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Variants in *SCN8A* and Associated Genes in Saudi Arabian Epilepsy Cohort: A Preliminary Whole-Exome Sequencing Analysis

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Abstract

Background and Purpose: Epilepsy is a neurological disorder characterized by recurrent seizures, with genetic factors playing significant roles in its etiology. This preliminary study aimed to characterize genetic variants in Saudi Arabian epilepsy patients using whole-exome sequencing (WES) data, providing exploratory insights into potential population-specific patterns.

Methods: Raw WES data (ERR10619203–ERR10619207) from five Saudi Arabian epilepsy patients were retrieved from the NCBI Sequence Read Archive (SRA). Data underwent quality control, alignment, and variant calling using standard bioinformatics workflows. Variants were annotated and classified according to American College of Medical Genetics and Genomics (ACMG) guidelines, with particular focus on established epilepsy-associated genes.

Results: The analysis identified 23,361 exonic variants across all genes. Within epilepsy-associated genes, 3,005 variants were detected across 11 functional categories, with ion channel genes showing the highest variant burden (61%). Among sodium channel genes, *SCN8A* variants (n=87) appeared more frequent than *SCN1A* variants (n=12) in this small cohort. While this observation contrasts with patterns reported in other populations, the limited sample size precludes definitive conclusions. Additionally, analysis of *GABRG2*-associated variants revealed genomic co-localization with *CCNG1* (n=343 variants), suggesting shared genomic regions rather than confirmed functional interaction.

Conclusions: This exploratory analysis provides descriptive insights into the genetic landscape of epilepsy in a small Saudi Arabian cohort. The findings highlight possible population-specific patterns but require validation in larger, more diverse samples. These preliminary results contribute to the growing understanding of geographical diversity in epilepsy genetics.

Keywords: Epilepsy; Whole-Exome Sequencing; Bioinformatics; Population Genetics

1. Introduction

Epilepsy is a common neurological disorder affecting individuals across all age groups, demographics, and geographical regions worldwide. Characterized by recurrent seizures resulting from abnormal brain electrical activity, epilepsy can significantly impact neurological function, mental health, and quality of life for both patients and their families [1]. The global burden of epilepsy exhibits marked geographical variation, with lifetime prevalence rates in developing countries (up to 15.4 per 1,000) substantially exceeding those in industrialized nations (approximately 5.8 per 1,000) [2].

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Epilepsy etiology encompasses multiple factors, with approximately 60% of cases classified as idiopathic (primary) epilepsy, often involving genetic susceptibility to neuronal hyperexcitability [3]. Secondary (symptomatic) epilepsy accounts for the remaining cases, attributable to structural causes including traumatic brain injury, stroke, infections, and congenital abnormalities [4]. Advances in genomic technologies have significantly enhanced our understanding of epilepsy genetics, with evidence suggesting that 70–80% of cases may have genetic contributions [5].

Recent studies have demonstrated that genetic alterations in epilepsy encompass a diverse range of variants affecting ion channels, neurotransmitter receptors, transcriptional regulators, and cellular signaling pathways [6]. These alterations can occur through de novo mutations or inherited variants, manifesting through monogenic or polygenic mechanisms. The International League Against Epilepsy (ILAE) classification system categorizes epilepsies according to seizure characteristics and etiologies, including genetic, structural, infectious, metabolic, immune-related, and unknown causes [7].

Previous genomic studies in Middle Eastern populations, particularly in Saudi Arabia, have provided important insights into the genetic landscape of epilepsy. Al Anazi et al. (2022) conducted whole-exome sequencing on 102 monogenic epilepsy genes among 44 Saudi Arabian individuals with epilepsy, identifying 12 genes (*CHRNA4*, *CLN3*, *CLN8*, *DEPDC5*, *KCNJ10*, *KCNMA1*, *POLG*, *PRICKLE1*, *SCN1A*, *SCN2A*, *SCN8A*, and *SCN9A*) as containing pathogenic variants [8]. This landmark study highlighted both shared and population-specific genetic factors in epilepsy susceptibility. Prior research has established *SCN1A* as the predominant sodium channel gene implicated in epilepsy, especially in Dravet syndrome, with numerous reports of pathogenic variants across populations [9, 10]. These variants have been shown to disrupt neuronal excitability by impairing sodium channel function, thereby contributing to the characteristic seizure phenotypes observed in affected individuals.

