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Case Report

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A homozygous start-codon variant in PEX10 presenting as West syndrome: a case report

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Abstract

Background: PEX10-related peroxisome biogenesis disorder type 6 (PBD6) belongs to the Zellweger spectrum disorders (ZSDs), a group of rare autosomal recessive diseases caused by defective peroxisome biogenesis. PEX10-related cases presenting initially with infantile spasms (West syndrome) are extremely rare, and the genotype–phenotype relationship of start-codon variants has not been systematically characterized.

Case presentation: We report a Chinese female infant with onset at 3 months of age of recurrent clusters of epileptic spasms, occurring more than 20 times per day, accompanied by delayed motor and cognitive development. Video-EEG showed atypical hypsarrhythmia, and epileptic spasms were recorded; West syndrome was diagnosed. Brain MRI showed diffuse bilateral T2-weighted hyperintensity of the cerebral white matter, ventricular enlargement, and a thin corpus callosum. After treatment with ACTH pulse therapy, topiramate, and clobazam, seizures decreased, but intermittent spasms persisted up to the last follow-up at 1 year of age. During follow-up, the patient developed global developmental delay, recurrent severe pneumonia (requiring non-invasive ventilation for respiratory failure), persistent liver dysfunction (peak ALT 453 U/L, peak AST 522 U/L), and hyperammonemia (peak 80 μmol/L). Tandem mass spectrometry and urine organic acid analysis showed no clearly specific abnormalities. Trio whole-exome sequencing (trio-WES) identified a homozygous start-codon variant in PEX10, NM_002617.4:c.3G>A (p.Met1?), with both parents being heterozygous carriers. The variant abolishes the normal translation initiation site; its pathogenic mechanism and its effect on protein function remain to be verified by functional studies.

Conclusions: This case expands the neurological phenotypic spectrum of PEX10-related disease and suggests that West syndrome may be an early manifestation of PEX10 start-codon variants. In infants with unexplained infantile spasms and developmental delay, genetic testing and screening for peroxisomal disorders should be considered even when routine metabolic screening is normal. The pathogenic mechanism of this variant requires functional validation.

Keywords: PEX10; Zellweger spectrum disorder; West syndrome; infantile spasms; peroxisome biogenesis disorder; start-codon variant

Background

Zellweger spectrum disorders (ZSDs) are autosomal recessive diseases caused by impaired peroxisome biogenesis, mainly due to variants in PEX (peroxin) genes [1–3]. Peroxisomes participate in key metabolic processes including β-oxidation of very-long-chain fatty acids (VLCFAs), phytanic acid metabolism, bile acid synthesis, and clearance of reactive oxygen species, and their dysfunction affects multiple organ systems, producing a continuous clinical spectrum from lethal neonatal disease to mild, late-onset neurological disorders [1–3]. Of at least 13 PEX genes associated with ZSD, PEX1 and PEX6 are the most common, whereas PEX10-related cases are relatively uncommon [2,3]. PEX10, located on chromosome 1p36, encodes a component of the PEX2–PEX10–PEX12 ubiquitin ligase complex that is essential

for peroxisomal matrix protein import [4,5]. Its loss causes peroxisome biogenesis disorder type 6 (PBD6), comprising subtypes PBD6A and PBD6B, both belonging to the ZSD [3].

Previous studies have shown marked clinical heterogeneity in PEX10-related disease, with phenotypes spanning classic Zellweger syndrome, neonatal adrenoleukodystrophy, and milder ataxic syndromes with childhood or adult onset [6,7]. Neurological involvement is the most common feature, including global developmental delay, hypotonia, white matter abnormalities, cerebellar ataxia, and seizures [3,6,7]. Infantile spasms (West syndrome) is a common developmental and epileptic encephalopathy of infancy [8,9]; however, PEX10-related cases presenting with it as the initial manifestation have been reported only extremely rarely.

Start-codon variants are a special class of loss-of-function variants that directly affect translation initiation. Recent studies suggest that some start-codon variants may generate partially functional truncated proteins through alternative translation initiation, resulting in relatively milder clinical phenotypes, and that their clinical impact is closely related to the presence of downstream alternative initiation sites [10,11]. However, genotype–phenotype correlations of PEX10 start-codon variants have not been systematically studied.

