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Short communication

Self-limited Neonatal Epilepsy associated with global developmental delay and high-threshold sensory profile: A novel *KCNQ3 de novo* variant unusually located in the voltage sensor S4 segment of the Kv7.3 channel subunit

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ABSTRACT

Background: Pathogenic variants in *KCNQ2* and *KCNQ3* are established causes of self-limited neonatal epilepsy (SeLNE). While over 300 *KCNQ2* variants have been reported, the number of *KCNQ3* variants is increasing, with an expanding phenotypic spectrum including Developmental and Epileptic Encephalopathy (DEE), Intellectual Disability (ID), and Autism Spectrum Disorder (ASD). In a single case study, we report the clinical, neuropsychological, and molecular characterization of a novel *KCNQ3* variant to refine genotype–phenotype correlations.
Methods: Deep phenotyping included standardized neurodevelopmental assessment (BSID-III) and sensory profiling (Toddler Sensory Profile-2). Genetic analysis was performed using a targeted Next-Generation Sequencing panel with Sanger confirmation of candidate variants. Variant interpretation followed ACMG criteria. Structural impact was assessed using AlphaFold-based modelling and FoldX in silico mutagenesis; pathogenicity was further evaluated with PolyPhen-2, SIFT, and MutationTaster.
Results: BSID-III showed global developmental delay (cognitive 75, language 63, motor 67, socio-emotional 75). Sensory assessment identified a high-threshold profile with increased Seeking and Registration patterns across modalities. NGS identified a de novo heterozygous *KCNQ3* variant (NM_004519.4:c.730G>A; p.Gly244Ser), absent from population databases. The variant affects a highly conserved residue (PhyloP100=7.9) within the S4 voltage-sensing domain. Structural modelling predicted destabilization (ΔΔG=+1.56 kcal/mol). In silico tools consistently supported a deleterious effect; ACMG classification was likely pathogenic (PS2, PM2, PP3). Seizures were effectively controlled with levetiracetam.
Conclusion: This study expands the *KCNQ3* mutational spectrum and supports the contribution of S4 domain variants to combined epileptic and neurodevelopmental phenotypes, highlighting the value of integrated deep phenotyping and molecular characterization.

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1. Introduction

Self-limited Neonatal Epilepsy (SeLNE) is a rare autosomal dominant genetic disorder affecting 5.3 newborns per 100,000 live births [1]. It is characterized by brief, often clustered, focal or generalized tonic seizures which can alternate sides from seizure to seizure, followed by apnoea and clonic movements with oculofacial features often associated with autonomic signs. The seizures begin between 2 and 7 days of life and spontaneously disappear within the first year of life.

Pathogenic variants of *KCNQ2/Q3*, encoding Kv7.2 and Kv7.3 voltage-gated potassium channel subunits, have been identified as the cause of Self-limited Neonatal Epilepsy (SeLNE), since the channels (Kv) regulate neuronal excitability by generating the sub-threshold K+ current (M-current).

However, the *KCNQ3* variants described so far are few compared to the over 300 pathogenic *KCNQ2* variants reported [2]. Although *KCNQ2/Q3* variants have been identified mainly in families with SeLNE, recently these variants have been reported also in subjects with Developmental Epileptic Encephalopathy (DEE), Intellectual Disabilities (ID), and Autism Spectrum Disorder (ASD) [3–6]. Furthermore, the above phenotypes are often the results of a qualitative clinical observations lacking of the quantitative measurements of standardizated neuropsychological assessment.

