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FABRY DISEASE

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Introduction:

Fabry Disease comes under hereditary familial diseases called lysosomal storage disorders. It is a genetic disease resulting from the accumulation of fatty substance in the cell, which is known as globotriaosylceramide. The accumulation of fatty substance, globotriaosylceramid, in the cells is due to the deficiency of enzyme α-galactosidase A. As a result of deficiency of α-galactosidase A, globotriaosylceramide accumulates in lysosomes, which in turn impairs the cell's ability to function properly.

In view of the above, accumulation of fatty substance in the cell and damage of tissues is the reason for significant symptoms of Fabry disease. One of the most visible symptoms of Fabry disease is reddish rashes over the body. Other symptoms of Fabry disease are non-specific such as pain, burning in hands and feet, fatigue and little sweating. Fabry disease also involves potentially life-threatening complications such as progressive kidney damage, heart attack, and stroke. Some affected individuals have milder forms of the disorder that appear later in life and affect only the heart or kidneys[2-4]. However an Australian study found that Fabry disease is rare and roughly 1 in 117,000 people.[1-3]

The irony of the inheritance of Fabry Disease is that, men with Fabry disease have a 100% chance of passing the altered gene to their daughters and 0% chance of passing it to their sons and a woman with Fabry disease passes the altered gene to her child (son or daughter) [3-4].

Fabry Disease can be easily diagnosed once the symptoms are identified. The confirmatory test

for diagnosis is genetic test. Further Fabry Disease can be diagnosed by enzyme assay that measures the galactosidase activity—prenatally in amniocytes or chorionic villi and postnatally in serum or WBCs[3-5].

One of the effective treatments for Fabry Disease is enzyme replacement with recombinant α-galactosidase A combined with supportive measures for fever and pain. In necessary cases, kidney transplantation is effective [5].

To conclude, Fabry disease is difficult to identify but easy to diagnose. Above all it is a multisystem progressive and hereditary genetic disease. Therefore a multi-disciplinary approach is required for the treatment of Fabry disease and replacing the deficient enzyme is one of the effective treatments of this disease. Last but not least, support and care from the loved ones are vital for its treatment.

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