Here we report a Chinese infant carrying a homozygous PEX10 start-codon variant, c.3G>A (p.Met1?), who presented with West syndrome as the initial manifestation together with severe developmental delay, white matter abnormalities, persistent liver dysfunction, and hyperammonemia.

Case presentation

General information and clinical course

The patient was a girl born at term by spontaneous vaginal delivery with a birth weight of 2.7 kg and no obvious history of perinatal hypoxia or asphyxia. The parents were non-consanguineous, and there was no family history. Feeding after birth was adequate, but early motor and cognitive development gradually lagged; at 3 months of age she still could not hold her head up and showed poor visual and auditory tracking.

At 3 months of age, without obvious precipitating factors, the patient developed recurrent clusters of spasms, manifesting as sudden flexion of both upper limbs, clenched fists, head-nodding movements, and forceful limb movements, sometimes accompanied by crying. Each spasm lasted about 1–2 seconds and occurred in clusters, up to more than 20 times per day, with decreased responsiveness after the episodes. The seizure frequency gradually increased

after onset, and she was admitted to the Department of Neurology of Hebei Children's Hospital. Neurological examination showed unstable head control, inability to roll over, poor visual and auditory tracking, and mildly decreased muscle tone.

The patient was successively treated with ACTH pulse therapy and the antiseizure medications topiramate and clobazam. Seizures decreased, but intermittent spasms persisted, indicating an incomplete response to treatment, including ACTH. During the disease course she developed recurrent severe pneumonia with repeated dyspnea and hypoxemia, and required non-invasive ventilation for respiratory failure; her condition stabilized after anti-infective and supportive treatment. In June 2026 she was readmitted for severe pneumonia and respiratory failure and improved with symptomatic treatment. Her most recent hospitalization was from 17 to 24 September 2026, at the age of 1 year, for fever and bronchopneumonia. At this last follow-up, epileptic spasms persisted despite treatment, occurring as isolated or clustered events lasting a few seconds, more than 20 times per day before admission, and they were still present intermittently at discharge. Cenobamate was added to topiramate, and clobazam was tapered to discontinuation. Developmentally, she still showed no visual fixation or sound tracking, could not follow objects, had no social smile, and could not hold her head up, roll over, or sit independently. The Alberta Infant Motor Scale total score was 7 (<5th percentile), and the 0–6-year child developmental examination gave developmental quotients (DQ) of 37 (adaptive), 41 (gross motor), 44 (fine motor), 40 (language), and 22 (personal–social). Head circumference was 40.5 cm, and limb muscle strength and tone were normal. Liver involvement persisted: aspartate aminotransferase was 84 U/L and total bile acids 114.6 μmol/L, the liver edge was palpable 3 cm below the costal margin, and abdominal ultrasound showed a mildly enlarged liver with increased and uneven parenchymal echogenicity.

Video-EEG

Video-EEG was recorded using the international 10–20 system. The first video-EEG (10 January 2026) showed a disorganized background with diffuse high-amplitude slow-wave activity. Interictal recordings showed multifocal sharp waves, spikes, and spike-and-wave discharges, most prominent in the central, parietal, and midline regions, giving an overall pattern of atypical hypsarrhythmia (Fig. 1A). Typical epileptic spasms were captured ictally, with the EEG showing diffuse low-voltage suppression followed by slow-wave activity (Fig. 1B), consistent with West syndrome. A follow-up video-EEG on 2 February 2026 again showed an abnormal background without a posterior dominant rhythm, atypical hypsarrhythmia with posterior predominance, and recorded epileptic spasms, indicating a

persistent developmental and epileptic encephalopathy.