Since it is difficult to predict the cognitive and behavior outcome in the life-span a formal neuropsychological assessment should be a crucial data to better define the genotype-phenotype correlation. However, available evidence delineates a clinically heterogeneous phenotype predominantly marked by ID, ASD with variable epileptic involvement. Interestingly, it has been reported that a subset of individuals with neurodevelopmental disorders associated with *KCNQ3* (*KCNQ3*-NDD) present *de novo* pathogenic missense variants located in S4 domain. To date, only 5 pathogenic variants of KCNQ3, affecting the S4 domain, have been reported. In particular, functional studies have demonstrated a GoF effect in 4 variants and only one variant with a LoF effect [7]. The variant we report affects the Gly244Ser amino acid residue of the protein, thus located near the functionally studied M240R (LoF) residue and associated with a SeLNE and "language delay" phenotype. Our finding represents the second pathogenic novel variant affecting the bottom of the S4 domain of KCNQ3 that shares the SeLNE phenotype but expands the associated cognitive impairments from a language delay to a global developmental delay (GDD).

2. Case study

Written informed consent for publication was obtained from the parents.

The proband, a 7-month-old male, was the second offspring of healthy consanguineous parents (second-degree cousins), with two previous pregnancies resulting in miscarriages. Following a complicated pregnancy, due to bleeding and placental abruption, he was born at 38 + 6 weeks of gestation by elective Caesarean section. The Apgar scores at birth were 9 and 10 (1′−5′). His weight was 2860 g (36th centile), length 48 cm (44th centile), and head circumference 33 cm (63th centile). He has a healthy older sister, aged 16-years-old.

On day 5, he exhibited a cluster of brief focal ictal events (3–5/day), switching sides from one seizure to another, characterized by staring, head and eyes lateralization, stertorous breath, upper limb jerkings, and perioral cyanosis. As paroxysmal events recurred, on day 6, the patient was referred to our attention. On admission, the newborn showed mild axial hypotonia and reduced spontaneous motility. Complete blood count, procalcitonin, CRP, blood cultures, glucose, serum electrolytes, neonatal metabolic screening, and a head ultrasound were normal.

A continuous EEG showed several ictal electroclinical patterns characterized by focal low amplitude and high frequency activity evolving into sharp-wave activity with decreasing frequency and increasing amplitude lasting 15 s, and switching sides from one seizure to another (Fig. 1A,B).

On day 7, he was given a bolus of phenobarbital (PB), 20 mg/kg, followed by 2.5 mg/Kg/day twice daily, resulting in seizure control from day 8. On day 20, when the EEGs and the seizure features appeared consistent with the diagnosis of SeLNE, we switched PB to Levetiracetam (LEV) at 15 mg/Kg/day, due to the latter's fewer side effects and the growing consensus regarding its efficacy. On the same day, the infant was discharged, and he has not experienced any further seizures to date, at the age of 15 months.

Brain MRI, performed at 1 month of age, didn't show any abnormalities.

Interictal EEG performed at 5 months showed a brief sequence of high-amplitude sharp waves in the left frontal-central regions (Fig. 1C).

At 7 months of age, the child was alert to his environment and showed a good level of attention and cooperation with a positive emotional tone, adequate adaptability, and sociability. He was consolidating the sitting position. The vocalisation was spontaneous and in response to the examiner, and he showed adequate eye contact and gross manipulation of objects. At 13 months, the child no longer had seizures even on a low dose of LEV, but he was unable to stand up, and he spoke one word. Therefore, the child underwent a formal neuropsychological assessment, including standardised testing for neurocognitive development and sensory processing skills performed by a trained psychologist blind to the child's medical history.

Neurocognitive development, assessed using the Bayley Scales of Infant and Toddler Development, Third Edition (BSID-III) [8], showed a global developmental delay. Significant delay were found in all the domains tested: cognitive (composite score = 75), language (composite score = 63) motor (composite score = 67), and in the socio-emotional behavior (composite score = 75) (see Table 1).

The sensory profile, assessed using the Toddler Sensory Profile-2 [9], showed elevated scores in both the Seeking (percentile = 90–99) and Registration (percentile = 96–99) quadrants (Table 2). In particular, the Seeking pattern fell within the "More than Others" range, reflecting an increased tendency to actively pursue sensory input, whereas the Registration pattern was classified as "Much More than Others", indicating a reduced passive detection of sensory stimuli. This pattern reflects a high-threshold sensory profile, characterized by the coexistence of both active (Seeking) and passive (Registration) self-regulation strategies. Consistent with the quadrant findings, elevated scores were also observed across several sensory system and behavioral responses sections. In particular, Auditory, Visual and Tactile systems fell within the "more than others" ranges whereas Motion fell within the "much more than others" range indicating generalized sensory processing alterations rather than modality-specific alterations.

