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Review on HAEMOPHILIA

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Abstract-

Haemophilia is a hereditary disorder that impairs body's ability to control blood clotting or coagulation when blood vessel is broken. It is mainly of 3 types: - Haemophilia A, Haemophilia B, vonwillbrand disease. In absence of factor viii it may happen. Female is the carrier of the disease & male are highly affected by it. No proper treatment has been found, but there is a way to minimize it. Many complications are observed I the disease like deep internal bleeding, joint damage, transfusion transmitted infection, etc. "joint Bleed", "muscle Bleed" form in the disease.

INTRODUCTION-

Haemophilia is a group of hereditary genetic disorder. The body looses ability to control blood clotting when blood vessel is broken. It is mainly of 3 types:- Haemophilia A(clotting factor vii deficiency) is the most common form of disorder, present in about 5,000-10,000 male born. Haemophilia (factor ix deficiency) happens in about 1 in 20,000-34,000 male born. Von-willibrabds disease happens due to insufficient level of factor viii and abnormality of platelets adhesiveness. It is an sex linked disorder – x chromosome disorder. It is more possible in men than women. A woman has two s chromosome but men has only one x chromosome. So defective gene is guaranteed to manifest in any male who carries it. The chance of female having 2 defective copies gene is very exceptional. Female can inherit the defective gene from their mother or father or it may be new mutation. In Haemophilia clotting factors levels for a normal clotting is missing. In severe haemophilia, a minor injury can result blood loss lasting for days or weeks or may not heal completely. In areas such as brain or inside joints, this can be fatal or permanently debilitating.

SIGNS AND SYMPTOMS:-

Characteristic symptom varies with severity.

1) In general symptoms are internal or external bleeding episodes which are called “bleed”. Patients with more severe haemophilia suffer more & bleeding occurs more frequently while patients with mild haemophilia suffer less, symptoms diminishes after surgery.
2) Moderate haemophlias are variable which manifest between severe & mild forms.
3) In both haemophilia A & B there is a spontaneous bleeding but a normal bleeding time, normal prothrombin time, normal thrombin time but prolong partial thromboplastin time.
4) The most characteristic type of internal bleed is a joint bleed where blood enters into joint space. It is the most severe haemophilia.
5) Females who are carriers have enough clotting factor from their one normal gene to prevent serious bleeding problem, though some may present as mild haemophilias.

COMPLICANTS:-

Severe complications are much more common in severe & moderate haemophilias.

Deep internal bleeding: deep muscle bleeding, leading to swelling pain of limb.

Joint damage: - from haemathrosis, potentially with severe pain & disfunction of joints.

TRANSFUSION TRANSMITTED INFECTION:-

From blood transfusion that are given during treatment. Intracranial haemorage is a serious medical emergency caused by build up of pressure iside the skull. It may cause nausea, disorientation, brain damage.

LIFE EXPECTANCY:-

Like most aspects of disorders, life expectancy varies with severity & treatment. By the 1980s the life span of average haemophiliac receiving appropriate treatment was 50-60 years. Now, medical science is improving day by day. With appropriate treatment males with haemophilia have normal life span approximately to 10years shorter than unaffected male.

CAUSE

Human beings have 46numbers of chromosomes. 2of them have sex determining capacity. In female they are called XX, in male they are one X & another Y.

So, structure of female chromosome = 44+XX.

Structure of male chromosome = 44+XY.

The gene which helps to synthesize factor VII present in X chromosome. The female have two X chromosomes, one is normal & another is defective. The females are called carriers. They have no problem to blood coagulation. But a carrier married with a normal male, if their baby is male, he has a chance to attack haemophilia.

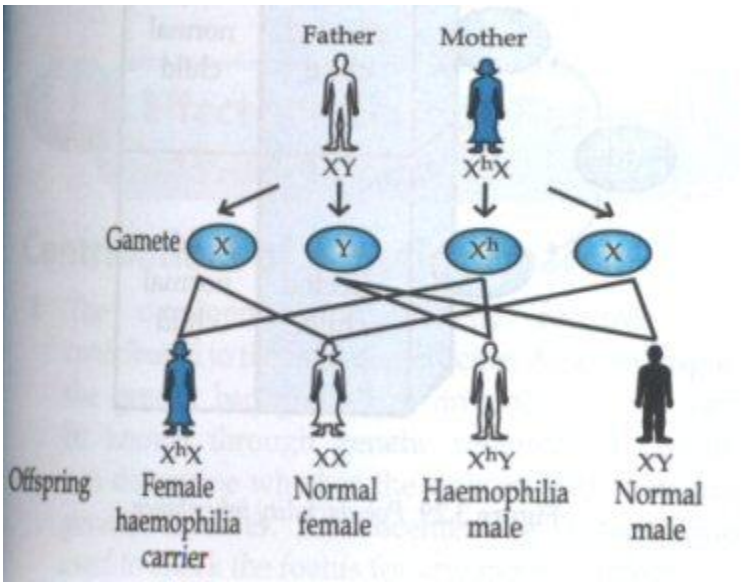

Fig:1. CLASSIFICATION OF HAEMOPHILIA BASED ON FACTOR LEVEL.

