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NEUROD2-related disorder with neonatal onset: case report and review of the literature

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Abstract

The *NEUROD2* gene encodes a transcription factor essential for neuronal differentiation and cortical development. Pathogenic variants cause a rare autosomal dominant neurodevelopmental disorder with variable expressivity, typically presenting in early infancy with developmental delay, epilepsy, and behavioral abnormalities. We report a newborn girl carrying a de novo heterozygous missense variant *NM_006160.4*:c.790G>A, p.(Ala264Thr), located outside the canonical basic helix-loop-helix (bHLH) domain. Soon after birth, she presented respiratory depression, hypotonia, feeding difficulties, and electrographic seizures. Magnetic resonance imaging (MRI) showed subcortical white matter hyperintensity, and the electroencephalogram (EEG) revealed abnormal background activity. During follow-up, epilepsy was controlled, but neurodevelopmental delay with autistic features emerged. This case represents the earliest reported clinical onset associated with *NEUROD2* variants and expands the phenotypic and mutational spectrum. It highlights that variants outside known hotspots can cause severe disease and supports including *NEUROD2* in the differential diagnosis of neonatal neurological impairment.

Keywords

NEUROD2 case report, neurodevelopmental disorder, neonatal hypotonia, neonatal seizures, autistic features

Introduction

The *NEUROD2* gene (OMIM #601725) encodes a neuronal basic helix-loop-helix (bHLH) transcription factor that plays a critical role in neuronal differentiation, synaptic maturation, and maintenance of excitatory-inhibitory balance within the central nervous system. Experimental studies have shown that *NEUROD2* is essential for the development and maturation of cortical neurons, and its dysfunction can disrupt neuronal circuit formation and synaptic plasticity [1–4].

Pathogenic *NEUROD2* variants have recently been implicated in a rare autosomal dominant neurodevelopmental disorder (OMIM #618374) characterized by variable expressivity, including global developmental delay, intellectual disability (ID), epilepsy, behavioral abnormalities, and, in some cases, structural brain changes [5–10].

To date, only a limited number of *NEUROD2* variants have been described, and most are clustered in the highly conserved bHLH domain, which is crucial for DNA binding and dimerization. Reported clinical presentations usually begin during infancy or childhood, often with infantile seizures, developmental delay, and abnormal electroencephalogram (EEG) findings. However, the phenotypic spectrum is still being defined, and neonatal presentation is unreported [5–10]. Expanding the number of well-characterized cases is essential for improving our understanding of genotype-phenotype correlations and the underlying disease mechanisms.

Here, we report the case of a newborn girl carrying a de novo heterozygous missense variant *NM_006160.4*:c.790G>A in the *NEUROD2* gene, associated with early neonatal hypotonia, feeding difficulties, and electrographic seizures. This case report is notable for the very early onset of symptoms**,** beginning immediately after birth, and for the identification of a novel variant located outside the typical mutational hotspot. It provides additional insight into the clinical variability and genetic landscape of *NEUROD2*-related disorders.

Timeline

Table 1 shows the timeline, giving a clear and concise roadmap of the case.

Table 1. Timeline.

Time	Features
At birth	Respiratory depression
First week of life	Hypotonia, hyporeactivity, and feeding difficulties
First month	Neonatal seizures
Second month	Persistent hypotonia
Third month	Discharge
6th to 12th month	Normal growth. Psychomotor retardation
24th month	Normal growth. Psychomotor retardation
30th month	Autistic features

Narrative

The baby girl was born at term via vaginal delivery with meconium-stained amniotic fluid. The pregnancy progressed normally, with a prenatal finding of a right pelvic kidney. At birth, she presented respiratory depression and was therefore ventilated, with subsequent recovery. Apgar scores were 2–6 and 7. Birth weight was 3,200 g (20th centile), length 51 cm (48th centile), and head circumference 33 cm (13th centile). Due to respiratory distress, she was admitted to the neonatal intensive care unit. Since the first days of life, she showed signs of altered neurological status, including hypotonia, hypo-reactivity, and feeding difficulties. During hospitalization, the baby presented with persistent hypotonia, hypo-reactivity, with a predominant behavioral state of sleep/drowsiness and crying episodes with stereotyped and abnormal movements. Repeated EEG evaluations showed an excessively discontinuous baseline EEG