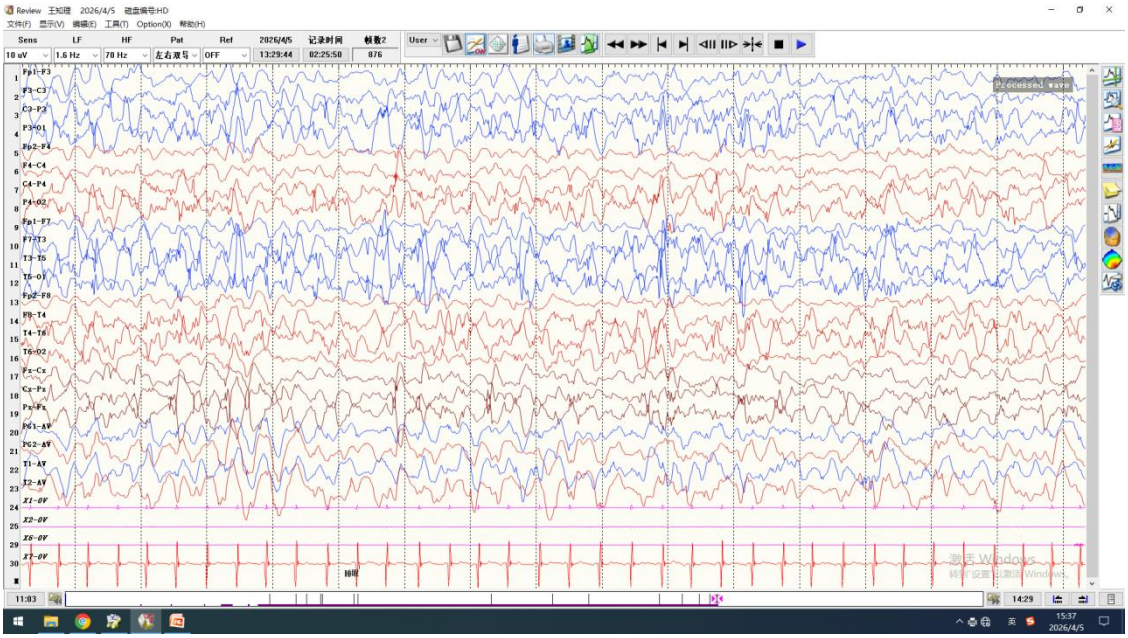


Fig. 1A First video-EEG showing atypical hypsarrhythmia.

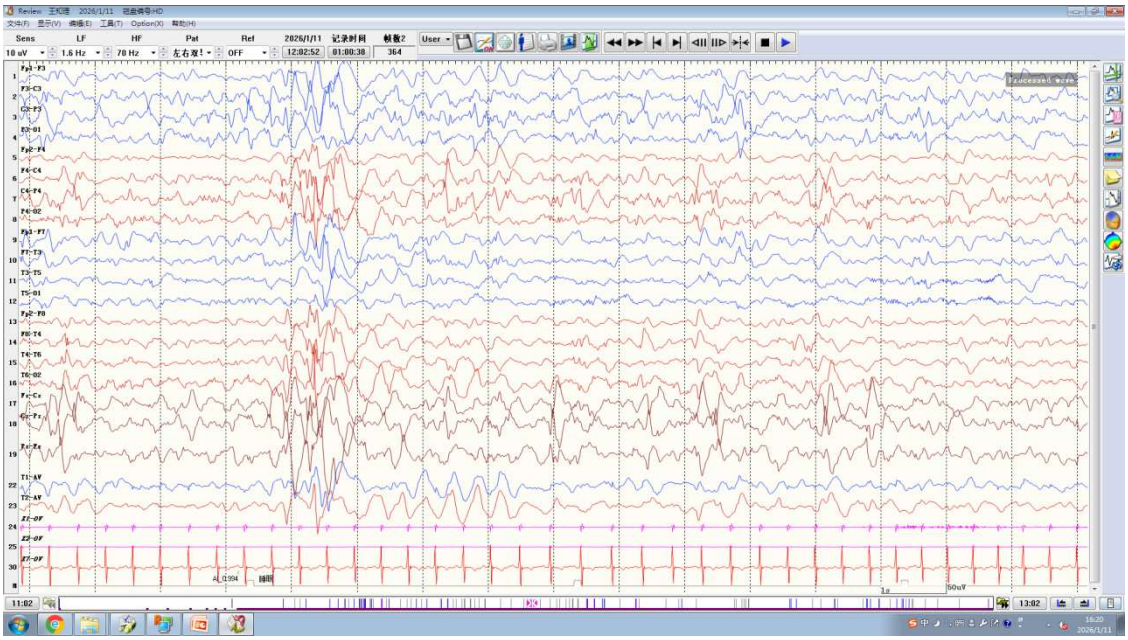


Fig. 1B Ictal EEG recording a typical epileptic spasm.

