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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/14141
DOI URL: http://dx.doi.org/10.21474/IJAR01/14141

RESEARCH ARTICLE

ADRENOLEUKODYTROPHY IN MOROCCAN CHILD: CASE REPORT

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Manuscript Info

Manuscript History
Received: 30 November 2021
Final Accepted: 31 December 2021
Published: January 2022

Abstract

A boy of 10 years was admitted in our hospital for recurrent generalized seizures, spastic tetraparesis.A gradual impairment of vision and abnormal behaviors noticed for last 3 years. His family history find an uncle (mother' s brother) deceaded at the age of 6 years without a stated diagnosis, examination finding revealed generalized hyperpimentation more marked on lips and finger nails.Neurogical examination find spastic tetraparesis, deep tendon reflexes were brisk and pupils were roud and reacting to light.Fundoscopic examination note bilateral primary optic atrophy, his cardiovascular, respiratory and abdominal systems were clinically normal. All routine investigations revealed normal finding exception biochemical features (very high ACTH level, Low basal cortisol), of primary adrenal failure. MRI of the head showed bilateral white matter abnormalities in parieto-occipital regions. The diagnosis of adrenoleukodystrophy was strongly suggested from the medical history, biochemical and radiological findings of brain. The purpose of our report case is to highlight this rare disease to all, because its progress can be delayed with early diagnosis and it' s incidence can be reduced by genetic counseling.

Introduction:-

Adrénoleukodystrophy (ALD),is an X-linked neurodegenerative desorder associated with progressive demyelinisation of cerebral white matter and adrenal insufficiency this peroxisomal disorder is caused by a mutation on ABCD1 gene located on Xq28, and results in defect in peroxisomal beta-oxidation and accumulation of the very long chein saturated fatty acid in all body tissues.Ze are reporting this case dur to its rarity.

Case report:

A 10 years old boy, was admitted In our hospital for recurrent generalized seizures and spastic tetraparesis.a gradual impairement of vision and abnormal behaviors noticed for last 3 years.His past medical history was unremarkable.His family history find an uncle (mother's brother) deceased at the age of 6, without a stated diagnosis.

General examination revealed a normally nourished boy, with diffuse hyperpigmentation more marked over lips and finger nails.Neurogical examination find spastic tetraparesis,deep tendon reflexes were brisk and pupils zere roud and reacting to light.Fundoscopic examination note bilateral primary optic atrophy,his cardiovascular,respiratory and abdominal systems zere clinically normal.Result of routine blood count serum electrolytes,liver function tests,renal function tests and urine analysis were normal.Serum cortisol level was low and ACTH level was very

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high.Magnetic Resonance imaging brain revealed diffuse symmetrical abnormal MR signals involving bilateral deep cerebral white matter(mainly parieto-occipital Regions),MR signals are hypointense on T1,non enhancing and hyperintense on T2 / Flair.

The diagnosis of adrenoleukodystrophy was made from history,biochemical and radiological finding,Then the treatment was started with prednisolone and phenytoin with control of fits but rest of symptoms showed on improvement.

Discussion:-

Adrenoleukodystrophy is an X-linked inherited disorder with considerable phenotypic variations.The defective gene is located on the long arm og the X chromosome (Xq27-q28).The clinical course in adrenoleukodystrophy is characterized by behavioral disorder ataxia,decreased hearing,visual loss,epileptic seizures,followed by mental deterioration,psychosis and death.adrenal insufficiency is a usual finding, but does not always precede neurologic disease.The important laboratory finding are low serum sodium and chloride level,and elevated potassium levels reflecting the atrophy of the adrenal glands.the latter? Results in reduced excretion of corticosteroids,low serum cortisol levels and lack of rise in 17-hydroxy-ketoster-oid after ACTH stimulation.Demyelination begins bilaterally in the occipital region,extending across the splenium of the corpus callosum.Typically,the process spreads out ward and forward as a confluent lesion,affecting the parietal,temporal,and finally the frontal white matter,cerebellar white matter,cerebellar peduncles and corticospinal tracts.Plain MRI show hypointense signal on T1 and hyperintense signal on T2 and Flair images.Features of primary adrenal insufficiency should be measured.

No clear effective treatments are available,therapy is directed towqrds control of seizures with anticonvulsivants and glycocorticoid replacement therapy,dietary restrictions of very long chain fatty acidand addition of glycerol trioleate to the diet has lowered the plasma levels but had only a marginal clinical effect.

Early diagnosis brings the possibility of genetic counseling,carrier detection, antenatal diagnosis and thus to reduce the incidence of this serious disease.

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