Overall, the sensory profile of our patient is consistent with a globally high sensory threshold influencing both perception and behavior in everyday contexts.

As the clinical data suggested a SeLNE diagnosis, we performed a molecular genetic analysis. A Next-Generation-Sequencing (NGS) panel of 1400 genes, including *KCNQ2* and *KCNQ3*, revealed a *de novo* heterozygous *KCNQ3* NM_004519.4:c.730G>A (p.(Gly244Ser)) variant, confirmed by Sanger sequencing. The variant is absent from local and public databases (GnomAD, ClinVar, 1000 Genomes Project database), and the allele frequency is very low ([ExAC] 0.00001). Structure-based modelling, using the AlphaFold model of KCNQ3, revealed the involvement of a conserved residue (PhyloP100: 7.9) in the voltage sensor S4 segment of the Kv7.3 channel subunit. In the wild-type structure, glycine (4 atoms) lacks a side chain and forms minimal local contacts. Substitution with serine introduces a polar side chain (6 atoms), altering the local packing environment. FoldX in silico mutagenesis predicted a destabilizing effect ($\Delta\Delta G = +1.56$ kcal/mol), supporting structural perturbation in this functionally constrained region of the voltage-sensing helix. (Fig. 2). The missense mutation Gly244Ser is predicted to be "probably damaging", "deleterious", and "disease-causing" based on the three in silico models, PolyPhen2, SIFT, and

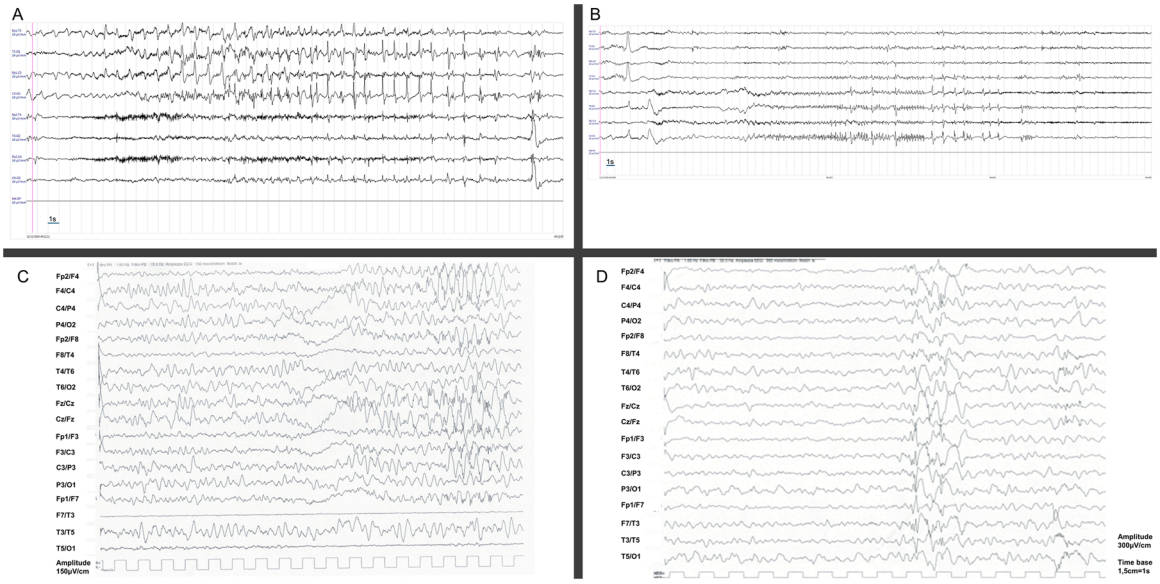

Fig. 1. The EEG shows several ictal electroclinical patterns characterized by focal low amplitude and high frequency activity evolving into sharp-wave activity with decreasing frequency and increasing amplitude lasting 40 s, and switching sides (**A** left frontal-central regions) from one seizure to another (**B** right frontal-central regions). **C**) The interictal EEG, performed at 5 months, shows a brief sequence of high-amplitude sharp waves in the left frontal-central regions. **D**) interictal EEG, performed at 5 months and 15 days in a patient with Self-Limited Familial Infantile Epilepsy.

