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Diagnosis and follow-up of a PCDH19 epilepsy patient

Yanzhao Chen[a,*], Yaming Xia[b,*], Lipeng Chen[c], Zhiping Liu[a], Bo Li[a], Keying Zhou[a,*] and Yongjian Yue[c]

Objective **Developmental and epileptic encephalopathy 9 (DEE9) is an X-linked genetic disorder characterized by the onset of seizures during infancy. Mutations in protocadherin 19 (*PCDH19*) are the main cause of DEE9. Our study aims to demonstrate the diagnostic process and long-term follow-up of a female pediatric case presenting with recurrent seizures.**

Methods **In the present study, a female child presented with recurrent epileptic seizures and findings of abnormal synchronous discharges on electroencephalograms. Whole exome sequencing (WES) was performed on the proband and her parents to identify potential genetic variants.**

Results **A heterozygous variant (NM_001105243: c.695A>G) in *PCDH19* was identified and validated using Sanger sequencing. Based on clinical features and genetic analyses, the patient was diagnosed with *PCDH19*-female limited epilepsy. Furthermore, a 4-year follow-up was conducted to assess the impact of the pathogenic variant on phenotype and treatment outcomes. The patient exhibited normal intelligence, which differed with the clinical features reported in other studies involving the same variant.**

Conclusion **WES confirmed the diagnosis of DEE9, and subsequent follow-up highlighted the effectiveness of the treatment. Therefore, genetic testing can improve the diagnosis of DEE9, particularly in cases with atypical symptoms, and provide valuable insights for genetic counseling and clinical treatment strategies. *Psychiatr Genet* 36: 26–31 Copyright © 2025 The Author(s). Published by Wolters Kluwer Health, Inc.**

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[a]Department of Pediatrics, [b]Department of Clinical Laboratory, Longhua Branch and [c]Department of Pulmonary and Critical Care Medicine, Shenzhen Institute of Respiratory Disease, Shenzhen People's Hospital (The First Affiliated Hospital of Southern University of Science and Technology, The Second Clinical Medical College, Jinan University), Shenzhen, Guangdong, China

Correspondence to Yongjian Yue, PhD, Department of Pulmonary and Critical Care Medicine, Shenzhen Institute of Respiratory Disease, Shenzhen People's Hospital, No. 1017 Dongmen North Road, Luohu District, Shenzhen 518020, China
Tel: +86 15919909356; e-mail: yueyongj@163.com

*Yanzhao Chen, Yaming Xia, and Keying Zhou contributed equally to the writing of this article.

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Introduction

Epilepsy is a recurrent chronic neurological disorder that is caused by sudden abnormal electrical discharge of brain neurons, resulting in short-term brain dysfunction. It affects approximately 10 million people in China (Ding *et al*., 2021). The prevalence of epilepsy in China has increased from 1990 to 2019 (Song *et al*., 2017; Liu *et al*., 2023). Improved diagnostic methods have led to the identification of an increased number of epilepsy cases. Nearly 30% patients with epilepsy exhibit resistance to antiepileptic drugs (Kavehei *et al*., 2019; Zhao *et al*., 2020). The main cause of developmental epileptic and encephalopathy is variability in the sodium voltage-gated channel alpha subunit 1 gene (*SCN1A*), and mutations in the protocadherin 19 gene (*PCDH19*) constitute the second most common cause (Liu *et al*., 2017). *SCN1A* plays a role in encoding the sodium ion channel, which is associated with various seizure disorders, such as generalized epilepsy with febrile seizures plus and Dravet syndrome (Wang *et al*., 2010). PCDH19, a member of the cadherin superfamily, is mainly expressed in the nervous system. *PCDH19* is located on Xq22, and its variants lead to developmental and epileptic encephalopathy 9 (OMIM# 300088) (Zhao *et al*., 2020).

