FM Publisher
Frontiers Journal of Case Reports and Images (FJCRI)
Volume 01, Issue 03, Article ID FJCRI-2019-12

Case Report *Open Access*

HEREDITARY MUTATION OF SLC6A5 GENE INDUCES HYPEREKPLEXIA (STARTLE DISEASE) IN TWO SISTERS: A TREATABLE NEUROPEDIATRIC DISEASE

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Citation: Bittmann, S., Luchter, E., Villalon, G., Weissenstein, A., & Moschüring-Alieva, E., (2019). China Restaurant Syndrome: An Unusual Neurological Experience Due To Glutamate. *Frontiers Journal of Case Reports and Images*, 1(3), 1-2.

Received Date: *05 October 2019;* ***Accepted Date:*** *15 October 2019;* ***Published Date:*** *20 October 2019.*

ABSTRACT

Hyperekplexia is primarily an autosomal dominant disease characterized by exaggerated startle reflex and neonatal hypertonia. If untreated it can be associated with sudden infant death from apnea or aspiration pneumonia and serious injuries. Different mutations of the 1-subunit of inhbitory glyzine receptor (GLRA1) could be found. Moreover mutations in the genes GLRB; GPHN and in our case of two sisters SLC6A5 were found. Clonazepame, a gammaamino- butyric acid (GABA) receptor agonist is the therapy of choice. An early diagnosis will lead to appropriate treatment and genetic counseling.

Keywords: *Hyperekplexia, Clonazepame*

CASE REPORT

"We report about a family with 5 children. The father is 43 years old, the mother 37 years. The family arrived from syria to Germany in 2015 and lived in Germany since one year as diagnostic treatment initiates. The mother described rhythmic intrauterine movements in the 4th months of pregnancy. These movements were also found in the 13 years old sister. N., the 5 years old girl, had seizures from 4 months of life. The time of the seizures lasted in the beginning 1 minute up to 2 hours. In syria, phenobarbital, valproate and levitiracetam were given. Further treatment in Germany consisted of 200 mg carbamazepine in the morning and the evening in an ambulatory setting. Semniologic analysis of the seizures revealed tonic-clonic form, lateral tongue bites, open eyes, variable time, about 2 minutes time. EEG analysis in Germany 2016 revealed a normal sleeping EEG, no epileptic potentials. Further EEG analysis later were without pathological signs. A vitamin B6 treatment was performed (3x140 mg daily). The clinical neurological evaluation revealed a global hyperreflexia without pyramidal signs. Molecular analysis showed a homozygote stopp variant in SLC6A5 gene. The other sister, A., 13 years-old, a good student at school, do not attend school sports because of fear to fall down suddenly, fear to hear loud sounds and voices. She had an episode of loss of bonus in moments of loud sounds and voices. She receives monotherapy of valproate. EEG analysis showed right sided occipital focal focus with spike wave activity without photo sensibility. Molecular analysis revealed a familiar homozygotic variante in SLC6A5 gene.

DISCUSSION

Hyperekplexia is based on muscular hypertonia and overwhelming reactions to tactical stimuli and defined as neurogenetic pediatric disease. Kirsten and Silfverskiold described the disease as, emotionally precipitated drop seizures (Kirstein et al., 1958). Newborns with hyperekplexia show diffuse muscular hypertonia, Hyperreflexia and startle reactions to sounds and voices. Startle reactions can be induced by tipping on labella region. Hypertonia and hyperreflexia relieves at the end of 1 year of life. Motor development is delayed to to fear for fall downs. Cases in literature are described, where children walk on knees not to fall down suddenly. 1992 the autosomal dominated hyperekplexia gene was found in linkage study on the long arm of chromosome 5 (5q33-35) (Baker et al., 1994; Ryan et al., 1922). In both children, a homozygote mutation of the SLC6A5 gene was found. This result is phenotypically the proof for hyperexplexia. SLC 6A gene codes for presynaptic glycol transporter. The gene influences the extracellular glycol concentration in neurotransmission situation. In hereditary hyperekplexia, symptoms were found in early newborn period with seizures with hypertonus of the muscles. In both children, the pathological homozygote variante of c.861 C A; p. Tyr287 in SLC6A5 gene were found. This mutation has consequences for both children: fear of sport activation due to to fall down in activity; fear of loud noises and therefore loss of muscle tones. Benzodiazepine treatment is the treatment of choice in this serious condition of single gene mutation.

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