Table 1
Scores on the Bayley Scales of Infant and Toddler Development, Third Edition (BSID-III).

Domain	Subscale	Patient	Interpretation
Cognition		75§	moderately low scores
Language		63§	severely low scores
	receptive	1*	
	expressive	6*	
Motor		67§	severely low scores
	fine	6*	
	gross	4*	
Socio-emotional behavior		75§	moderately low scores

Legend: § = composite scores (mean = 100; sd = 15); * = scaled scores (mean = 10; sd = 3).

Table 2
Scores on the Infant/Toddler Sensory Profile-2 (ITSP).

Domain	Subscale	Patient's raw scores	Percentile	Interpretation
Sensory Quadrant				
	Seeking	35	90–99	More than the others
	Avoidance	15	11–86	Like the rest
	Sensitivity	21	11–85	Like the rest
	Registration	25	96–99	Much more than the others
Sensory Systems				
	General	20	16–86	Like the rest
	Auditory	15	84–95	More than the others
	Visual	22	87–99	More than the others
	Tactile	13	84–96	More than the others
	Motion	22	100	Much more than the others
	Oral	7	4–87	Like the rest
Behavioral Responses Associated with Sensory Processing		14	89–96	More than the others

Mutation Taster, respectively.

According to ACMG criteria PS2, PM2, PP3, with a score of 6 points (6 pathogenic-0 benign), the variant was predicted as "Likely pathogenic" by Germline.

3. Discussion

The genetic heterogeneity, the wide distribution of the voltage-gated K+ channel (Kvs) family, and their consequent great functional diversity are considerably increasing our knowledge of their role in many diseases, including the nervous system. Simultaneously, research in this field has increased, involving different disciplines such as neuroscience, genetics, neurophysiology, and pharmacology, which is showing great interest in identifying selective therapeutic molecules.

Recent experimental pharmacological studies on different classes of Kv7 modulators suggested that the β-hydroxybutyrate (BHB) restored Kv7.2 +Kv7.3 M240R currents to those of wild-type [7]. Further, it has been documented the efficacy of Quinidine and Fluoxetine in blocking all GoF (Gain of Function) in KCNT2 variants. Whereas Loxapine and Riluzole were effective in activating some LoF (Loss of Function) variants while blocking others [10]. Interestingly, also the effects of some PUFAs on KCNQ3 variants have been investigated, specifically the derived compound N-arachidonoyl amine (NAA+). NAA+ partially reversed the effects of the GoF R227Q mutation, whereas N-arachidonoyl taurine (NAT-) reversed the effects of the LoF R236C mutation [2].

The most pathogenic variants in KCNQ3 identified in typical SeLNE are commonly LoF missense variants involving the pore region (S5, S6, and intervening loop) [7].

Recently, two de novo missense mutations, located in the voltage sensor S4 segment of the Kv7.3, have been identified in eleven children with GDD, ASD, and frequent sleep-activated multifocal epileptiform discharges. The mutations involve the first two arginine residues (R1: R227Q and R2: R230C/S/H) whose neutralization tends to destabilize the resting voltage-sensitive domain configuration, thereby causing constitutive channel activation and resulting in strong GoF effects [11].