DEE9 is also known as epilepsy and mental retardation limited to females (EFMR). DEE9 is an X-linked genetic disorder that affects heterozygous females. It is characterized by the onset of seizures during infancy without cognitive impairment, various intellectual disturbances, and autistic features (Zhao *et al*., 2020). Since variants in *PCDH19* were initially reported to be related to DEE9, several studies have revealed associations between the clinical features of epilepsy and pathogenic variants in *PCDH19* (Chen *et al*., 2023; Kowkabi *et al*., 2024). These studies have demonstrated that next-generation sequencing (NGS) can clarify the pathogenic mechanism underlying DEE9 and yield information on gene mutation sites, which is useful for diagnosing individuals

suspected of epilepsy and can provide a basis for personalized treatment, genetic counseling, and risk assessment of the disease.

In the present study, we identified a pathogenic de novo variant showing a connection to the phenotype of *PCDH19* in an infant girl. Our study introduces a new aspect of the genetic pathogenicity of DEE9 that may promote the diagnosis and treatment of this form of epilepsy.

Materials and methods

Subjects

The proband, suffering from repeated convulsions, was admitted to the pediatric ward of Shenzhen People's Hospital in July 2020. MRI of the brain and electroencephalography (EEG) were performed, with EEG examinations conducted biannually during follow-up. Demographic characteristics and clinical manifestations were recorded. The levels of serum ceruloplasmin and electrolytes, infection markers, and antibodies against autoimmune encephalitis (serum14 items) were analyzed. Intellectual quotient and cognitive abilities were assessed by the Wechsler Intelligence Scale for Children, Fourth Edition, and Child Behavior Checklist. Informed consent was obtained from the proband's parents in accordance with the 1964 Declaration of Helsinki ethical standards. The project was approved by the ethics committee of Shenzhen People's Hospital.

Whole exome sequencing and data analysis

Whole exome sequencing (WES) of samples from the proband and her parents was conducted using a NovaSeq platform (Illumina, San Diego, California, USA). Variants were analyzed using a Genome Analysis Tool Kit with respect to the reference genome hg19 (Marini *et al*., 2012). Annotation was conducted by Annotate variation (ANNOVAR) (Wang *et al*., 2010), and an in-house pipeline was used to filter the common variants. Rare and coding variants were retained after filtering using minor allele frequency in the 1000 Genome Project and GnomAD databases of the eastern Asian population (cut-off frequency, 0.001). Only rare and coding variants were considered as potential genetic variants. MutationTaster, Likelihood Ratio Test, and PolyPhen-2 were used for evaluating the pathogenic variants. The American College of Medical Genetics and Genomics (ACMG) guidelines were used for variant interpretation and pathogenicity assessment (Wang *et al*., 2010).

Validation through Sanger sequencing

Genomic DNA was extracted from peripheral blood samples obtained from the patient and her parents using a QIAamp DNA Blood Kit (Qiagen, Hilden, Germany), following the manufacturer's protocol. The primers were designed using online tools of IDT PrimerQuest software (https://sg.idtdna.com/Primerquest/Home/Index). The primer sequences of gamma-aminobutyric acid receptor subunit delta (*GABRD*) and *PCDH19* were as follows: *GABRD*-F, 5'-CCAGAACAGTGCTGCATCC-3'; *GABRD*-R, 5'-CCTCACCTCCGATGCCA-3'; and *PCDH19*-F, 5'-TGGATCGCTGGCGTTGA-3'; *PCDH19*-R, 5'-GAGACGCAGTCGCACTACA-3'. All candidate genes segments were amplified through PCR. Candidate rare variants among subjects were genotyped using a Sanger sequencing platform. Sequences were analyzed using Chromas (http://technelysium.com.au/wp/chromas/).