pattern and recorded isolated electrographic seizures with minimal clinical correlation (stereotyped movements like rowing and pedaling). Antiepileptic therapy with levetiracetam was therefore administered. The infant required non-invasive respiratory support for several days and prolonged orogastric tube feeding due to poor or absent sucking, although swallowing capacity was substantially preserved. By 2 months of age, she was able to feed herself with a bottle. She was therefore discharged and followed up (see Table 1). At 24 months of age, the patient presented normal growth and epilepsy control with levetiracetam. At 30 months of age, a comprehensive neurodevelopmental assessment was performed by a multidisciplinary team comprising a child neuropsychiatrist and a clinical psychologist, both with expertise in neurodevelopmental disorders. The patient demonstrated persistent deficits in social communication and social interaction across multiple contexts, as well as restricted patterns of behavior, meeting diagnostic criteria for autism spectrum disorder (ASD), according to DSM-5 clinical criteria. The assessment included clinical observation, developmental history, and standardized diagnostic instruments. The diagnosis was supported by the Autism Diagnostic Observation Schedule (ADOS-2, administered by clinicians) and the Autism Diagnostic Interview-Revised (ADI-R), completed with the primary caregiver. Thus, at 30 months of age, the patient exhibited a neurodevelopmental disorder characterized by global developmental delay and ASD. Ongoing multidisciplinary follow-up continues to monitor developmental progress and therapeutic interventions.

Diagnostics

Several diagnostic investigations were carried out in the neonatal period, including brain magnetic resonance imaging (MRI), which revealed hyperintensity of the subcortical white matter, and genetic testing. Array CGH, Prader-Willi, and myotonic dystrophy genetic tests resulted in normal. Trio exome sequencing revealed a heterozygous missense variant in exon 2 of the *NEUROD2* gene: *NM_006160.4*:c.790G>A, leading to the amino acid substitution p.(Ala264Thr). The variant was confirmed by Sanger sequencing and was shown to be de novo (absent in both parents) (Table 2). In silico predictive tools consistently indicated a deleterious effect: SIFT predicted "damaging" (score 0.00), PolyPhen-2 predicted "probably damaging" (score 0.999), and the CADD score was 28.9 (strongly suggestive of pathogenicity). Splice AI predicted no significant impact on splicing (max score 0.02). The affected residue (alanine at position 264) is highly evolutionarily conserved across species, supporting its functional importance. Population database analysis showed that the variant is absent in gnomAD v4.0 (including all subpopulations), consistent with a rare disease-causing allele. Applying the ACMG/AMP guidelines [11, 12], the variant met the following criteria: PS2 (de novo in a patient with the disease and no family history), PM2 (absent from population databases), and PP3 (multiple lines of computational evidence supporting a deleterious effect). The overall classification framework is consistent with subsequent recommendations [13]. Collectively, these data support the classification of the variant as likely pathogenic. Monoallelic variants in the *NEUROD2* gene have recently been associated with a neurodevelopmental disorder (OMIM #618374) with autosomal dominant inheritance and variable expressivity. The *NEUROD2 NM_006160.4* :c.790G>A variant has been submitted to ClinVar (SUB15787434) and is under processing (accession number SCV007105870). Parental consent for clinical description has been obtained.

Patient perspective

The parents of the infant were actively involved throughout the diagnostic and therapeutic journey. They provided written informed consent for all procedures, including genetic testing, and for the publication of this report. Upon learning of the de novo *NEUROD2* variant and its likely pathogenicity, the parents expressed a mixture of relief at having a definitive diagnosis and concern regarding their daughter's long-term neurodevelopmental prognosis. The parents reported that the most challenging aspects during the neonatal period were the infant's prolonged hypotonia, feeding difficulties requiring orogastric tube feeding, and the emotional distress associated with the hospital stay. They appreciated the structured communication from the neonatal intensive care unit staff and the early involvement of the psychology unit,

Table 2. Summary of published *NEUROD2* cases (2019–2024)*.

