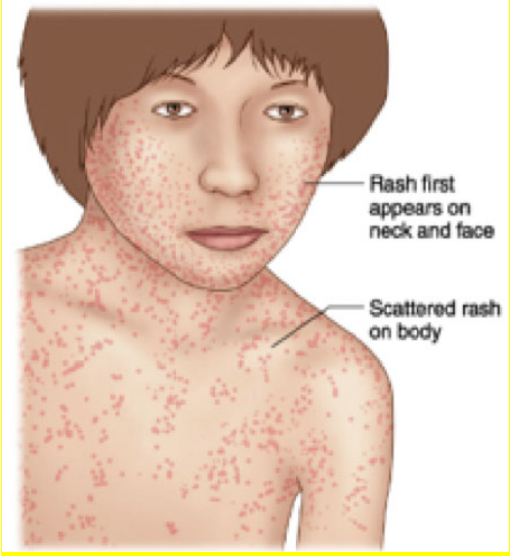


Figure 4: the rubella rash *(Image: Rubella symptoms . cited 2/06/10)*

Treatment and Prevention

Treatment

Currently there is no treatment that can kill the rubella virus. Because a virus causes rubella, antibiotics are not effective *(KidsHealth. Cited 23/05/10)*. Therefore, treatment focuses on providing relief of rubella symptoms as the body takes its own course to fight the infection. This is called supportive care *(Schoenstadt, Rubella Treatment. 2006)*. Supportive care, in the case of rubella includes:

- Increase fluid intake
- Rest
- Medications such as acetaminophen, ibuprofen or paracetamol to control fever and pain.

There are also a number of alternative treatments that have been suggested to make patients more comfortable during the period they have rubella. These treatments include ginger tea or clove tea, which some people believe make the disease run its course faster. Other suggested herbs for the treatment of rubella symptoms include peppermint, cicada, witch hazel and eyebright *(Rubella - Treatment. Cited 26/05/20)*. But unless there are complications, rubella will resolve on its own *(Children's Hospital Boston. Cited 23/05/10)*.

Prevention

Rubella can be prevented by the use of the rubella vaccine. The widespread immunization against rubella is critical in controlling the spread of the disease *(KidsHealth. Cited 23/05/10)*. Since 1969, the vaccine has been available for the prevention of rubella. Rubella vaccine can be given to a person by itself or in a combination with other vaccines (such as measles or mumps). Rubella vaccine is contained within the MMR (measles, mumps, rubella) vaccine and the MR (measles, rubella) vaccine *(Monson. 2009)*.

MMR is a live, attenuated, childhood combination vaccine that provides protection against the measles, mumps and rubella virus. MMR contains the safest and most effective forms of each vaccine, providing immunity to most people *(Monson. 2009)*. This vaccine is generally given in two doses. The first dose is usually administrated when a child is 12 to 15 months old, and will protect them for the rest of their life. However, for 5% of children the first dose of MMR does not work *(Centers for Disease Control and Prevention, 1995)*. Therefore a second dose is given to a child at the age of four to six. However, if 28 days have passed since the first dose was administrated, the second dose can be given to a child before the age of four *(Rubella (German Measles). 2009)*. People ho should get the vaccine include hospital or medical workers, international travellers, women of childbearing age and young adults *(Children's Hospital Boston. Cited 23/05/10)*.

Social and Economic Factors

Before vaccinations began, 8 out 10 people were infected with rubella during their childhood. Most people got the disease when they were 5 to 10 years of age. Now, because of the MMR vaccine, rubella has become a rare incidence in the developed world. However in the developing world, in places such as Latin America and the Caribbean, rubella still remains an endemic. It is estimated in the developing world, that 110,000 or more infants are born with Congenial Rubella Syndrome (CRS) each year. This is because the world's developing countries still lack the rubella vaccination, because of its high costs. It is estimated that the annual cost to treat rubella and CRS in the developing world begins at US$ 50,000 *(MMR (measles mumps and rubella). Cited 1/06/10)*.

In the developing world, in place such as Latin America and the Caribbean, rubella remains an endemic. It is estimated that around 20,000 or more infants are born with CRS each year in these countries. In developing countries the annual estimated cost to treat CRS starts at US$ 50, 000.