Conversely, a novel Kv7.3 (M240R) variant, located in the voltage-sensing S4 transmembrane segment of Kv7.3 subunit, was reported as responsible for disease in a family with SeLNE (one of whom also had a

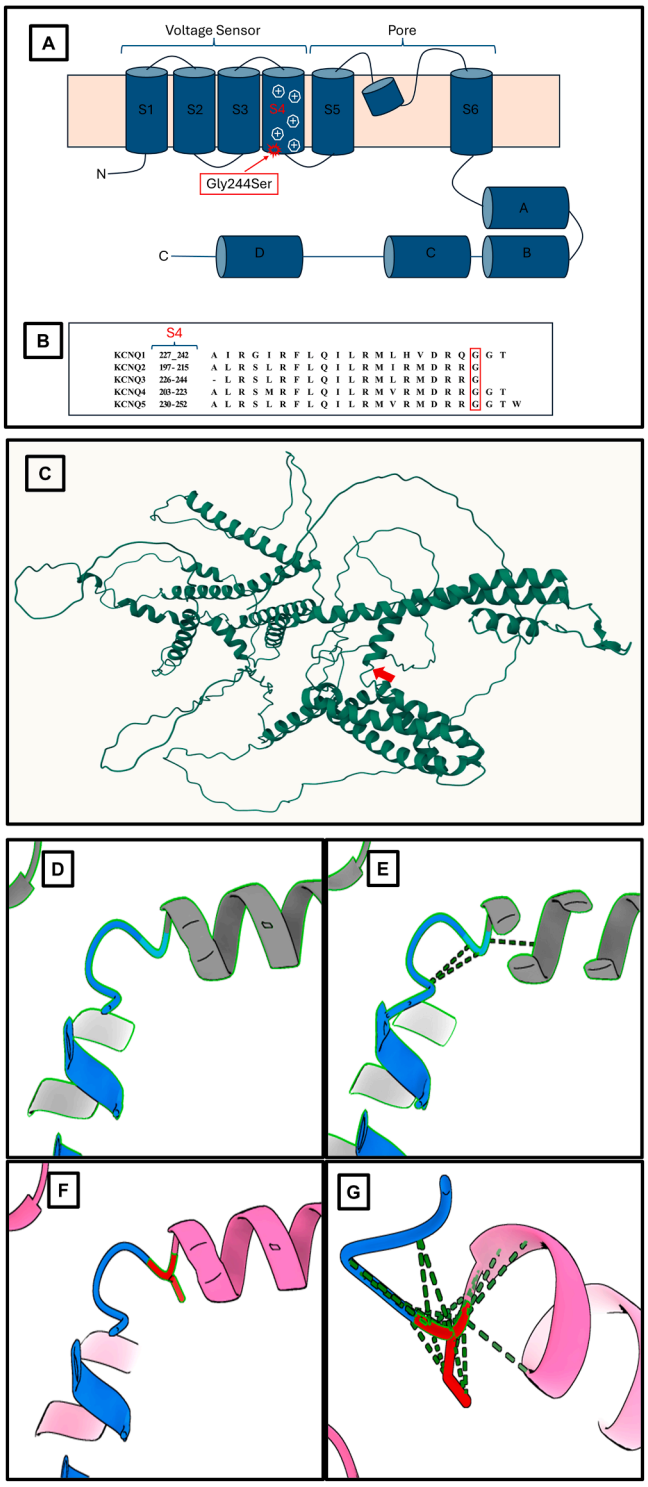

Fig. 2. **A**) Schematic representation of a single Kv7.3 subunit, with the arrow indicating the position of the mutation under investigation (highlighted in red). The letters A to D represent the four putative alpha-helical domains identified in the C-terminal region of Kv7.3. **B**) shows the sequence alignment of the S4 segments of the specified Kv subunits, and the conservation of the amino acid residues. The red box highlights the mutation. **C**) Global structural representation of KCNQ3 based on the AlphaFold model. A red arrow indicates the position of Gly244. **D**) Local structural view of the wild-type Gly244 residue, shown in cyan stick representation. Glycine contains only four atoms and lacks a side chain. S4 domain shown in blue. **E**) Local interaction environment surrounding wild-type Gly244 is shown in dashed green lines. **F**) Structural model of the p.Gly244Ser mutant, generated by FoldX-based in silico mutagenesis. The substituted Ser244 residue is displayed in red stick representation, highlighting the introduction of a side chain that increases the number of atoms at this position from four (Gly244) to six (Ser244) and introduces additional steric bulk. **G**) Local interaction network of Ser244 in the mutant structure is shown in dashed green lines. The newly introduced serine side chain establishes additional contacts with surrounding residues, altering local packing at the C-terminal boundary of the S4 helix (blue). Energetic analysis using FoldX mutagenesis predicted a destabilizing effect for the p.Gly244Ser substitution ($\Delta\Delta G = +1.56$ kcal/mol).