Neuroimaging

Brain MRI (at 4 months of age) was independently reviewed by two experienced pediatric neurologists. Plain scans showed fine, closely spaced gyri; diffuse T2-weighted hyperintensity of the bilateral cerebral hemispheric white matter; widened bilateral frontotemporal extra-axial spaces; bilateral ventricular enlargement with pointed posterior horns; and a thin corpus callosum with a cavum septum pellucidum (Fig. 2), indicating widespread white matter involvement and abnormal brain development consistent with peroxisome biogenesis

disorders.

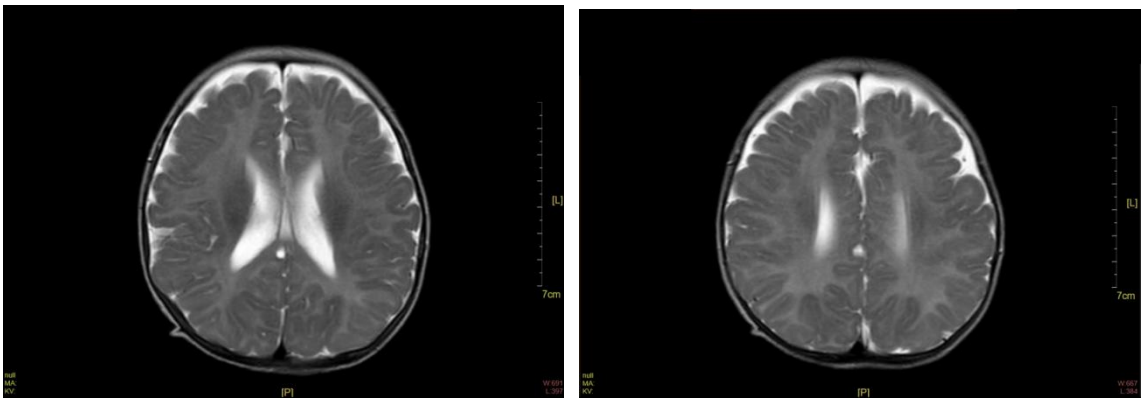


Fig. 2 Brain MRI. A: Abnormal hyperintensity of the bilateral cerebral white matter, bilateral ventricular enlargement, and a thin corpus callosum; B: Closely spaced fine gyri and abnormal cortical development.

Laboratory and metabolic investigations

Amino acid and acylcarnitine profiles were measured by tandem mass spectrometry (MS/MS) on a dried blood spot, and urine organic acids (132 analytes) by gas chromatography–mass spectrometry (GC–MS) on dried urine filter paper; both samples were collected at 4 months of age. Repeated testing during the disease course showed persistent liver dysfunction: peak alanine aminotransferase (ALT) 453 U/L, peak aspartate aminotransferase (AST) 522 U/L, and peak total bile acids 114.6 μmol/L. Blood ammonia remained elevated, with a peak of 80 μmol/L. Amino acid and acylcarnitine profiles showed no significant abnormalities, apart from a mildly elevated C18 dicarboxylic acylcarnitine (C18DC, 0.20 μM; reference 0.01–0.10), a mildly decreased C3/C2 ratio (0.01; reference 0.02–0.20), and a mildly elevated C16/C3 ratio (3.79; reference 0.10–3.50). Urine organic acid analysis likewise showed no specific abnormal metabolites, with only mildly elevated 4-hydroxyphenyllactic acid (20.8; reference 0.0–20.0) and 2-hydroxysebacic (2-hydroxydecanedioic) acid (29.4; reference 0.0–12.0). Peroxisome-specific markers, including VLCFAs, phytanic acid, pristanic acid, bile acid intermediates, and plasmalogens, were not measured in this patient, and functional validation was not performed owing to technical limitations.

Genetic testing

The patient and her parents underwent trio whole-exome sequencing (trio-WES). Reads were aligned to the human reference genome GRCh37/hg19, and variants were annotated using the ClinVar, HGMD, and gnomAD databases. Pathogenicity was classified according to the American College of Medical Genetics and Genomics (ACMG) guidelines, and candidate variants were validated in the family by Sanger sequencing.