Results

Clinical features

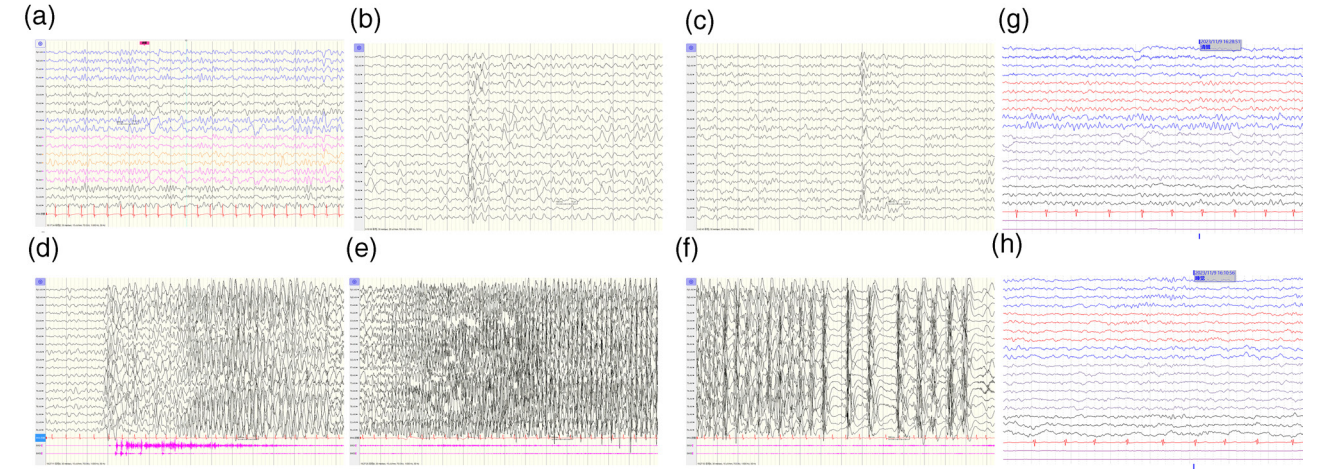
The proband was a 3-year-10-month-old girl who had suffered from recurrent epileptic seizures for more than 2 years. At the age of 18 months, she was afflicted by a 3-day-long episode of cluster febrile seizures characterized by troubled breathing, loss of contact, eyes rolled upwards, drooling, and shaking limbs, each lasting approximately 30 s to 1 min. Seizures recurred with fever and lasted for 2–7 days. She suffered from convulsions (with fever) twice or thrice a year between 2018 and 2019. When seizures were recurring, although without fever, and she did not exhibit any signs of intellectual disability (ID) or autism, she was inattentive and impulsive. She exhibited a slight delay in language acquisition. Her parents had a nonconsanguineous marriage and no family history of febrile convulsions or epilepsy. Clinical data and physical examinations did not indicate any signs of abnormalities. MRI findings of the brain were normal. Serum ceruloplasmin and electrolyte levels were normal, and infection markers and antibodies against autoimmune encephalitis were negative.

Analysis of electroencephalograms and diagnosis of epilepsy

In the awake state with the eyes closed, a poorly formed and fairly sustained posterior dominant rhythm of 5–6 Hz of moderate amplitude was noticed, which attenuated with eye opening. Background activity predominantly consisted of frequent activity with minimal delta, moderate theta, small alpha, and small beta activities. No significant regional asymmetries of background activity were noticed (Fig. 1a). During the interictal period, a small number of high-amplitude, generalized spike and wave or sharp-wave discharges (2–3.5 Hz) was noticed in the awake period, whereas a small number of high-amplitude spikes/sharps, and spike/sharp and waves were prominent in the bifrontal areas during awake and sleep periods (Figs. 1b, c).

Seizure onset was signaled by a sudden jerk of the body while sleeping, eyes opened, staring to the right, right upper limb raised, stiffness of the extremities, posture maintenance, and slight clonus. Synchronous EEG was characterized by generalized spike and wave, sharp-wave

Fig. 1



Polygraphic EEG and EMG channel waves showing a resting state and a focal seizure during sleep. (a) Background occipital rhythms lower than the normal rhythms in the awake state are shown. (b, c) Focal and generalized spikes and spike-slow waves particularly in the front head area during the interictal period. (d–f) A partial episode, which is suspected to have originated in the frontal area, is monitored and rapidly generalized. (g, h) Follow-up revealed normal spike and wave during awake and sleep after treatment. EEG, electroencephalography; EMG, electromyography.

discharges, or triphasic waves intermixed with low-amplitude fast activity. Generalized low-amplitude fast activity appeared in the initial portion of the epoch, which gradually increased in frequency and amplitude and finally developed into continuous generalized spike and waves superimposed with a large number of muscle artifacts synchronous with electromyography bursts, lasting approximately 50–60 s (Figs. 1d–f). These features provide strong evidence for the diagnosis of epilepsy. The disease phenotype was early onset infantile epileptic encephalopathy.

