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ISSN: 2161-1041
Hereditary Genetics: Current Research

Opinion Article

Genetic Disorder Haemophilia: Its Diagnosis and Management

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DESCRIPTION

Haemophilia, often known as haemophilia (from the Ancient Greek (haîma) "blood" and (phila) "love of"), is a genetic illness that is primarily inherited and affects the body's capacity to form blood clots, which are necessary to stop bleeding. As a result, people bruise easily, experience prolonged bleeding after injuries, and are more likely to experience internal bleeding into joints or the brain. Only after an accident or during surgery may those with a mild type of the illness experience symptoms. While bleeding into a joint may cause irreparable harm, bleeding into the brain may cause convulsions, chronic headaches, or a loss of consciousness.

Haemophilia may be divided into two main categories: haemophilia A, which is caused by low levels of clotting factor VIII, and haemophilia B, which is caused by low levels of clotting factor IX. Typically, they are inherited from one's parents *via* an X chromosome that contains a gene that is not functioning. Rarely, a novel mutation may appear during the early stages of development or haemophilia may appear later in life as a result of antibodies developing against a clotting factor. Haemophilia C, which is brought on by low levels of factor XI, Von Willebrand disease, which is brought on by low levels of a substance called von Willebrand factor in their blood, and parahaemophilia, which is brought on by low levels of factor V, are some of the other types.

The intrinsic route, a clotting mechanism required when a blood vessel's endothelium is damaged, is rendered ineffective by haemophilia A, B, and C. Pregnancy, inflammatory diseases, and malignancies are linked to acquired haemophilia. By analysing the blood's clotting potential and clotting factor concentrations, the diagnosis is made.

One method of prevention is to remove an egg, fertilise it, and test the embryo before putting it in the uterus. The technological item or process in research can be thought of as human embryos. In order to treat haemophilia, missing blood clotting factors are replenished. This may be carried out routinely or during periods of bleeding. Replacement procedures can be done in a hospital or at home. Either human blood or recombinant techniques are used to create the coagulation factors. 20% of persons can acquire anti-clotting factor antibodies, which makes therapy more challenging. People with mild haemophilia A may use the drug desmopressin. Early human studies for gene therapy are underway. Around 1 in 5,000–10,000 male infants are born with haemophilia A, while this number is closer to 1 in 40,000 for haemophilia B. Females are rarely seriously impacted by haemophilia A or B because both conditions are X-linked recessive diseases. Some girls who have an inactive gene on one of their X chromosomes may exhibit modest symptoms.

Diagnosis

If there is a family history of the disorder, haemophilia might be identified before, during, or after delivery. Parents have a variety of possibilities. Haemophilia is typically only discovered after a kid starts to walk or crawl, even if there is no family history of the condition. They could bleed from their joints or bruise easily. Mild haemophilia may not be identified until much later, typically following an accident or a dental or surgical operation. Before Pregnancy, To ascertain the likelihood of passing the illness on to a child, genetic testing and counselling are both available. In order to do this, a tissue or blood sample may be examined for indications of the genetic mutation that results in haemophilia. During pregnancy, the haemophilia gene can be tested for by a pregnant woman with a family history of haemophilia. These tests consist of Chorionic Villus Sampling (CVS) usually between weeks 11 and 14 of pregnancy; a tiny sample of the placenta is extracted from the womb and tested for the haemophilia gene. Amniocentesis: During weeks 15-20 of pregnancy, a sample of amniotic fluid is obtained for testing. The woman may discuss this with the doctor in charge of her care because there is a slight chance that these procedures could result in issues like miscarriage or early labor. After birth, after a child is born, if haemophilia is suspected, a blood test may typically confirm the diagnosis. If there is a history of haemophilia in the family, umbilical cord blood can be

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Received: 04-Nov-2022, Manuscript No. HGCR-22- 19033; **Editor assigned:** 07-Nov-2022, PreQC No. HGCR-22-19033 (PQ); Reviewed: 21-Nov-2022, QC No. HGCR-22-19033; **Revised:** 28-Nov-2022, Manuscript No. HGCR-22-19033 (R); **Published:** 05-Dec-2022, DOI: 10.35248/2161-1041.22.11.229

Citation: Twain E (2022) Genetic Disorder Haemophilia: Its Diagnosis and Management. Hereditary Genet.11: 229.

examined at birth. Additionally, a blood test can determine whether a kid has haemophilia A or B and the severity of illness.

Management

There is no permanent treatment. Replacement of the deficient blood clotting factors is the main method of treatment and prevention of bleeding episodes.