Dose Positive CHD8 have a Role in Ketogenic Diet Response in Autism: A Case Re- port

Amal AlQassmi[1*], Maha AlOtiabi[2] and Fahad AlRabeeah[3]

[1]Department of Pediatric Neurology and Epilepsy, King Saud Medical City, Riyadh, Saudi Arabia

Introduction:

Autism is a brain disorder that limits a person's ability to commu- nicate and relate to other people. It first appears in young children, who fall along a spectrum from mild to severe. Some people can nav- igate their world, some have exceptional abilities, while others strug- gle to speak [1,2]. Autism Spectrum Disorders (ASDs) affect about one child in 68, striking nearly five times as many boys as girls.
Before a child turns three, careful observers can see signs of au- tism [1]. Some children develop normally until 18-24 months old and then stop or lose skills. Signs of an ASD can include: Repeated mo- tions (rocking or spinning), avoiding eye contact or physical touch, delays in learning to talk, repeating words or phrases (echolalia), get- ting upset by minor changes. It's important to note that these signs can occur in children without ASDs, too [3].

The use of the ketogenic diet to treat epilepsy dates as far back as Hippocrates time. The major way to induce ketosis at that time was using fasting or starvation. Since then, modern reports put the use of the KD in treating childhood epilepsy starting in the 1920's

Ketogenic diet had been tried in treating Autism, but no clear hy- pothesis or mechanism of the Diet in Autistic spectrum.

Aim:

This study tests the effects of ketogenic diet feeding in Autism and their behavior, a child with autism spectrum disorder.

Case Report

We present a case of 7 and 6 months old Saudi boy who present- ed to neurology with history of speech delayed and behavior issue that had been concern by parents since age of 2 years they did seek medical advice, but no clear or major help was provided by general physician till age of 4 years where he had been referred to neurology for assessment.

After we did full history and examination which was normal apart from social developmental, speech delayed. Blood work up (CBC and chemistry) was normal, bone profile and renal function was normal. Metabolic screening (serum amino acid, urine organic acid, quanti- tative amino acid, CDT) all was normal. MRI (Magnatic Resonance Imaging) brain normal, EEG (Electro Encephalogram) was normal, BEAR (Hearing Test) was done normal, patient seen by psychiatry and the Autism Rating Scale score assessment was diagnosed as au- tism scored 49, IQ assessment was 85%, SNP array was not signifi- cant, patient was diagnosed with Autism , whole exome came positive for CHD8:NM_001170629:ex- on26:c.4984C>T:p.R1662X heterozygous (autosomal dominant).



Features of Ketogenic Diet Plan

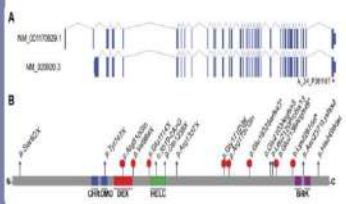


Figure 1
Spectrum of *CHD8* mutations in autism spectrum disorder

Discussion:

Autism Spectrum Disorders (ASD) include a complex neuro de- velopmental condition characterized by abnormal social interaction, verbal and non-verbal communication [1]. Limited scientific advanc- es have been made regarding the causes of ASD, with general agree- ment that both genetic and environmental factors contribute to this disorder . ASD has been associated to metabolic dysfunction

n our patient, it had CHD8 gene mutation, in spite different gene mutations had been discovered in Autism but none of them had dif- ferent presentation or describe the severity of behavior changes in Autistic child and non-had addressed the use of ketogenic diet in this different type of mutations.

In a case study of a child with autism and epilepsy, following stan- dard treatment non-response, the individual was placed on KD (1.5:1 lipid : non-lipid ratio) with adjunct anticonvulsant therapy [22]. The patient was in ketosis. After initiation of the diet several benefits en- sued including the resolution of morbid obesity and the improvement of cognitive and behavioural features of the disorder. After several years on the diet, the patient's score on the Childhood Autism Rating Scale decreased from 49 to 17, a change from a rating of severe au- tism to non-autistic, and IQ increased by 70 points. Fourteen months following the initiation of the diet the patient was also seizure free.

Although the exact molecular mechanisms underlying the effect of the KGD are still under investigation, several scenarios are reported below to explore the potential therapeutic effects of the KGD in ASD.

Conclusion:

Positive gene mutation of autism may be an important factors in showing the response of high lipid diet in those children.

Corresponding author: Amal AlQassmi, Department of Pediatric Neurology and Epilepsy, King Saud Medical City, Riyadh, Saudi Arabia, Tel: +966 570006241; E-mail: dr_alqasmi@yahoo.com

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