Case	Reference	Sex	Variant (cDNA)	Variant (protein)	Inheritance	Age at onset	Epilepsy	Developmental/Behavioural features	EEG/Cerebral MRI (brief)	Other features	Treatment/Outcome
1	[5]	F	c.(388G>C)	p.Glu130Gln	de novo	Infancy	DEE/Infantile-spasms	Severe global DD, hypotonia	Abnormal EEG; MRI: hyper-intensity of the putamen and white matter, corpus callosum thinning	Hyperkinetic movements	ASM; ketogenic diet; persistent seizures
2	[5]	M	c.(401T>C)	p.Met134Thr	de novo	Infancy	DEE/Infantile spasms	Severe global DD	Abnormal EEG; MRI: diffuse cerebral volume loss	Dysphagia	ASM; poor control
3	[6]	M	c.(488T>C)	p.Leu163Pro	de novo	Adolescent	No epilepsy	Global DD; ID	Normal EEG: MRI: normal	Ventricular septal defect; caffè-au lait spot	Supportive therapies
4	[6]	F	c.(703G>A)	p.Ala235Thr	Parental data unavailable	Adolescent	No epilepsy	DD; ASD	NA	NA	Supportive/Surveillance
5	[8]	F	c.(385C>T)	p.Arg129Trp	de novo	Infancy	No epilepsy	DD/ID ± ASD	Abnormal EEG; MRI: hyper-intensity of the putamen and white matter, corpus callosum thinning	Central obesity, large central teeth, tapering fingers	Supportive therapies
6	[8]	M	c.(388G>C)	p.Glu130Gln	de novo	Infancy	DEE/Infantile spasms	DD/ID ± ASD	Abnormal EEG; MRI diffuse cerebral atrophy	Dysphagia, microcephaly	ASM; symptomatic
7	[8]	M	c.(804 C>A)	p.Arg268Trp	From the affected father (patient 8)	Childhood	No epilepsy	DD/ID ± ASD	NA	Inverted nipples; aggressive behaviour	Symptomatic
8	[8]	F	c.(804 C>A)	p.Arg268Trp	Parental data unavailable	Adult	No epilepsy	Mild ID	NA	NA	NA
9	[7]	F	c.(388G>C)	p.Glu130Gln	de novo	Infancy	DEE/Infantile spasms	Global DD; later ASD traits	EEG: multifocal t spikes; MRI: delayed myelination, hyperintensity in globi pallidi and central tegmental tracts	NA	Vigabatrin + high-dose prednisolone; Relapsing-remitting epilepsy
10	[9]	F	c.(388G>A)	p.Glu130Lys	de novo	infancy	Rett-like features	Severe DD, stereotypies, Rett-like phenotype	EEG/MRI abnormalities	NA	Symptomatic
11	[10]	F	c.(388G > C)	p.Glu130Gl)	de novo	infancy	DEE/infantile spasms	DD/ID; dysmorphic features	EEG: hypsarrhythmia/MRI: no structural abnormalities	NA	ASM; prednisone; partial response described

Table 2. Summary of published *NEUROD2* cases (2019–2024)*. (*continued*)

Case	Reference	Sex	Variant (cDNA)	Variant (protein)	Inheritance	Age at onset	Epilepsy	Developmental/Behavioural features	EEG/Cerebral MRI (brief)	Other features	Treatment/Outcome
12	Present case	F	c.(790G>A)	p.Ala264Thr	de novo	Neonatal	Neonatal hypotonia; neonatal seizures	DD/ASD trait	EEG poorly structured/MRI: subcortical white matter hyperintensity	Pelvic kidney; Dysphagia	Levetiracetam; symptomatic

ASD: autism spectrum disorder; ASM: anti-seizure medication; DD: developmental delay; DEE: developmental and epileptic encephalopathy; ID: intellectual disability; MRI: magnetic resonance imaging; NA: not available. *Two patients with chromosome deletion including the *NEUROD2* gene, described by Runge et al. [8], are not included in the table.

which offered emotional support and facilitated bonding. Following discharge, the family engaged in an early intervention program comprising physiotherapy and speech therapy.

Discussion

The present case of a newborn carrying a de novo heterozygous *NM_006160.4*:c.790G>A *NEUROD2* variant contributes to the expanding phenotypic and genotypic spectrum associated with *NEUROD2*-related neurodevelopmental disorders. Pathogenic variants in this gene have been linked to an autosomal dominant neurodevelopmental disorder characterized by variable expressivity, including developmental delay, ID, epilepsy, and behavioral abnormalities. In most reported cases, clinical manifestations begin during infancy, and variants tend to cluster in the bHLH domain, which is essential for DNA binding and dimerization [5–10]. Compared to previously described patients, our case shows an unusually early and severe neonatal presentation (Table 2). The patient manifested cardiorespiratory depression at birth, required respiratory support, and exhibited neurological abnormalities, including hypotonia, hyporeactivity, feeding difficulties, and early-onset seizures. Brain MRI revealed hyperintensity of the subcortical white matter, consistent with previous reports of white matter signal abnormalities and delayed myelination in *NEUROD2*-related disorders. However, in contrast to many previously published cases in which symptoms typically become apparent during early infancy, our patient displayed significant neurological impairment since birth, suggesting that the pathogenic effect of this variant may interfere with neuronal development at very early stages. Two additional clinical findings merit comment. The meconium-stained amniotic fluid is likely attributable to perinatal distress rather than a direct effect of the *NEUROD2* variant. The right pelvic kidney is not a recognized feature of *NEUROD2*-related disorders and is considered an incidental finding. A comparison of this case with prior literature highlights both shared and distinctive features (Table 2). Reported patients with *NEUROD2* variants frequently presented with developmental delay and epilepsy, often manifesting as infantile spasms during the first months of life (developmental epileptic encephalopathy). EEG abnormalities, such as multifocal spikes or hypsarrhythmia, were common. MRI findings were usually nonspecific, including white matter hyperintensity and delayed myelination, without major structural malformations. Treatment responses were variable; some patients showed temporary improvement with vigabatrin and steroids, whereas others required multiple antiepileptic drugs. Developmental outcomes were generally poor, with persistent cognitive and motor impairment, although severity varies considerably even among individuals carrying variants affecting similar domains [5–10]. The *NM_006160.4*:c.790G>A *NEUROD2* variant identified in this case has been rarely reported and is located further downstream from the most frequently mutated residues, which typically cluster in the core bHLH domain [1, 5–10]. Its presence in a highly conserved region, absence from population databases, de novo occurrence, and predicted functional impact support its classification as likely pathogenic. The severe and early neurological phenotype observed here may suggest that variants outside the canonical hotspot regions can still profoundly disrupt *NEUROD2* function. Previous functional analyses of