Trio-WES identified a homozygous variant in PEX10, NM_002617.4:c.3G>A (p.Met1?), located at the translation initiation codon and expected to abolish the normal translation start site. Sanger sequencing showed that both parents (aged 29 and 31 years) were heterozygous carriers without related phenotypes (Fig. 3), consistent with autosomal recessive inheritance. The variant was classified as likely pathogenic according to the ACMG guidelines (PVS1_Moderate + PM5 + PM2_Supporting + PM3_Supporting). PVS1 was applied at moderate strength in line with the ClinGen recommendations for initiation-codon variants, and PM5 was applied because a different amino acid change at the same residue (Met1), c.2T>C (p.M1T), has been classified as pathogenic/likely pathogenic. The variant (chr1:2343939, GRCh37; rs1393899190) is listed in ClinVar as pathogenic/likely pathogenic (Variation ID: 2733820), has a gnomAD v3 frequency of 0.0000066 (0 in East Asians), and is absent from the other databases. Considering the clinical manifestations, neuroimaging changes, and parental carrier results, the patient was diagnosed with PEX10-related PBD6; because peroxisomal biochemical testing was not performed, PBD6A and PBD6B could not be distinguished. The karyotype was 46,XX, and no pathogenic or likely pathogenic copy-number variants, structural variants, uniparental disomy, or dynamic mutations were detected. Heterozygous variants of uncertain significance in RELN, ASPM, CNTNAP1, POLR1A, and SPATA5L1, each inherited from a phenotypically normal parent, were considered insufficient to explain the phenotype.