Most children receive a routine vaccination against rubella. In many states this vaccination is required for school attendance. However, it is estimated that 10% to 20% of women of childbearing age have still not been vaccinated. People who are most at risk of contracting this disease are foreign-born immigrants who have come from countries where the rubella vaccine is not routinely used and those native-born citizens who refuse immunization because of religious or philosophical reasons *(Rubella Risk. Cited 1/06/10)*.

There are also a number of people who should be vaccinated for the disease. This is because they have an increased risk of contracting the disease. These people include:

- Non-pregnant women of childbearing age.
- Those attending university, trade school or postsecondary school.
- Those working in a hospital, medical facility, child-care center or school.
- Those planning to travel overseas or taking a cruise.

(Rubella. 2008)

Over the past decade, the incidence of rubella has decreased in all age groups, but has especially decreased among children. Therefore, adults account for an increasing number of the few cases that still occur. More than 70% of rubella cases since 2000 have been among adults, compared with 29% in 1991 *(Schoenstadt. 2003)*.

Interesting facts about rubella

Some interesting facts about rubella include:

- The name "rubella" is derived from Latin, meaning little red *(Resource Packages: German Measles (Rubella). 2008)*.
- Rubella is also known as German measles because two German physicians first described the disease in the mid-eighteenth century *(Rubella. Cited 26/05/2010)*.
- The symptoms of rubella start to appear 14 to 21 days after it was first contracted.
- The rubella rash usually lasts 3 days, lymph nodes may remain swollen for a week or more, and joint pain can last for more than 2 weeks.
- Rubella is contagious from seven days before, until seven days after the rash appears.
- 20% to 50% of people with rubella do not exhibit its symptoms.
- As many as 7 million women of childbearing age are susceptible to rubella.
- Up to 7% of young adults are susceptible to the rubella virus.

(Schoenstadt. 2003).

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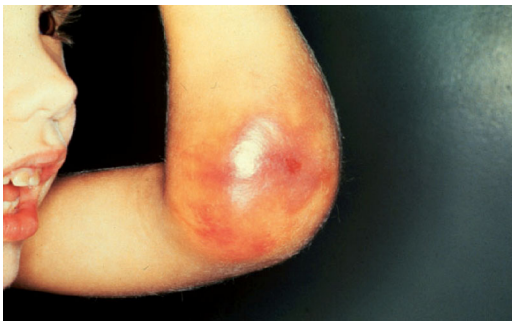
HAEMOPHILIA

PART B



(Image: DIAGNOSIS OF

What is Hemophilia?

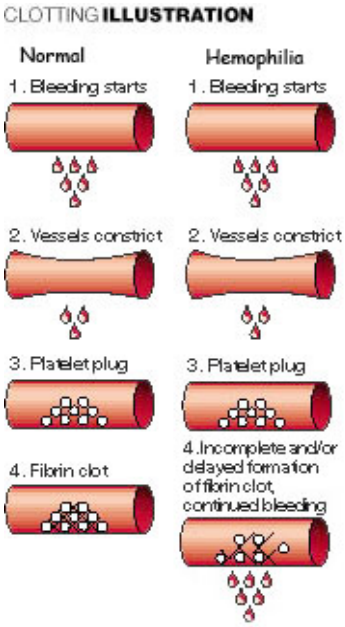


Figure 1: Blood clotting *(Image: Blood clotting. Cited 2/06/10)*

Hemophilia is a rare, sex-linked inherited disease in which the blood does not clot normally *(Schoenstadt. 2008)*. Hemophilia is a genetic disorder, which means that it is a result of one or more abnormal genes that was either inherited (passed on from parent to child) or occurred during the fetus's development in the womb *(Griffin, Hemophilia. 2007)*. People who have it may bleed for a longer period of time following an injury or accident *(Schoenstadt. 2008)*. Human blood contains certain proteins, known as clotting factors. Clotting factors work with platelets (small blood cell fragments) and fibrin to help clot the blood and allow for a blood vessel to heal after an injury *(Griffin. 2007)*. This is seen in figure 1. Our bodies have 12 clotting factors that work together to stop bleeding. Having too little of factor VIII (8) or IX (9) is what causes hemophilia. Therefore, a person with hemophilia is deficient in only one factor, either factor VIII or factor IX, but not both, and as a result their blood cannot clot properly. There are two major types of hemophilia, Hemophilia A and Hemophilia B. Hemophilia A is a factor VIII deficiency and Hemophilia B is a factor IX deficiency *(Griffin, Hemophilia. 2007)*. While this disorder usually occurs only in males, there is the rare exception in which a female will have it. The symptoms of hemophilia include bleeding and bruising, and the treatment in most cases involves replacement therapy, in which the missing clotting factors are replaced *(Schoenstadt. 2008)*. This disease can range from mild to severe, and in its most serious form can lead to death *(Hemophilia – Definition. Cited 27/05/10)*.