other *NEUROD2* variants demonstrated reduced transcriptional activity and impaired neuronal differentiation, supporting a loss-of-function mechanism as the pathogenic basis. Notably, the Ala235Thr variant reported by Mis et al. [6] shares key structural features with the present case—both are alanine substitutions in the C-terminal region outside the bHLH domain—yet is associated with a substantially milder phenotype, underscoring the complexity of genotype-phenotype correlations within this region. The clinical course of this patient also highlights the heterogeneity and variable expressivity of *NEUROD2*-related disorders. Some reported individuals have milder phenotypes with developmental delay but no epilepsy, whereas others present with severe epileptic encephalopathy and profound neurological deficits. This variability may be influenced by the location and nature of the variant, modifier genes, and epigenetic factors.

Overall, this case reinforces the importance of including *NEUROD2* in the differential diagnosis of neonates presenting with unexplained hypotonia, feeding difficulties, and early-onset seizures [1, 5–10, 14]. It also highlights the value of early exome sequencing in identifying de novo variants in genes associated with neurodevelopmental disorders, enabling accurate diagnosis and counseling [15–17].

Conclusions

In conclusion, we report a newborn with a de novo heterozygous *NM_006160.4*:c.790G>A *NEUROD2* variant presenting with neonatal onset, thereby expanding both the phenotypic and mutational spectrum of *NEUROD2*-related neurodevelopmental disorders. This case introduces a novel missense change associated with severe early presentation, demonstrating that pathogenic *NEUROD2* variants can exert profound clinical effects even when located outside the most well-characterized functional domains. As additional cases are identified, further refinement of genotype-phenotype correlations, improved prognostic accuracy, and the development of targeted therapeutic strategies will become increasingly feasible for this rare but clinically significant disorder.

Abbreviations

ASD: autism spectrum disorder

bHLH: basic helix-loop-helix

EEG: electroencephalogram

ID: intellectual disability

MRI: magnetic resonance imaging

Declarations

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Author contributions

LL: Conceptualization, Investigation, Formal analysis, Visualization, Writing—original draft, Writing—review & editing. IG: Investigation, Writing—original draft, Writing—review & editing. CR: Investigation, Writing—original draft, Writing—review & editing. NB: Investigation, Writing—original draft, Writing—review & editing. MB: Investigation, Writing—original draft, Writing—review & editing. VC: Investigation, Writing—original draft, Writing—review & editing. EB: Investigation, Writing—original draft, Writing—review & editing. MP: Validation, Writing—review & editing, Supervision. AB: Validation, Writing—review & editing, Supervision. All authors read and approved the submitted version.

Conflicts of interest

The authors declare that there are no conflicts of interest.

Ethical approval

This study was conducted in accordance with the principles of the Declaration of Helsinki. According to institutional policy and Italian national regulations, formal ethics committee review is not mandatory for retrospective case reports describing routine clinical practice, provided no experimental interventions are performed, and no additional biological samples are collected beyond those obtained for standard diagnostic purposes. All data presented in this report were obtained as part of routine clinical care.

Consent to participate

Informed consent to participate in the study was obtained from the patient's parents.

Consent to publication

Written informed consent for publication of clinical details and genetic findings was obtained from the patient's parents. Patient data were anonymized and handled in strict compliance with the European Union General Data Protection Regulation (GDPR) (EU 2016/679) and Italian privacy law.

Availability of data and materials

The datasets supporting the findings of this study are available from the corresponding author upon reasonable request.

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