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Genetic insights into progressive myoclonic epilepsies: A case study of *KCTD7* mutation in an Iranian-Azeri-Turkish family

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ABSTRACT

Progressive Myoclonic Epilepsies (PMEs) are a rare and heterogeneous group of epileptic disorders often with progressive neurologic deterioration. The intensity of the clinical features varies depending on the underlying genetic etiology. This study aims to identify the genetic mutation associated with PME in a family belonging to the Iranian-Azeri-Turkish ethnic population. A 5-year-old boy and his 8-year-old sister, presenting with PME-related electroclinical features such as myoclonic seizures and progressive cognitive and motor decline, underwent comprehensive clinical evaluations, including pedigree analysis, laboratory tests, and EEG assessments, followed by Whole-Exome Sequencing (WES) to identify potential disease-causing mutations. We identified a novel homozygous mutation (c.14C > T) in the *KCTD7* gene in both siblings, confirmed through Sanger sequencing. This mutation was not observed in a cohort of 430 healthy individuals from the same Iranian-Azeri-Turkish ethnic background, providing strong evidence for its pathogenic role. This finding advances our understanding of the genetic basis and phenotypic diversity of PMEs, but further research is needed to elucidate how *KCTD7* mutations contribute to epilepsy and neurodegeneration.

1. Introduction

The Progressive Myoclonic Epilepsies (PMEs) are rare and caused by diverse group of underlying genetic etiologies. PMEs are characterized by drug-resistant epilepsy, mainly myoclonic seizures, abnormal electroencephalographic findings, cerebellar signs such as ataxia and tremor, and progressive psychomotor deterioration which appear in an individual with prior normal development and cognition. The age of onset, clinical features, course of the disease, and prognosis are highly dependent on the underlying etiology. There may be a family history, often with an autosomal recessive pattern; however, PME can also be sporadic [1].

Several specific subtypes of PME have been identified, including Unverricht-Lundborg disease (Progressive Myoclonic Epilepsy Type 1), Neuronal Ceroid Lipofuscinoses (NCL), Lafora disease Myoclonic, Sialidosis, and Myoclonic Epilepsy with Ragged Red Fibers (MERRF syndrome). The majority of these subtypes are inherited in an autosomal recessive pattern, with various genetic mutations linked to each form [2]. For instance, mutations in the *CSTB* gene cause Unverricht-Lundborg disease, the most common type of PME worldwide, while *EPM2A* mutations are linked to Lafora disease [3,4].

Identifying the genetic cause is essential for distinguishing PME subtypes and guiding treatment, yet the etiology remains unknown for many patients, underscoring the need for continued research in this field.

In this study, we present a novel homozygous mutation **(NC_000007.14 (NM_153033.5): c.14C > T)** in the *KCTD7* gene, discovered through whole-exome sequencing (WES), in a 5-year-old boy originating from Iranian-Azeri-Turkish ethnic group and his 8-year-old sister, both exhibiting the same clinical manifestations. Both siblings, from a non-consanguineous family, display clinical features consistent with Progressive Myoclonic Epilepsy, contributing to the broader understanding of the genetic landscape and phenotypic variability associated with PMEs.

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2. Materials & methods

2.1. Case analysis

A 5-year-old boy from a family with a non-consanguineous marriage was referred to the Division of Pediatric Neurology at Mardani Azari Children's Hospital in East-Azerbaijan Province (IRAN) due to drug-resistant epilepsy and neurodevelopmental regression. His sister, who is three years older, exhibited the same clinical features, while both parents appeared healthy with no signs of illness. To diagnose the condition, we conducted pedigree analysis, clinical assessment, laboratory data analysis, and long-term Video EEG monitoring. Written informed consent was collected from all participants or their guardians. The study received approval from The Ethics Committee of the University of Tabriz.

2.2. Genetic analysis

2.2.1. Whole exome sequencing

The patient was referred to the genetic laboratory for WES analysis to identify mutations responsible for the disease in his genome. Informed consent was acquired from the family before collecting blood samples. EDTA tubes were utilized for the collection of blood samples from the proband as well as his parents and affected sister. Genomic DNA was extracted from peripheral blood, quantified, and then underwent WES analysis by applying the Agilent SureSelect Human All Exon V7 kit for Target Enrichment [5,6].

