X-ADRENOLEUKODYSTROPHY

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Relevance: X-linked adrenoleukodystrophy (X-ALD) belongs to peroxisomal disorders which damages the central or peripheral nervous system and/or the adrenal cortex. The pathogenesis is based on mutation in the ABCD1 gene, causing ineffective P-oxidation of fatty acids following a decrease in the activity of acetyl-CoA, leading to accumulation of very long chain fatty acids in tissues. Most commonly presents in males.

There are several phenotypes of disease: cerebral adrenoleukodystrophy (CALD), adrenomyeloneuropathy (AMN) and isolated adrenocortical insufficiency (IAI). CALD is characterized by progressive behavioral, cognitive, and neurologic deficits. Onset of symptoms ranges from childhood (3 to 11 years) to adolescence (12 to 18 years) and adulthood.

Purpose: To study the clinical features of X-ALD in children.

Objective: To describe the patient with severe childhood cerebral X-ALD, to analyze clinical criteria, the diagnosis and management.

Materials and methods. 7-year-old child, with a two-month history of decreased visual acuity, behavioral changes, learning deficits and social isolation which slowly manifested after catching a cold, sub-febrile temperature lasting several days. On neurological examination, he presented muscle spasticity, hyperreflexia, static ataxia, gait abnormality, speech delay, dysphagia. Hyperpigmented lesions on skin folds, hypotonia, weight loss suspects the primary adrenal insufficiency. Biochemical testing shows abnormally elevated very long- chain fatty acids (VLCFAs), high C-reactive protein, hyponatremia, low cortisol level.

The diagnosis was verified by MRI examination which showed bilateral white matter signal alteration in parieto-occipital regions. On molecular genetic testing detected ABCD1pathogenic gene.

Results: The patient received targeted therapy of hematopoietic stem cell transplantation (HSCT) that significantly reduced the progression of disease.

Conclusion: CALD causes progressive cerebral demyelination and adrenal insufficiency. Proper clinical approach, biochemical and genetic testing, MRI-findings are essential in diagnosis. HSCT is the most successful treatment option.

References:

1. Мубитдинова Х.Хамраева Г.Назаров, Б.Фатхуллаева , Д.& Муминова , Н. (2024).

Circadian' rhythm of total peripheral vascular resistance in acute renal failure in c hildren aged 7.1-18 years old. in Library, 7(1), 3-12. извлечено от https://inlibrary.uz/index.php/archive/article/view/36896

2. Хамраева, Г., Абдуллаев, Х., & Нуруллаева, Д. (2024). Оптимизация седации при малоинвазивных хирургических манипуляциях у детей с онкогематологическими заболеваниями . in Library, 2(2), 518-524. извлечено от https://inlibrary.uz/index.php/archive/article/view/36899
3. Акилов, Х., Хамраева, Г., Саитазизов, Х., & Чориев, Х. (2024). Surgical outcome of congenital heart defects with pulmonary hypertension in infants, in Library, 3(3), 8-13. извлечено от https://inlibrary.uz/index.php/archive/article/view/36902