speech delay). The mutation, adding a further positively charged residue at the bottom of S4 segment between the R5 (R239) and R6 (R242) residues, resulted in a significant decrease in channel sensitivity to voltage of approximately 10 mV and destabilizing the activated conformation of the voltage sensor, thereby preventing pore opening, resulting in LoF effects [7].

Our Gly244Ser finding documents the second novel mutation of the bottom S4 causing a SeLNE phenotype. The mutation introduces an uncharged polar residue into the voltage-sensing S4 helix of the Kv7.3 subunits, following R5 and R6, and neighboring the pathogenic mutations R236C and M240R previously identified and functionally characterized.

The lack of a functional study of the Gly244Ser mutation is a limitation of the study. However, it is hypothesized that the change of residue polarity (Gly apolar – Ser polar) and of different size (Gly <Ser) is enough to destabilize the activated conformation of the voltage sensor, resulting in structural perturbation of the voltage-sensing domain. These findings strengthen the view [7] that the bottom of the S4 helix is a region very sensitive to substitutions of amino acid residues that, by modifying their charge, polarity or size, result in a LoF of the channel [11]. However, the current clinical profile of our patient reveals a SeLNE, GDD and a high-threshold sensory profile suggesting that environmental sensory input often fails to reach the threshold required for consistent behavioral registration unless it is sufficiently intense or salient. Therefore, the proband shows clinical characteristics of the LoF (M240R) profiles. Furthermore, we do not rule out that the cognitive-behavioral profile may also depend, to some extent, on a possible modifying effect of other genetic factors related to the consanguinity of the patient's parents.

The analysis of the data previously reported and those present in this study suggests that Levetiracetam, like sodium channel blockers, is proving to be effective in controlling seizures. However, the duration of treatment remains unresolved, considering that relapses in the long term are not rare [12]. Some personal clinical and electroencephalographic observations lead us to believe that the withdrawal of an anti-seizure treatment before 6 months is not recommended.

Notably, the onset of Self-Limited Familial Infantile Epilepsy at 5 months and 15 days in one of our patients with KCNQ3- Ser617Thr mutation is consistent with the above considerations (Fig. 1D) [13]. Furthermore, the comparison of his interictal EEG with that of the patient here reported (SeLNE), at the age of 5–6 months, showed similar epileptogenic abnormalities. Suggesting that at least up to this age, the deficit of the KCNQ3 channel function has not been sufficiently compensated by the inhibitory system still in ongoing maturation.

CRediT authorship contribution statement

Giuseppe Donato Mangano: Conceptualization, Supervision, Methodology, Formal analysis, Data curation, Writing – original draft, Writing – review & editing. **Gabriele Di Pasquale:** Writing – original draft. **Francesco Fabrizio Comisi:** Writing – original draft. **Vita Maria Angileri:** Investigation, Resources. **Vincenzo Antona:** Investigation, Resources. **Vincenzo Duca:** Investigation, Resources. **Vincenzo Salpietro:** Conceptualization, Writing – review & editing. **Giuseppa Renata Mangano:** Conceptualization, Supervision, Formal analysis, Data curation.

Declaration of competing interest

Giuseppe Donato Mangano, Gabriele Di Pasquale, Francesco Comisi, Vita Maria Angileri, Vincenzo Antona, Vincenzo Duca, Vincenzo Salpietro, and Giuseppa Renata Mangano declare no conflicts of interest.

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