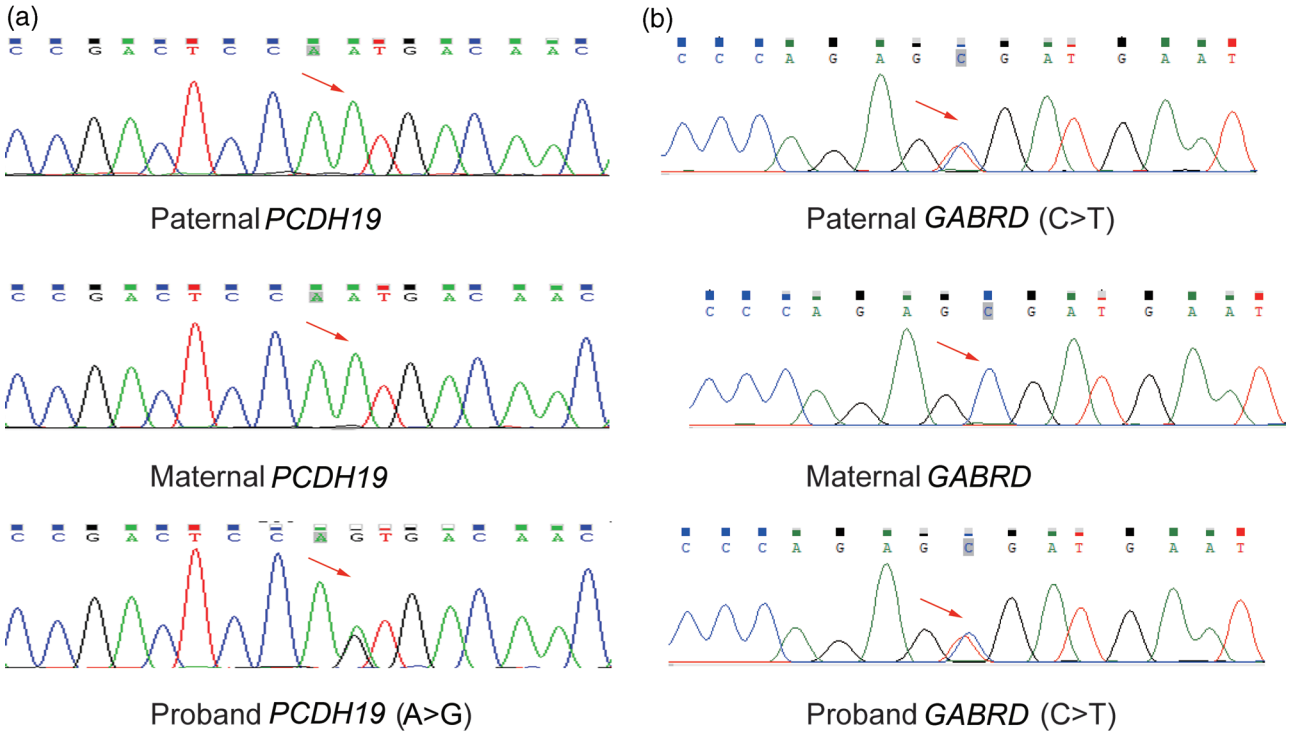
Whole exome sequencing revealed a *PCDH19* c.695A>G missense pathogenic variant