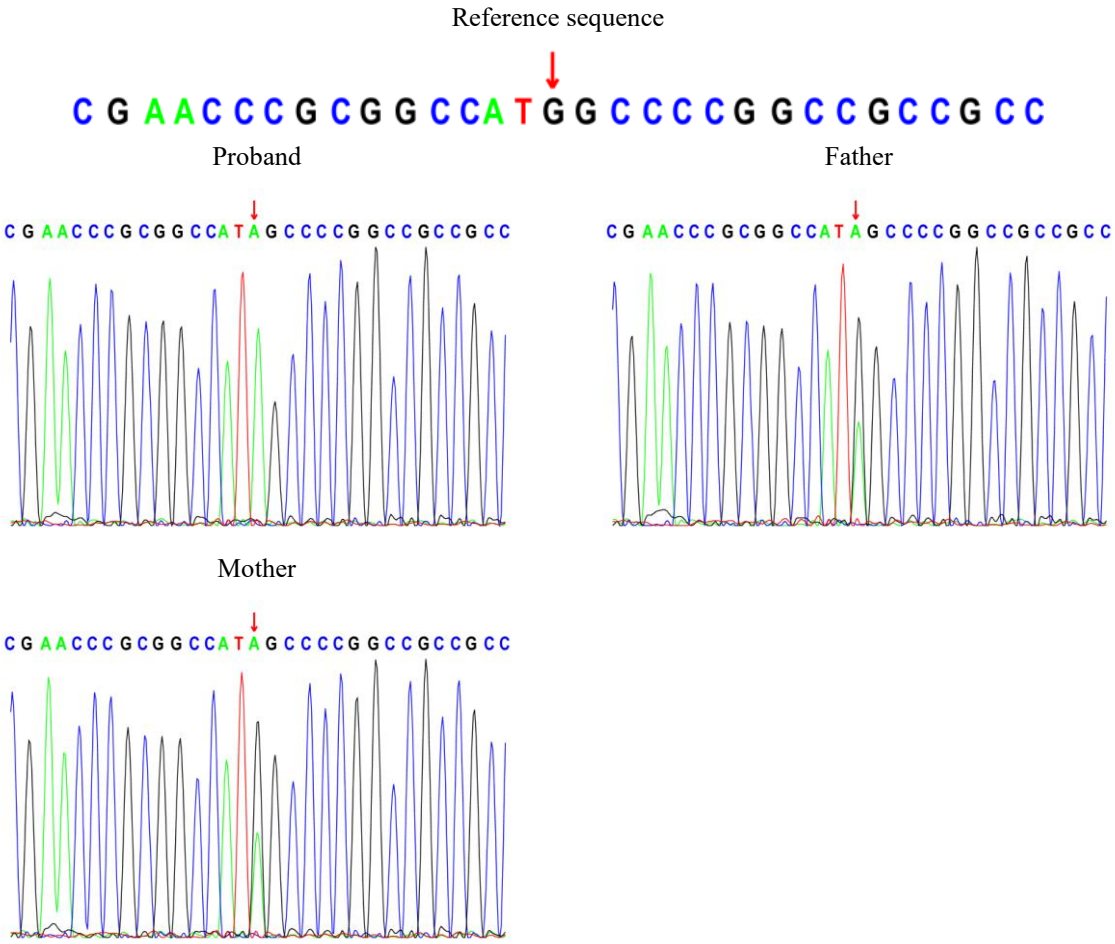


Fig. 3 Sanger sequencing verification in the family. Arrows indicate position c.3: the proband is homozygous for A, and both parents are heterozygous G/A.

Discussion and conclusions

West syndrome may be an early neurological phenotype of PEX10-related disease

West syndrome is a developmental and epileptic encephalopathy with infantile onset, characterized by epileptic spasms, hypsarrhythmia, and developmental arrest or regression [8,9]. Many inherited metabolic diseases can cause West syndrome, including mitochondrial diseases, disorders of amino acid metabolism, organic acidemias, and peroxisomal disorders [12].

Previously reported PEX10-related cases mainly presented with ataxia, cerebellar atrophy, peripheral neuropathy, hearing loss, and mild cognitive impairment [6,7,13–15]. Régal et al. described PEX10-related autosomal recessive ataxia [6], and the cases subsequently reported by Yamashita, Renaud, and Zhang et al. were likewise characterized mainly by slowly progressive ataxia [7,13,14]. However, to the best of our knowledge based on our literature search, there has been no report of a homozygous PEX10 start-codon variant

presenting initially as West syndrome.

Peroxisomes play important roles in neuronal migration, axonal development, myelination, and lipid metabolism, and their dysfunction can impair brain development and neural network formation [1,2]. We therefore speculate that impaired PEX10 function may precipitate infantile epileptic encephalopathy by affecting neural network development; in this patient, the response to treatment, including ACTH, was only partial.

Start-codon variants and the phenotype of this patient

Start-codon variants usually cause failure of translation initiation and have traditionally been regarded as loss-of-function variants [10,11]. However, some start-codon variants may generate partially functional truncated proteins through alternative translation initiation, thereby attenuating disease severity [10]. The patients reported by Yamashita et al. carried the start-codon variant c.2T>C (p.M1T) (in compound heterozygosity with p.C307Y); all survived to adulthood and showed a relatively mild neurological phenotype [7], suggesting that PEX10 start-codon variants may not completely abolish protein function.

The c.3G>A variant in this patient abolishes the translation initiation site. Theoretically, if a usable in-frame downstream ATG exists, translation could re-initiate and produce an N-terminally truncated protein that retains partial function, which might be related to the patient not dying in the neonatal period; however, no prediction analysis or functional validation was performed in this study, and this explanation is only speculative. It should be noted that, although this patient has survived to 1 year of age, she already has severe neurodevelopmental impairment, persistent epileptic spasms, recurrent severe pneumonia, and persistent liver dysfunction; therefore, the available evidence is insufficient to classify this case as a mild phenotype, and the long-term prognosis requires continued follow-up.

Compared with the previously reported c.2T>C (p.M1T) cases, our patient had a markedly earlier age at onset and a more severe neurological phenotype characterized by West syndrome and developmental and epileptic encephalopathy. This suggests that such variants can cause not only slowly progressive ataxia in childhood but also severe infantile-onset neurodevelopmental impairment and epileptic encephalopathy.

Normal routine metabolic screening does not exclude PEX10-related disease

The traditional diagnosis of peroxisomal disorders relies mainly on measurement of metabolic markers such as VLCFAs, phytanic acid, pristanic acid, bile acid intermediates, and plasmalogens [1,3]. However, a growing number of mild ZSD cases have been found to have

only mild, or even normal, metabolic abnormalities [15,16]. In this patient, routine acylcarnitine and urine organic acid profiles showed no specific abnormalities, and these tests do not reflect peroxisomal function; the diagnosis was finally established by trio-WES. Blomqvist et al. similarly reported a patient with PEX10-related mild ZSD who had minimal metabolic abnormalities but prominent neurological symptoms [16]. Therefore, in children with unexplained developmental delay, epilepsy, white matter abnormalities, and liver dysfunction, PEX-related disease should still be considered even if routine metabolic screening results are normal, and genetic testing should be performed early.