Occurrence

Hemophilia is quite a rare disease that mostly affects males. Hemophilia occurs worldwide and can affect a person of any race. Approximately 1 out of every 5,000 male babies is born with Hemophilia A, while 1 out of every 30,000 male babies is born with Hemophilia B. This shows that Hemophilia B is far less common *(Parks. 2009)*.

Hemophilia in females is extremely rare, though it can occasionally happen. It is more common for females to be carriers of the gene that causes hemophilia, which results in the female having either very mild symptoms or none at all *(Facts on the Condition. Cited 31/05/10)*.

Cause

Hemophilia is not contagious like a cold or flu. It is usually inherited, which means that it is passed down from a parent to their child *(Human Diseases and Conditions. Cited 29/05/10)*. Specifically, hemophilia is caused by mutations in the factor VIII (F8) or factor IX (F9) genes. Mutations in the F8 gene cause Hemophila A, while Hemophilia B is caused by mutation in the F9 gene. These genes are located on the X chromosome, which determines the gender of a baby. In most cases hemophilia is passed from a female carrier to her son, however, it is possible for boys with hemophilia to be born to mothers who are not carriers when there is a random mutation in the gene as it is passed on to the child. Although it is rare, it is possible for a girl to be born with hemophilia *(Schoenstad, Causes of Hemophilia. 2008)*. Both Hemophilia A and B are caused by a genetic defect present on the X chromosome. Approximately 70% of people with hemophilia inherit the disease, while the remaining 30% develop the disease due to spontaneous genetic mutation *(Hemophilia – Causes. Cited 25/05/10)*.

Inherited Hemophilia

Hemophilia is inherited in an X-linked recessive pattern. This is because the disorder is located on the X chromosome. Therefore, it is caused when a defective X chromosome gene is inherited. Men and women each have 23 pairs of chromosomes. Half of the chromosomes come from the father, and the other half comes from the mother. Women have two X chromosomes, while men have one X and one Y chromosome *(Hemophilia – Causes. Cited 25/05/10)*.

The genes for making clotting factors are located on the X chromosome. This means that males are more likely to have hemophila compared to females, as they only have one X chromosome *(Hemophilia – Causes. Cited 25/05/10)*. A female carrier of hemophilia has the defective gene for F8 or F9 on one of her X chromosomes. Therefore, there is the risk that she will pass the defective gene on to her children.

- If she has a son, there is a 50% chance that he will have hemophilia.
- If she has a daughter, there is a 50% chance that she will be a carrier.

(Schoenstad, Causes of Hemophilia. 2008).

Males who have hemophilia cannot pass the disease on to their sons; however, all of their daughters will be carriers of the disease *(Schoenstad, Causes of Hemophilia. 2008)*. For a female to develop hemophilia, she would have to receive the defective chromosome from her father, who would have hemophilia, as well as the defective X chromosome from her mother, who would be a carrier. Although this is not impossible, it is highly unlikely *(Griffin, Hemophilia. 2007)*.

These patterns of the inheritance for hemophilia can summarised in a pedigree, like the ones shown below in Figure 2. These two pedigrees show how hemophilia is carried on the X chromosome and is passed down to the offspring. They also show the differences between the inheritance of hemophilia from a carrier mother or an infected father.