WES was performed for identifying the relevant genetic variants of candidate genes obtained from the online database MalaCards. Three coding variants were identified in epilepsy-associated candidate genes. One heterozygous variant (NM_001105243:c.695A>G) was observed in exon 1 of *PCDH19* of the proband (Fig. 2a; Table 1). This de novo variant was not observed in her parents. ACMG interpretation indicated that it was a likely pathogenic variant, while ClinVar demonstrated that this pathogenic variant was associated with DEE9. The proband also carried a nonsense heterozygous variant (NM_000815:c.71C>T) in exon 2 of *GABRD*, which is a paternally-inherited variant and considered benign (Fig. 2b). One rare variant of spectrin alpha, nonerythrocytic1 (*SPTAN1*) was likely a benign variant (Table 1). Based on the nontypical inheritance mode of the family and known association between *PCDH19* and DEE9, the *PCDH19* (NM_001105243:c.695A>G) variant was identified as the most promising pathogenic variant. Clinically recurrent epileptic seizures and abnormal synchronous discharges in the EEG combined with the discovery of *PCDH19* variant, led to the diagnosis of DEE9.

Treatment of seizures and follow-up results

After admission to the hospital, the patient continued to suffer from frequent convulsions, which manifested as clustered seizures for 6–10 times a day, mostly while crying or sleeping. To manage these symptoms, phenobarbital sodium and a small dose of mannitol were administered for sedation and dehydration, respectively, followed by the administration of vitamin B6 and additional treatments. This regimen successfully reduced the frequency of convulsions. A 15-h video EEG revealed abnormal brain activity for the child. Levetiracetam tablets (62.5 mg) were orally administered twice a day as antiepileptic treatment since July 2020, and the dose was gradually increased to 250 mg. Seizures did not recur, even during fevers. Following discharge, the patient regularly attended the pediatric outpatient clinic and remained free from seizure attacks, except for episodes associated with severely weakened immunity, until the last follow-up in July 2024. Throughout this period, the patient demonstrated normal intelligence and cognitive abilities, which differed with the clinical features reported in other studies involving the same variant (Table 2) (Breuillard *et al.*, 2016; Liu *et al.*, 2017; Chemaly *et al.*, 2018). She attended a classical school system without requiring additional assistance. EEG displayed normal spike and wave during awake and sleep after 3 years of treatment on December 2023 (Figs. 1g, h).

Fig. 2



Variants in (a) *PCDH19* (c.695A>G) and (b) *GABRD* (c.71C>T) of the proband, confirmed through Sanger sequencing. The evaluated nucleotide positions were marked with arrows.

Table 1 **Rare variants of epilepsy-associated genes identified in the subject**

Chromosome	Start	refGene	Alteration	Snp138	ExAC EAS	P/M/L[a]	ACMG
Chr1	1 956 383	*GABRD*	NM_000815:c. 71 C > T:p.A24V	Rs748341188	0.0001	D/D/D	Uncertain significance
ChrX	99 662 901	*PCDH19*	NM_001105243: c. 695 A > G:p.N232S	Rs587784299	NA	D/D/D	Likely_pathogenic
Chr9	1.31E + 08	*SPTAN1*	NM_001130438: c. 1691 G > A:p.R564H	Rs201168391	0.0002	B/N/N	Likely_benign

ACMG, American College of Medical Genetics and Genomics; ExAC EAS, East Asian of Exome Aggregation Consortium.
[a]MutationTaster (M), Likelihood Ratio Test (L), PolyPhen-2 (P) pathogenic evaluation results of variants. The variants were categorized as follows: D (Deleterious or Disease-causing), B (Benign), and N (polymorphism or Neutral).

Table 2 **Clinical features in other studies involving the same variant (c.695A>G) in *PCDH19***

Studies	Age (months)	Seizures type	Fever sensitivity	Seizure clusters	Intellectual
Breuillard *et al.* (2016)	9	Generalized/ focal seizures	Yes	Yes	Mild delay
Liu *et al.* (2017)	5	Multiple seizure types	Yes	Yes	Disability
Chemaly *et al.* (2018)	12	Generalized/ focal seizures	Yes	Yes	Mild delay
Present study	18	Generalized/ focal seizures	Yes	Yes	Normality

Discussion

This study investigated a classic case of epilepsy with recurrent epileptic seizures and abnormal synchronous discharges in the EEG. Three rare missense variants, including a pathogenic de novo variant (NM_001105243:c.695A>G) in *PCDH19*, and two benign variants *SPTAN1* and *GABRD* (c.71C>T), were identified. The patient was diagnosed with DEE9 based on the clinical features and genetic analyses. A 4-year follow-up revealed normal EGG patterns and normal intelligence, highlighting the positive impact of levetiracetam therapy in treating genetic DEE9.