Genotype–phenotype correlation

Warren et al. found that nonsense, frameshift, and splice variants causing severe protein truncation usually correspond to classic severe Zellweger syndrome, whereas missense variants that retain partial function are more common in neonatal adrenoleukodystrophy and milder phenotypes [17]. Recently reported mild PEX10 patients mainly carry missense variants, with some carrying start-codon variants (such as c.2T>C), and present mainly with ataxia, cerebellar atrophy, and peripheral neuropathy [6,7,13–15]. Our patient carries a homozygous start-codon variant yet presented with West syndrome in infancy and severe developmental delay, suggesting that the clinical phenotype of PEX10 start-codon variants may be much broader than previously recognized. Taking this case and previous reports together, we believe that PEX10-related disease is not simply an ataxic syndrome or classic ZSD, but may encompass a continuous phenotypic spectrum from infantile epileptic encephalopathy to slowly progressive neurodegenerative disease in adulthood.

Limitations

This study has the following limitations. First, peroxisome-specific metabolic markers such as VLCFAs, phytanic acid, pristanic acid, and bile acid intermediates were not measured, so peroxisomal functional deficiency could not be verified biochemically. Second, no fibroblast functional experiments or protein expression analyses were performed, so the pathogenic mechanism of the variant and whether alternative translation initiation occurs remain speculative. Third, the previously reported c.2T>C cases were compound heterozygotes, so the influence of the other allele cannot be excluded, which limits direct comparison with our patient. Fourth, the patient has been followed only until 1 year of age, and the long-term prognosis and disease course remain unclear.

Conclusions

We report a Chinese infant with a homozygous PEX10 start-codon variant, c.3G>A (p.Met1?), who presented with West syndrome accompanied by white matter abnormalities, severe developmental delay, persistent liver dysfunction, and hyperammonemia, further expanding the clinical phenotypic spectrum of PEX10-related disease. In infants with unexplained infantile spasms and developmental delay, genetic testing and screening for peroxisomal disorders should be emphasized. The pathogenic mechanism of this variant and its impact on the clinical phenotype still need to be confirmed by functional experiments.

Declarations

List of abbreviations: ZSD, Zellweger spectrum disorder; PBD6, peroxisome biogenesis disorder type 6; VLCFAs, very-long-chain fatty acids; EEG, electroencephalography; Trio-WES, trio whole-exome sequencing; ACMG, American College of Medical Genetics and Genomics; MRI, magnetic resonance imaging; ACTH, adrenocorticotropic hormone; ALT, alanine aminotransferase; AST, aspartate aminotransferase; MS/MS, tandem mass spectrometry; GC–MS, gas chromatography–mass spectrometry.

Ethics approval and consent to participate: This study was conducted in accordance with the principles of the Declaration of Helsinki, and informed consent was obtained from the patient's guardians. Ethical approval was granted by the Medical Research Ethics Committee of Hebei Children's Hospital (approval No. 2025034-51).

Consent for publication: Written informed consent for publication of the case details and accompanying images was obtained from the patient's guardians. A copy of the consent form is available on request from the editorial office.

Availability of data and materials: The datasets generated and analysed during the current study are not publicly available because the patient's legal guardians did not consent to the deposition of the patient's genetic data, including the raw sequencing data, in public repositories. The variant reported here, PEX10 NM_002617.4:c.3G>A (p.Met1?), is publicly available in ClinVar (Variation ID: 2733820). All other data supporting the findings are included in this article.

Competing interests: The authors declare that they have no competing interests.

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collection, analysis, interpretation of data, or writing of the manuscript.

Authors' contributions: YD and ZN collected the clinical data, analyzed and interpreted the patient data, and drafted the manuscript. WW assisted with data collection and with the analysis and interpretation of the genetic, EEG, and imaging data. XL and FC designed and supervised the study and critically revised the manuscript. YD and ZN contributed equally to this work. All authors read and approved the final manuscript.

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