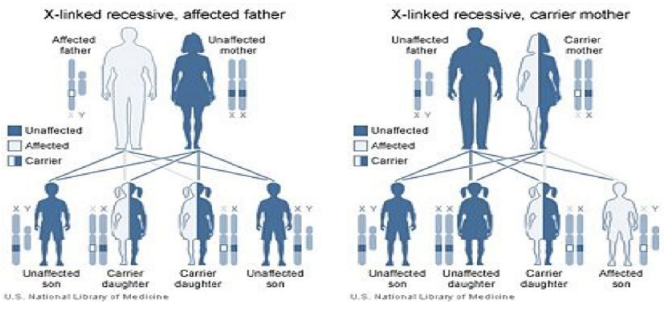


Figure 2: Pedigree of Hemophilia

Spontaneous Genetic Mutation

About 30% of all people with either Hemophilia A or B are the first members of their family to ever have the disease. These individuals get the disease through spontaneous genetic mutation. This is caused during the early development of the fetus, where some random genetic accident (mutation) can cause a defect in the X chromosome, resulting in the development of hemophilia *(Hemophilia – Causes. Cited 25/05/10)*.

Symptoms

For people with hemophilia, the common symptoms include bleeding and bruising. The symptoms of hemophilia vary, depending on the severity of the factor deficiency and the location of the bleeding. The severity of hemophilia symptoms can be categorized as mild, moderate, or severe. In severe cases, symptoms most commonly include internal bleeding in the joints. *(Schoenstadt, Hemophilia Symptoms. 2008)*.

Early Symptoms

In most children, the first symptoms of hemophilia often include:

- Heavy bruising and bleeding from the gums as they begin to get their baby teeth.
- Bumps and bruises from frequent falls as they learn to walk.
- Swelling and bruising from bleeding in the joints, soft tissue and muscles.

(Schoenstadt, Hemophilia Symptoms. 2008).

Symptoms in Children and adults

The most common symptoms in older children and adults include things such as:

- Bleeding in the joints, which is also known as hemarthrosis.
- Bleeding and bruising in the soft tissue and muscles.
- Bleeding in the mouth from a cut, bite or loss of teeth.
- Nosebleeds for no apparent reason.
- Blood in the urine, which is also known as hematuria. This is a result from bleeding in the kidneys or bladder.
- Blood in the stool. This is a result from bleeding in the intestines or stomach.
- Bleeding in the brain.

(Griffin. 2007).

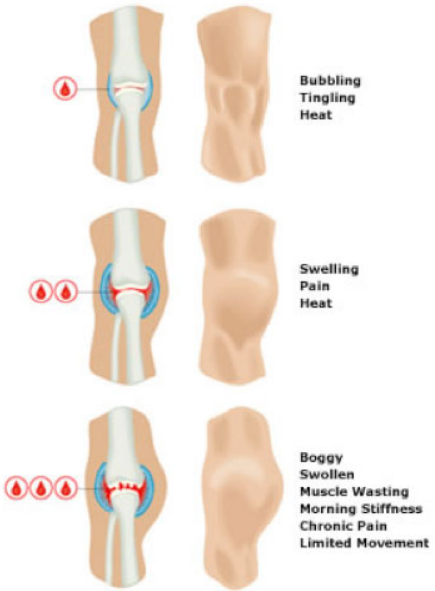


Figure 3: Swollen Joint Symptoms
(Image: Bleeding Joints. Cited 31/05/10)

Bleeding in the Joints

Bleeding in the joints is the most common symptom associated with hemophila. This bleeding often occurs with an injury and can last for days if it is not treated. Although bleeding can occur in any joints, it mostly occurs in the knees, ankles and elbows. The symptoms of bleeding in the joints include:

- Tightness in the joint.
- Pain and swelling
- A warmth feeling in the affected area.
- Loss of movement in the joint.

The development of these symptoms are illustrated in figure 3.
(Schoenstadt, Hemophilia Symptoms. 2008).

Treatment and management

Treatment

Hemophilia is a lifelong disease with no permanent cure. However, it can be successfully treated temporarily.

The treatment of hemophilia is determined by how severe the disease is *(Treatment Overview. 2009)*. In most cases of hemophilia, treatment involves replacing the clotting factor that is too low or missing, in what is known as replacement therapy. Other options for treatment include a synthetic hormone know as desmopressin and antifibrinolytic drugs *(Schoenstadt, Hemophilia Treatment. 2009)*.

