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Atypical Presentation of Celiac Disease in a Girl with Epilepsy and Retinopathy – Albanian Case

Aferdita Tako[1], Mira Dervishi[1], Sonila Tomori[1], Aida Bushati[1, 2], Paskal Cullufi[1, 2]

[1]Department of Pediatrics, University Hospital Center "Mother Teresa", Tirana, Albania

[2]Department of Pediatrics, University of Medical Sciences, Tirana, Albania

Email address:

mira02bet@yahoo.com (A. Tako)

Abstract:

Aims: To report a case with celiac disease in a girl presented with epilepsy, behavioural disorder and retinal dystrophy.
Method: E.B. was hospitalised at age 11 with convulsive seizure (the third in a year) and irritable mood, hyperactivity, school refusal and eating problems. She was born in term with APGAR score 9/10 and weight 2800 gr. Her infancy and early childhood passed normally besides being a picky eater, not an easy child to obey and sleeping difficulties. When she got 8 years started to have decreased in vision, sleep disturbances, irritable mood and an iron deficiency anaemia was present. A year later she suffered night blindness and was diagnosed with conic retinal dystrophy. The molecular biology test resulted negative. At age 10 she had her first convulsive seizure early in morning as left hemi convulsion generalised secondarily. Her sleep patterns were dominated by sleepwalking episodes mostly around 2 hours after falling asleep associated with fear and psycho-motor agitation. She started to have nightmares and inability to remember these episodes. MRI scan without contrast resulted normal and she started treatment with carbamazepine 400 mg/daily. Five months later she had her second convulsive seizure with the same convulsive pattern: starting as hemi convulsion and afterwards generalised secondarily. EEG resulted in diffuse slow waves at fronto-temporal region. Gastrointestinal symptoms were constipation and abdominal discomfort.
Results and Discussion: Laboratory tests showed the following:

IgA anti-tTG ELISA	95.7 UI/ml
Intestine biopsy	villous atrophy
Endomysial antibodies IgA (EMA)	positive (1:20) - high
Complement factors C3; C4	C3=76.5mg/dl [79-152]; C4=14.5 mg/dl [16-38]
Functional thyroid tests TSH, FT4	normal
Anti TPO (anti peroxidase antibodies)	68.1U/ml (slitely elevated >75 = positive)
25-Oh-D3	11.48 ng/ml (very low)
Retinole	in the lowest normal range
folic acid and B12, B6 vitamine	Normal
Proteine electrophoresis Albumine	39.8 % [56-68]
Proteine electrophoresis gamaglobuline	33.0% [9-18]
P.electroph. alfa 1,2 & beta globuline	normal

It was confirmed celiac disease and she started a gluten free diet and continued anti-epileptic medication switched to valproic acid. Immunoglobulin 100% IgG (Intratec) I/V was given during hospitalisation 2gr/kilo/day in 5 consecutively days.
Conclusion: Celiac disease has intestinal and extra-intestinal manifestations, caused by immune alterations or by nutrient malabsorption. The ophthalmic symptoms are rare within the extra intestinal manifestations, but should be investigated in patients with celiac disease and taken into consideration as the first systemic manifestations of the disease. Epilepsy, vitamin D&A deficiency, ophthalmologic manifestation in celiac disease are strongly correlated with an autoimmune pathway. Important for treatment consideration are gluten free diet, oral retinol and vitamin D daily, antiepileptic drugs and immunotherapy.

Keywords

Celiac Disease, Retinal Dystrophy, Behavioural Disorder, Epilepsy, Autoimmune Pathway