44+xx =normal female

44+XY= Normal male

44+XX^h= Carrier female

44+X^hY= Haemophilic male

CLASSIFICATION OF HAEMOPHILIA BASED ON FACTOR LEVEL:-

TYPE	% NORMAL FACTOR LEVEL
Severe	<1
Moderate	(1-5)
Mild	(6-30)

CLINICAL FEATURES OF HAEMOPHILIA:-

Joint bleed- Main symptoms of joint bleed are bleeding in the knees, elbow or other joints. This bleeding can happen without external or internal injury. First the bleeding causes tightness in joint with no real pain or any visible sign of bleeding. The joint become swollen, hot to touch & painful to bend. Two symptoms are related to joint bleed:-

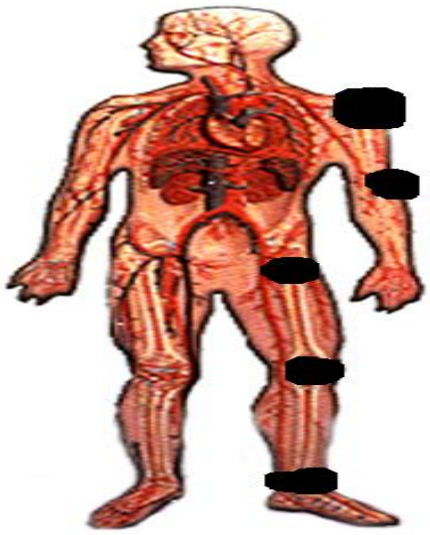

FIG: 2. This is a diagram of the joints most commonly affected by Hemophilia. It most often occurs at the knees, hips, ankles, shoulders, and elbows.

A. Sub acute haemarthrosis
   - Synovium becomes inflamated
   - Develops after repeated bleed into joints.
   - Hyperthrophy, hyperplasia.
   - Hemosiderosis: haemoglobin of intra articular blood is degraded & iron deposits into joint spaces.

B. Chronis athropathy:-
   - Progressive destruction of a joint.
   - Enzymes begin to destroy articular cartilage.
   - Microfaction & cyst formation in surodial bone.

Muscle bleed: - The bleeds are one common type of bleeds for persons with haemophilia. Muscle bleed can happen in a group of muscle. Bleeding into muscles or soft tissue. Sites of muscle bleeds are calf, upper arm, shoulder area. Symptoms are pain, swelling, muscle spasm, warmth, less flexibility of muscle & skin tightness.

MANAGEMENT:-

No proper is found in haemophilia.

- Factor VII is given by injection form.

- Plasma may be given
- Cryoprecipitate is used for this disease.

At present some treatment are useful to treat haemophilia.

GENE THERAPY:-

On 10th December 2011, a team of British & American investigators reportated successfully He, B gene therapy. The gene investigator inserted F9 gene into an adeno-associated virus & vector which has a propensity for liver where factor 9 is produced & remains the chromosomes as not to disrupt other genes. To prevent rejection the patients were primed with steroids to suppress their immune response.

PREVENTIVE EXERCISE:-

- Some specific helpful for the affected people.
- Exercise increases flexibility, tone, strength of muscle, increase ability of joints.
- The exercises are stretching of calves, ankle circles, elbow flexions, standard sports warm-up.

CONTRAINDICATIONS:-

- Anticoagulants such as heparin & warfarin are contraindicated for people with haemophilia, as these can make clotting difficult.
- Other medications- ibuprofen, naproxen sodium, because they are well kown to have side effect of prolong bleeding.
- Football, hockey, boxing, rugby should be avoided by people with haemophilia.

OTHER MEDICAL TREATMENT:-

- Analgesic(no aspirin)
- Anti-ninflamatory medications
- Psychological counseling
- Good dental care
- Education- life long management

EDUCATION OF PATIENT & FAMILY:-

- Importance of early factor of replacement
- Use helmet when riding tricycle/bicycle
- Footwear

CONCLUSION:-

- Haemophilia disease is harmful in human
- It is very serious problem for human & infants
- Many treatment had been discovered for the disease
- Many girls are carrier of the disease, boys are highly affected by the disease.
- Doctor suggests controlling the bleeding. You also need treatment to precaution during childbirth.
- There is no proper treatment found for the disease, but some treatment is used for minimizing the disease. So it is a great gain for medical science.

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