The primary treatment for moderate to severe hemophilia is replacement therapy, which replaces the blood's deficient clotting factor. Replacement therapy can be used to prevent bleeding (preventive therapy) or to stop bleeding when it occurs (demand therapy) *(Schoenstadt, Hemophilia Treatment. 2009)*. The clotting factors that are used to treat this disease can either come from donated blood or from laboratory produced clotting factors, called "recombinant" factors. The clotting factors are transfused into a person's blood through an intravenous (IV) line, that be given in the hospital, at a doctor's office or at home *(Griffin, Hemophilia. 2007)*.

People, who undergo preventive therapy, receive it on regular basis to keep the levels of clotting factors high enough so that the bleeding will not occur. This hemophilia treatment is usually given to people with severe cases, and is often used in children to prevent damage to their joints. Preventive treatment is can be intensive and expensive, and is generally given to patients at their home. People, who suffer with a mild or moderate case of hemophilia, undergo demand therapy. This is given as it as needed or on demand to stop the bleeding as soon as possible after it begins. This type of replacement therapy aims to prevent damage to joints and muscles and to prevent other parts of the body from bleeding *(Schoenstadt, Hemophilia Treatment. 2009)*.

Desmopressin (DDAVP) is a synthetic hormone that is used to treat people with mild or moderate Hemophila A. This hormone stimulates the release of factor VIII and von Willebrand factor that is stored in the blood vessels *(Human Diseases and Conditions. Cited 29/05/10)*. The von Willebrand factor carries and binds to factor VIII, enabling it to stay in the bloodstream longer. This therefore increases the level of these proteins in the blood. This temporary treatment is usually given by an injection or in a nasal spray. Antifibrinolytic drugs, including tranexamic acid and aminocaproic acid, are medicines used with clotting factors for the treatment of hemophilia. They are usually given to patients as a pill, to help keep clots from breaking down. *(Schoenstadt, Hemophilia Treatment. 2009)*.

A simple treatment for hemophilia is to apply first aid as soon as possible to limit the amount of bleeding and damage. RICE – Rest, Ice, Compression, and Elevation – is an important technique used to treat bleeding in the muscles and joints. The application of ice is useful in decreasing inflammation and may help to decrease the pain and limit the bleeding. Elevation of the injured area is used to slow blood loss by lowering the pressure in the joint or muscle *(Schoenstadt, Hemophilia Treatment. 2009)*.

Management

People living with hemophilia can take a number of steps to remain healthy, and prevent illness and bleeding problems. There are many things that can be done to enable a person living with the disease to live a healthy life *(Human Diseases and Conditions. Cited 29/05/10)*.

For parents who have children with hemophilia, there are a number of things they can do to try and prevent or reduce the occurrence of bleeding. Doctors recommend choosing soft toys with no sharp corners and padded clothing, such as elbow pads and kneepads, while the child is learning to walk examinations *(Human Diseases and Conditions. Cited 29/05/10)*. Parents should also encourage their children to tell them when they sense a bleed, as a quick infusion can prevent any long-term damage *(Griffin, Hemophilia. 2007)*. Dental care is just as important for children with hemophilia, so parents should teach their children to regularly clean their teeth and visit a dentist, to prevent tooth decay and gum disease. Finally parents can take their children to get regular check-ups that include joint and muscle examinations *(Human Diseases and Conditions. Cited 29/05/10)*.

Exercise is important for adults and children with hemophilia. Regular exercising helps keep the muscles flexible and strengthens the joints, therefore decreasing bleeding episodes. Sports such as swimming, cycling and walking are great because they don't put pressure on the joints. However contact and team sports are not safe for people with hemophilia *(Griffin, Hemophilia. 2007)*.

It is also important for people with hemophilia to maintain a healthy weight, as extra kilograms can strain the body *(Griffin, Hemophilia. 2007)*.

Finally another great way to manage hemophila is for parents to send their children to a camp where they can meet and interact with other children who have hemophilia. This aims to help children work toward being able to give themselves clotting factor replacement therapy, allowing them to gain control of their condition *(Griffin, Hemophilia. 2007)*.

Control

The hemophilia gene runs in families and is passed down from a parent to their child. People with a family history of hemophilia can undergo genetic counseling before having children. Genetic counseling is a service that provides information and support to people who have, or may be a risk of a genetic disorder, and is a way of controlling the occurrences of that disease. Genetic counseling can help couples determine the risk of a child being born with hemophilia or of being a carrier of the gene. This then enables them to make an informed choice about having children where there is a possibility that the child might have hemophilia. Genetic counseling may include genetic testing and prenatal screening *(Hemophilia Genetics: Hemophilia in Families. 2010)*.