Sequencing of the enriched library was performed on the Illumina NovaSeq 6000 platform, producing Sequence reads of 150 base pairs (bp) that were aligned to the UCSC human reference genome (GRCh38/hg38 and GRCh37/hg19) using CLC Genomics Workbench software (version 21) and the Burrows-Wheeler Aligner (BWA) [7]. The use of Picard and Trimmomatics V0.39 allowed for the filtering out of Reads with low quality scores and duplicates (Qbase $<$ 20) to maintain high data quality [8]. Sorting and indexing of BAM files were performed using Samtools [6,9]. Variant calling for single nucleotide variants (SNVs) and small insertions or deletions (Indels) was performed using GATK version 4.2.0.0 and DeepVariant version 1.1.0 [10]. Following variant calling, the wANNOVAR web tool was employed to annotate the variants [11]. An in-house variant filtering pipeline was used to detect variants responsible for the disease, adhering to the guidelines from the Sherloc criteria and the American College of Medical Genetics and Genomics (ACMG) [12]. The filtration process excluded synonymous variants and those located in deep intronic regions, as well as variants in upstream, downstream, and ncRNA regions, to eliminate polymorphic variants. The regions surrounding the exons, however, were retained, as they could harbor variants with potential functional impact and key regulatory elements. Subsequently, variants with a frequency exceeding 0.05 in diverse populations were omitted from the analysis, as they are regarded as polymorphic. The VCF file was submitted to the CADD database for further analysis [13]. The relevant filtration was applied with a Phred score of $\geq$ 10, and the data was subsequently retrieved from the CADD database to finalize the filtration using the MutationTaster database [14]. In the next step, by using the Online Mendelian Inheritance in Man (OMIM) database, genes potentially associated with the disease symptoms were carefully chosen from the list generated by the MutationTaster database for further assessment [15]. Web-based tools and resources, including VarSome were used to interpret and screen variants for their potential to cause disease [16]. After identifying the disease-causing variant associated with the proband's clinical features, Primers were designed to target the specific segment of the genome sequence that contained the pathogenic variants. After designing the primers, Sanger sequencing was performed for the patient, his sister, and their parents. Additionally, the secondary structure of RNA transcribed from the chosen alleles and the impact of the identified variant on the structure and function of the proteins encoded by these alleles was analyzed utilizing online tools [17,18].

2.2.2. Segregation analysis

Sanger sequencing was performed on the proband, his affected sibling, and their parents to confirm the likely pathogenic variant and provide additional evidence of its association with the disease. The ABI 3500 Genetic Analyzer, manufactured by Applied Biosystems, was utilized for Sanger sequencing. For this purpose, primers were specifically designed to amplify Target sequence for Sanger sequencing, enabling precise analysis of the variant. The primer sequences were as listed: forward, 5′-GCCAGAGGACCAATCAGTGC-3′ and reverse, 5′-GTCGTCTTCGGCGTCAGAG-3′. During validation step, DNA samples from the proband, his sister, and parents were amplified via PCR using specifically designed primers, with Taq DNA polymerase and an annealing temperature of 61 °C, followed by Sanger sequencing of the PCR products to confirm the presence of the variant.

2.2.3. Population study

A cohort of 430 healthy individuals from the Iranian-Azeri-Turkish ethnic background was examined for the presence of the variant through analysis of whole-exome sequencing (WES) data. This comprehensive screening serves as a control group to assess the variant's frequency in the general population, providing valuable context for its potential pathogenicity and its role in disease causation within this specific ethnic group.

3. Results

3.1. Case history

The case was a 5-year-old, right-handed male from a non-consanguineous family of Iranian-Azeri-Turkish descent. He had normal birth history and typical development until the age 2 years. At that time, he began experiencing brief myoclonic seizures triggered by fevers. He was treated by sodium valproate and clobazam, achieving acceptable seizure control initially. However, over the following months and years, the frequency of seizures progressively increased. Other antiseizure medications, including ethosuximide, topiramate, and levetiracetam, were tried but did not significantly reduce seizure frequency.

His habitual seizures were myoclonic, but he also experienced sporadic generalized tonic clonic seizure. Treatment with prednisolone and IVIG provided only temporary improvement in seizure frequency. His neurodevelopmental status has been progressively deteriorating. Neurological examination revealed ataxic gait, dysarthria, and dysmetria. The myoclonus poses challenges with certain movements, such as standing up from a sitting position or changing direction while walking, which often results in falls to the left side. Ophthalmic examination was normal. Although speech was significantly affected, cognitive abilities remained acceptable. Muscle strength was normal, with intact deep tendon reflexes (DTR).

