Fabry Disease
(A Kind of Lysosomal Storage Disease)

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Fabry disease is an X-linked disorder. This condition is an inherited disorder that begins in childhood, causes signs and symptoms of many parts of body.

Characteristic clinical features of disease, include episodes of pain, particularly in the hands and feet (acropares thesia), small dark red spots (on the skin) called angiokeratomas, decreases ability to sweat (hypohidrosis, crnealopacity, disorders in Gastrointestinal tract, ear ringing (tinnitus), Hearing impairment and nephrologic problem.

Some problems potentially are life threatening; Nephrologic problems, cardiac disorders and stroke.

The Fabry disease is found in 1/117000 people or 1/40000 males. zAlso late-onset form (the milder dorm) is more common than the classic form.

This condition is caused by mutation in the GLA gene. This gene lead to make an enzyme called alpha-galactosidase A. This enzyme breaks down a fatty substance called globotriasylceramide. Mutation in GLA gene causes structural and functional disorder, globotriaosylcermide accumulates in blood vessels in the skin, cells in the kidney, heart and nervous system.

Absence of Alpha-galactosidose A activity lead absence of alpha-galactosidase and decrease of the enzyme activity causes the milder form of Fabry disease.

Some women are carrier of the disease and may show symptoms of Fabry disease.

These symptoms include; burning sensation in the hands that may increase in hot weather. Other Symptoms of Fabry disease include: Fever, decreased sweating, GI problems especially after eating.

Enzyme replacement therapy is used to treat patients, Faberazyme (agalsidase beta) decreases golobotria-asylceramide (GL-3). Approximately 1% of patients who treated by fabrazyme have experienced severe allergic reactions includes; Swelling of mouth, throat and face. Swelling disorder and low blood pressure are reported. Patients who have had allergic reaction during infusion treatment with anti fever and antihistamines before next infusion is recommended.

Keywords: Fabry disease; Childhood; Enzyme replacement therapy

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