Genetic testing involves examining a person's DNA – taken from cells in a sample of blood, or sometimes from other body fluids or tissues – for an abnormality that signifies a disease. For families with a history of hemophilia, genetic test are used for carrier screenings. This involves identifying an individual who may or may not be a carrier of the gene that causes a certain disease. This test allows potential or known carriers within a family to be identified. This can help people determine the potential risks of a child being born with hemophilia or as a carrier of the gene *(Gene testing. 2008)*.

Prenatal testing is a way of stopping in further occurrences of hemophilia, but is however a personal decision to be made by a couple. Prenatal testing involves testing the fetus during the early stages of pregnancy, to determine whether or not the fetus has the certain genetic disease. This examination can help a couple to determine whether or not to continue with the pregnancy *(Prenatal Diagnostic Testing. Cited 1/06/10)*.

Genetic counseling is important in regulating the incidence of hemophilia and in controlling any further occurrences of the disease *(Biology in Focus)*.

New Treatments and Research

New advances have been made in the treatment of hemophilia, allowing most patients to live a full and healthy life. At present, there is no cure for hemophilia, but many trials with gene therapy are underway. Gene therapy is the replacement of a defective gene in a person or organism that is

suffering from a genetic disease. Hemophilia gene therapy research seeks to replace the defective gene that causes hemophilia with a normal, fully functional, gene that will raise the level of the deficient clotting factor *(Human Diseases and Conditions. Cited 29/05/10)*. Researchers have been successful in developing healthy replacement genes for use in hemophilia clinical trials. These genes will be transferred into the body using a vector, which is a method of transportation for a gene. In past clinical trials, gene vectors have been used to cure lab animals of Hemophilia A and B. The future for gene therapy clinical trials will involve human subjects, as researchers need to know if the new gene can be passed on to future generations. Gene therapy will allow people to have continuous infusions of clotting factors, either under the skin or in pill form. It may also eventually help people with hemophilia to begin producing their own clotting factors *(Future Therapies. Cited 30/05/10)*.

Other researchers have also started to produce genetically engineered animals cells into which they have inserted the genetic sequence to produce human factor VIII *(Human Diseases and Conditions. Cited 29/05/10)*. An example of this is a new product known as ADVATE Antihemophic Factor. This product is produced using genetically engineered Chinese hamster ovary cells that have been altered to produce the deficient factor VIII. This treatment is used for people with Hemophilia A and prevents and controls bleeding episodes *(New treatment for hemophilia. 2003)*.

Cryoprecipitate therapy is another new treatment for hemophilia. This is similar to factor VIII replacement therapy, but cryoprecipitate uses donated blood, where the plasma is separated and then quickly frozen and then thawed, so that the factor VIII can be separated from the plasma. This is used for the treatment of Hemophila A. Lyophilization is a similar treatment that rapidly freezes and dehydrates the plasma. The freeze-dried plasma is then quickly dissolved, so that the clotting factors can be separated from the plasma and infused into a patient with hemophila *(New treatment for hemophilia. 2003)*.

Finally, the development of clotting factors made in a laboratory has virtually eliminated the risk of contracting a transfusion-related infection of HIV or hepatitis C. These two infections are a problem in hemophilia treatment as they are caused from the donated blood used for replacement therapy *(Future Therapies. Cited 30/05/10)*.

Interesting facts about hemophilia

Some interesting facts about hemophilia include:

- The name hemophilia is derived from the Greek words, “hemo” meaning blood and “philia” meaning tendency towards.
- Hemophilia has been indirectly known about since the second century AD.
- Hemophilia has played an important role in Europe’s history, for it suddenly cropped up in the children of Great Britain’s Queen Victoria and Prince Albert.
- Hemophilia is referred to as the “Royal Disease” because it was inherited through the royal families of Europe through the descendents of Queen Victoria.
- A carrier mother has 50% chance of passing the disease to her daughter, making her a carrier as well.
- An affected father will always pass on the affected gene to his daughter, making her a carrier.
- An affected father can never pass on the disease to his son.
- Hemophilia is a lifelong disease.

(Schoenstad, Causes of Hemophilia. 2008).

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