Family history is notable; his 8-year-old sister exhibits a similar, though milder, clinical course. Her seizures started at the age of 2 years as febrile generalized tonic-clonic seizures. She later developed afebrile seizures, including generalized tonic-clonic and myoclonic seizures. Currently, her habitual seizures are myoclonic in nature. She is on a regimen of sodium valproate, ethosuximide and clobazam, which partially control her myoclonic seizure. Despite this, she is able to perform daily independent activities such as walking, feeding, and attending school. However, her school performance is poor, likely due to hyperactivity and attention deficits.

The parents report that in the morning on awakening, her action tremor and myoclonus worsen with voluntary movements, interfering with fine motor skills. Neurological examinations revealed a mildly ataxic gait accompanied by action tremor and myoclonus. (Her medical evaluation was conducted outside of our center).

3.2. Laboratory evaluation

Metabolic studies, including lactate, ammonia, pyruvate, and metabolic screening, were within normal limits. Liver function tests, complete blood count (CBC), cholesterol, triglyceride, and thyroid function tests (TFT) were within normal limits as well. Standard electrodiagnostic studies, including nerve conduction velocity (NCV) and electromyography (EMG), were within normal limits. A brain MRI performed at age 3 years was reported as normal. Visual evoked potential (VEP) and auditory brainstem response (ABR) tests were also within normal limits. Long-term video EEG monitoring revealed multifocal erratic myoclonic seizures that were time-locked with generalized spike and polyspike waves. The interictal EEG showed a slow background, poorly organized sleep features, and frequent generalized spike and polyspike waves.

3.3. WES data

The proband was sent to the genetic laboratory for further genetic evaluation using WES. A homozygous transition from C to T at position 14 (c.14C > T) (p.Thr5Met) in exon 1 of the *KCTD7* gene has been identified (Fig. 1). Sanger sequencing for the proband, his sister, and parents was performed for further evaluation (Fig. 1). The findings indicated that the parents' genotypes were heterozygous for the mutation, while the patient and his affected sister were homozygous for **NC_000007.14 (NM_153033.5):c.14C > T** variant, confirming the autosomal recessive pattern of the disorder.

3.4. Bioinformatics analysis

A variety of bioinformatics tools and databases were applied to predict the functional impact of the genetic variant, assess its pathogenicity, and interpret its potential effects on protein structure. To predict the pathogenicity of the variant, the following tools were used: CADD (Combined Annotation-Dependent Depletion) score (Phred Score = 22.8), InterVar (classified as uncertain significance, located in a mutational hotspot or critical and well-established functional domain, and absent from controls), and Franklin by Genoox (classified as a variant of uncertain significance, VUS).

Analysis using the RNAsnp web-based tool indicated that the mutation is unlikely to significantly alter the local RNA secondary structure, as evidenced by a non-significant p-value of 0.2418. Additionally, I-TASSER predictions revealed minimal structural differences between