Some epilepsies can be divided into several genetic syndromes, including Dravet syndrome (MIM# 607208) caused by variants in *SCN1A* (Claes *et al.*, 2001), *CDKL5*/*STK9* Rett-like epileptic encephalopathy (Tao *et al.*, 2004; Weaving *et al.*, 2004), and DEE9 associated with variants in *PCDH19* (Dibbens *et al.*, 2008). PCDH19 plays an important role in the disease process and diagnosis of DEE9. *PCDH19* contains six exons that encode

a protocadherin protein and contribute to the function of cadherin (Zhao *et al*., 2020). Although the physiological role and pathogenic mechanisms of PCDH19 remain unclear, it may contribute to maintaining neuronal homeostasis (Gerosa *et al*., 2022).

Epilepsy related to heterozygous female children with clinical manifestations of *PCDH19* variant shows a diverse phenotype. Variants in *PCDH19* of heterozygous females and somatic mosaic males cause DEE9 that demonstrates ID and autistic features (Kolc *et al*., 2019). In our study, the patient did not exhibit any signs of ID or autism, which contrasts with the reported cases of autistic features (Breuillard *et al*., 2016; Chemaly *et al*., 2018) and ID (Liu *et al*., 2017) in other carriers of the same variant. The three reported cases exhibited classical clinical features, including generalized/focal seizures, fever sensitivity, and seizure clusters, which were consistent with our present case. The age of onset for PCDH19-related epilepsy ranges between 1 and 70 months, with a median of 10 months (Dibbens *et al*., 2008). Mental retardation is not a certain clinical manifestation in most DEE9 cases. Some patients with a variant in *PCDH19* present certain clinical manifestations that overlap with Dravet syndrome without *SCN1A* mutations (Liu *et al*., 2017). PCDH19 plays a major role in infantile-onset familial or sporadic epilepsy in female patients (Depienne *et al*., 2009; Marini *et al*., 2010). In our study, classic clinical manifestations were observed in the proband, and clinical manifestations included a combination of fever-induced seizures, abnormal EEG, and genetic defects. Based on the clinical features, genetic findings, and inherited pattern, the proband in our study was diagnosed with DEE9 owing to a variant (NM_001105243:c.695A>G) in *PCDH19* (Depienne and LeGuern, 2012; Liu *et al*., 2017). Further follow-up revealed normal intelligence and EGG patterns, highlighting the positive impact of genetic counseling and clinical treatment in managing DEE9. Compared to previously reported cases (Depienne and LeGuern, 2012; Liu *et al*., 2017), our study provides a detailed depiction of the phenotype associated with this variant (NM_001105243:c.695A>G), along with valuable long-term follow-up outcomes on the treatment of genetic DEE9.

Epilepsy has a variable age of onset, with or without fever sensitivity, cognitive impairment, and resistance to antiepileptic treatment (Kavehei *et al*., 2019). Genetic factors significantly affect epilepsy (Zhang *et al*., 2024). Presently, early diagnosis and implementation of effective treatment strategies for efficient clinical management of patients with PCDH19-related epilepsy is challenging. Therefore, early and accurate diagnosis of this complex heterogeneous disease using genetic testing, which reveals the genetic architecture and phenotypic and genetic complexities of epilepsy, is of utmost importance.

In conclusion, NGS-based genetic analyses may help improve the diagnosis and treatment of PCDH19-related epilepsy. Moreover, by utilizing the high-throughput results following genetic analyses, an effective genetic counseling and clinical treatment guidance for DEE9 may be developed.

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The project was approved by the ethics committee of the Ethics Committee of Shenzhen People's Hospital. Informed consent was obtained from the proband's parents in accordance with the 1964 Declaration of Helsinki ethical standards.

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The datasets used or analyzed during the current study are available from the corresponding author on reasonable request.

Conflicts of interest

There are no conflicts of interest.

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