A case study patient with the neuronal ceroid lipofuscinoses

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Introduction & Aim: The Neuronal Ceroid Lipofuscinoses (NCLs) are a group of related hereditary neurodegenerative disorders that occur at a frequency of between 2 and 4 in 100,000 live births. There is no effective treatment for NCL and all childhood forms are eventually fatal. Several forms of NCL are differentiated according to age of onset, pathology, and genetic linkage. The aim of this study was to describe the clinical and electroencephalographic findings of children with neuronal ceroid lipofuscinoses, respectively.

Case Report: A male patient of 14 years at the time was born with a body weight - 3400, height - 50 cm at birth by Caesarean section, during pregnancy, the mother had a mitral valve insufficiency. The child developed according to age till 4 years. He was complained to lack of walking, hand tremor, weakness in the lower limbs, Vinpocetine 5mg and Hopantenic acid 750 mg per day was administrated. At the age 5 years was complained to generalize myoclonic seizures by every hour and a slack paraparesis. MRI and EEG at the age 5 years revealed mixed hydrocephaly and disorganization of cortical rhythm, irritation of medium structures, damage to stem structures. Electromyography admitted degenerative changes with a decrease in bioelectric activity in the muscles of both limbs. Treatment scheme was Valproic acid 500mg per day, however progressed delay mental and psychomotor development, increasing paroxysmal myoclonic seizures. Phenotypic changes developed such as severe deformity of the skeleton, the underdevelopment of the middle third skull, while forming of trema and diastema (Fig. 1), cachexia. In the following years, he exhibited a progressive loss of expressive language and only can follow commands to smile or look in all specific directions and became bed-dependent, also present amaurosis. Multiple pathology of internal organs is also noted (Tab.1). thrombocytopenia developed (PLT 30*109/l) (Tab.2) due to VPA 800 mg per day

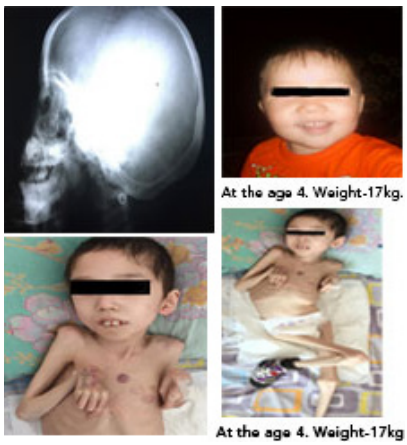

Figure 1: Phenotypic changes developed such as severe deformity of the skeleton, scoliosis, cachexia

Figure 2: X-ray of skull at the age 5, trema and diastema of teeth.

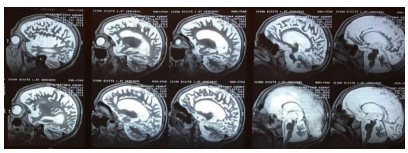

Figure 3: The MRI at the age 10 years

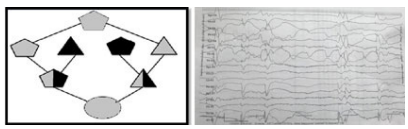

Figure 4: The genealogical tree of presented patient.
Figure 5: EEG during the morning sleep

Results: Karyotype analysis report a single specimen has not detected any abnormalities in structure, karyotype 46,XY. The test based on Clinical Medical Institute of Urumqi General Hospital, Landzhou Command, PLA. In 2017 MRI demonstrated residual organic encephalopathy with atrophy of the large brain, gray matter, which is characteristic for NCL. Based on genealogical tree (Fig.4), was admitted a consanguineous marriage (first-degree cousins).

Conclusion: Based on MRI, phenotypical changes and genealogical tree of this patient, enzymatic and genetic studies should be performed since accurate enzymatic or molecular diagnosis will enable treatment and family counseling. Better awareness of the disease will facilitate timely strategies for the specific management of NCL disease will decrease a high mortality rate. Disease-specific management, genetic counseling, and new therapies in development for NCL disease make early and accurate diagnosis of this severe neurodegenerative disease essential.

Biography

Baurzhan Orazaliev is currently pursuing his Pediatric Intern at Karaganda Medical University. He has published 9 articles and participated to discrepancy international student conferences.