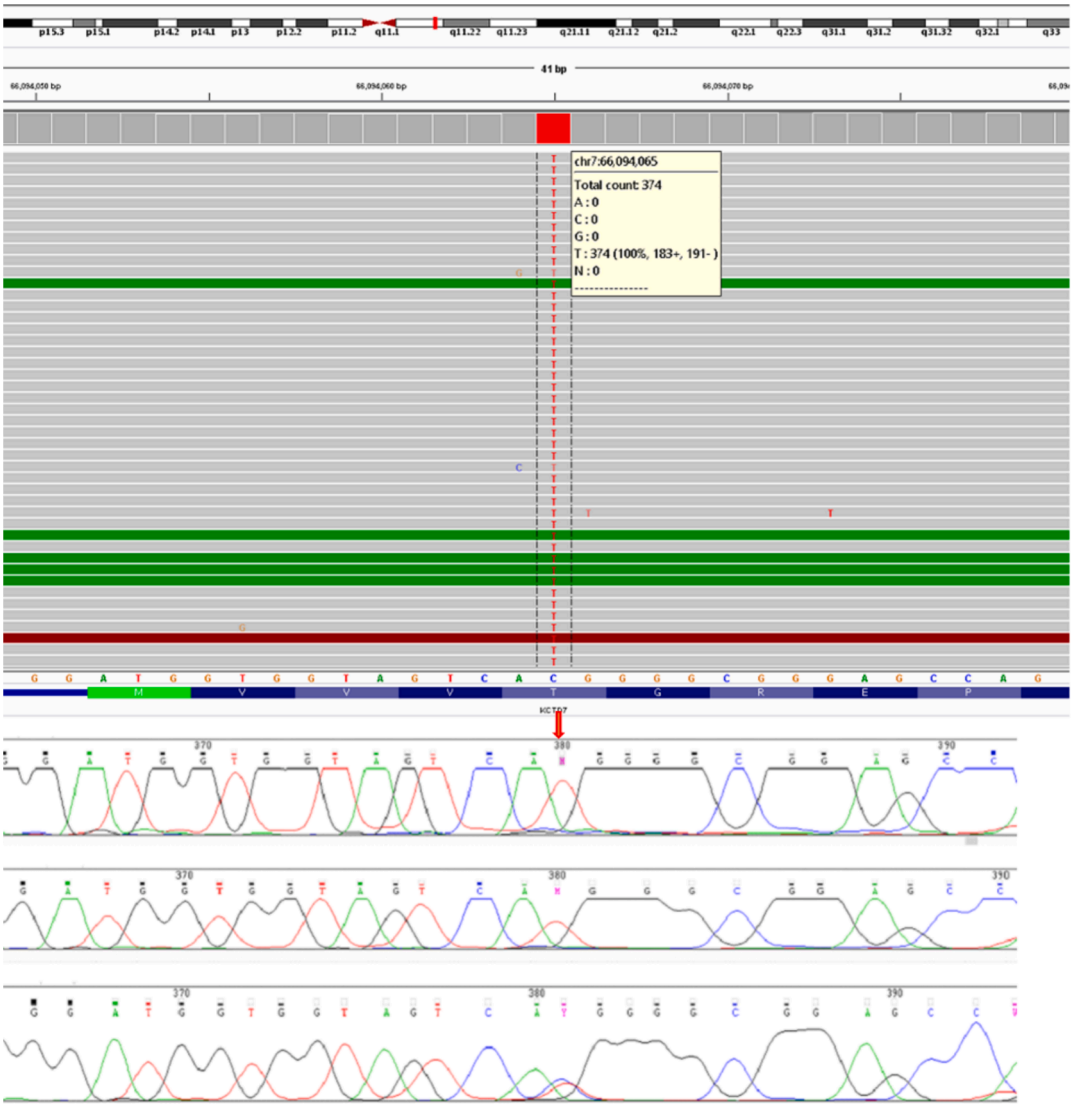

Fig. 1. Whole exome sequencing and Sanger sequencing results in the proband and his affected sister and parents. The pathogenic variant in the KCTD7 gene (c.14C > T) is illustrated in the IGV figure of the BAM file of the proband (upper figure). This potentially pathogenic variant is shown in a homozygous state in the proband and his affected sister (upper graphs) and heterozygous state in his parents (lower graph).

the wild-type and mutant proteins, with conserved secondary structure elements and similar RMSD, TM-scores, and C-scores. These results suggest that the overall protein fold and modeling quality remain largely unaffected.

3.5. Population screening

This variant was not found in 430 healthy individuals from the Iranian-Azeri-Turkish ethnic group or in public population databases such as the 1000 Genomes Project and gnomAD, strongly supporting its pathogenicity.

The ACMG guidelines for interpretation of sequence variants is being used to determine pathogenicity of this novel variant. The absence of the mutation in our cohort and in published public datasets (PS4), combined with the results of segregation analysis showing co-segregation with the disorder in multiple affected family individuals (PP1), and the highly specific phenotypes of patients with *KCTD7*-related diseases (PP4) suggest that the identified variant is most likely pathogenic.

