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EC NEUROLOGY
Short Communication

Fabry Anderson Disease: Leukoencephalopathy

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Very rare X-linked disease definitely occurs in children, adults 18 - 50 years old and rarely with parents > 50 years old. The important cause is the mutation in the gene B-galactosidase.

An important lysosomal storage metabolism disease actually comes early on, but the discovery of the cause depends on brain mapping or WES (Whole Exome Sequences), MRI is also very important as it is mandatory for multiple sclerosis too different.

Also, important to remember as white matter lesions are known as leukoencephalopathies.

In fact, we find that such metabolic disorders, diseases, have various incomplete recessive X-linked, because women XX - as in men XY - have similar clinical symptoms, in the heterozygous case.

Accompanying symptoms

Angiokeratoma corporis diffusum, affects the kidneys, liver, skin, blood vessels and autonomic nervous system, acute abdomen pain, unexplained fever is also known, tingling sensations and various pains in the thighs and lower legs, especially the feet and hands, which are treated with pain reliever tablets such as gabapentins, Carbamazepine and Phenytoin are treated, dry eyes as seen with (Cornea Verticillata) in the SLIT LAMP and the complications usually come later, but with kidney and heart failure.

Stroke and TIAs are known these days because of high levels of cholesterol and sugar in the blood, relatively high levels of clotting in the blood.

Diagnose:

- GFR, S. Creatinine for Kidneys.
- ECG for heart.
- EEG for shingle and epilepsy.
- Non-contrast MRI scans to detect cerebral, small brain and brain stem blood disorders.
- Skin biopsy: because of keratoma/hyperkeratotic signs and skin allergy/rash.
- Blood Enzyme B galactosidase/trihexosylceramide B galactosidase activity. ***Very important because this enzyme deficiency in the blood affects the disease.
- Leukocytes in the blood.

Treatment:

- Kidney transplant.
- Pain reliever therapy.
- Complications treatment such as stroke in the stroke unit.

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