4. Discussion

PMEs are linked to pathogenic variants in multiple genes, including *CSTB*, *NHLRC1*, *EPM2A*, *NEU1*, *MT-TK*, *KCNC1*, *GOSR2*, *SCARB2*, *PRICKLE1*, and the *KCTD7* gene on chromosome 7q11 [19]. Epilepsy, Progressive Myoclonic 3 (PME3), with or without intracellular inclusions is a rare genetic form of PME and is caused by homozygous or compound heterozygous mutations in the *KCTD7* gene (potassium channel tetramerization domain containing protein 7), following an autosomal recessive inheritance pattern [20]. Broad expression of the *KCTD7* gene is observed across various regions of the brain, particularly in the hippocampus, cerebellum, and corticospinal tracts and the protein encoded by this gene belongs to the potassium channel tetramerization domain-containing family. Impaired KCTD7 function is linked to a depolarized resting membrane potential and elevated excitability, which can cause epileptic seizures [19]. The *KCTD7* gene has been linked to progressive myoclonic epilepsy (PME) through various homozygous mutations, identified in consanguineous and non-consanguineous families across diverse ethnic backgrounds, consistently presenting with myoclonic seizures, neurological decline, and developmental delays [20,21,22,23,24,25]. In this study, we report a novel homozygous mutation **NC_000007.14 (NM_153033.5): c.14C > T** in *KCTD7* in a 5-year-old Iranian-Azeri-Turkish boy and his 8-year-old sister from a non-consanguineous family. Bioinformatics analysis revealed that the secondary structure of the transcribed mutant RNA and the structure of the encoded protein exhibited no notable differences compared to the wild type. However, the observed phenotype indicates that the reported mutation likely leads to protein dysfunction, which contributes to the disease. The proband and his sister exhibit a phenotype characterized by early-onset myoclonic seizures and developmental regression following seizure onset. Their clinical presentation aligns with the definition of PME, as both experienced an initial phase of normal development followed by neurological decline and multifocal myoclonus as the predominant type of epileptic seizure. Most PMEs are autosomal recessive diseases, consistent with the cases presented. The age of onset for PME typically ranges from five months to three years; in this study, seizure onset in both siblings was observed at two years of age, aligning with findings reported by Van Bogaert et al., Kousi et al., and Farhan et al [20,22,24]. PMEs are often drug-resistant and have poor responses to treatment. The daughter in the studied family did not respond to any prescribed antiepileptic medications. However, the son experienced a reduction in seizure activity after receiving Prednisolone and IVIG treatment. Consistent with our findings, a previous report by Sai Chandar Dudipala et al. in 2021 noted a decrease in seizure frequency in an affected Indian girl after receiving prednisolone [25]. In all patients, EEGs showed abnormalities, predominantly characterized by highly frequent generalized and/or multifocal spike and wave discharges. The case's EEG results, which showed frequent epileptiform discharges, were consistent with this. Brain MRI findings in cases of PME lack specific diagnostic patterns and may appear normal at the onset of the disease, However, as the disease advances, cerebral or cerebellar atrophy and white matter changes may become more evident. In the present cases, brain MRIs were reported as normal.

The highly specific phenotypes observed in both patients with *KCTD7*-related diseases (PP4), combined with segregation analysis indicating co-segregation with the disorder in several affected family individuals (PP1), the identification of a homozygous state in both affected individuals (PM3), and the absence of the mutation in our control group and in published public datasets (PS4), strongly suggest that the identified variant is likely pathogenic. However, a limitation of this study is the need for functional validation of our findings. Future functional studies using organoids and biochemical approaches like immunoprecipitation could elucidate the impact of mutations on neuronal development and protein interactions compared to wild-type conditions.

4.1. Conclusion

In this study, we report a novel likely pathogenic variant (**NC_000007.14 (NM_153033.5): c.14C > T**) of the *KCTD7* gene in two siblings presenting with progressive myoclonic seizures, cognitive regression and global developmental delay, whose parents were heterozygous carriers of this variant. The identification of this likely pathogenic variant via WES provides a likely genetic diagnosis, informs therapeutic strategies for affected families, and advances understanding of the disease's genetic basis.

Ethical Statement: This project was ethically approved by the Tabriz University of Medical Sciences (IR.TBZMED.REC.1403.879) and written formal consent was obtained from the parent/guardian.

CRediT authorship contribution statement

Haneieh Honarmand: Writing – review & editing, Writing – original draft, Investigation, Data curation, Conceptualization. **Mortaza Bonyadi:** Writing – original draft, Validation, Supervision, Conceptualization. **Mohammad Barzegar:** Writing – review & editing, Project administration, Conceptualization.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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