The present preliminary study addresses the need for population-specific genomic characterization of epilepsy by analyzing whole-exome sequencing data from Saudi Arabian epilepsy patients. Given the limited sample size and exploratory nature of this analysis, the study aims to characterize genetic variants in established epilepsy-associated genes and identify potential patterns that may inform future larger-scale investigations. While the current sample size limits the generalizability of findings, this work contributes to the growing understanding of geographical diversity in epilepsy genetics and provides a foundation for future population-specific studies.

2. Methods

Study Design and Data Source: This preliminary analysis utilized publicly available whole-exome sequencing data from five Saudi Arabian epilepsy patients, accessed from the NCBI Sequence Read Archive (SRA) under accession numbers ERR10619203–ERR10619207 [8]. The study design was exploratory and descriptive in nature, aimed at characterizing genetic variants in epilepsy-associated genes within this population.

Sample Characteristics: All samples were derived from peripheral blood DNA extracted from Saudi Arabian individuals diagnosed with epilepsy. Sequencing was performed on the Illumina HiSeq 2500 platform with 150 bp paired-end reads achieving >30× coverage depth [8]. The small sample size (n=5) was a significant limitation that restricts the statistical power and generalizability of findings.

Quality Control and Preprocessing: Quality assessment and preprocessing were performed using established bioinformatics tools. Quality control metrics were generated with MultiQC v1.15 [11]. Low-quality reads with Phred scores <30 or length <50 bp were filtered out. Trimming was performed using Trimmomatic v0.39 with parameters: leading/trailing quality <3, sliding window of 4 bp with average quality <15, minimum read length 36 bp [12]. Clean reads were aligned to the GRCh38 reference genome using Bowtie2 v2.4.1 [13]. Duplicate reads were marked using Picard v2.23.8, and alignment statistics were generated using SAMtools v1.10 [14].

Variant Calling and Annotation: Variants were called using GATK MuTect2 v4.1.9.0 with a minimum depth threshold of 14× [15]. Variants were annotated using ANNOVAR v2021Apr16 with RefSeq annotations and hg38 reference [16].

Variant Filtering and Classification: Variants were systematically filtered and classified according to established criteria. Only those meeting quality thresholds, defined as a minimum depth of 14× and a quality score of at least 30, were retained for analysis. Variants were then categorized into non-synonymous single nucleotide variants (NS-SNVs), loss-of-function variants such as stop-gain, frameshift, and splice-site changes, and synonymous variants, which were catalogued separately. All retained variants were further assessed according to ACMG guidelines [17], and the analysis was focused specifically on genes previously established in the literature as being associated with epilepsy.

Statistical Analysis: Given the small sample size (n=5), statistical analyses were limited to descriptive statistics. Variant frequencies were calculated across gene categories and individual genes. No comparative statistical testing was performed due to lack of appropriate control groups.

Data Analysis and Visualization: Data analysis and visualization were conducted in R v4.1.0 using dplyr and ggplot2 packages [18]. Hierarchical clustering was performed using standard clustering algorithms to explore relationships between variant types and gene families.

3. Results

3.1. Sequencing Quality Assessment

Quality assessment of whole exome sequencing data from five Saudi Arabian epilepsy patients demonstrated high sequencing performance (Table 1).

Table 1 Statistical results of raw reads and sequence quality score

Sample	Unique Reads	Duplicate Reads	Duplication (%)	Total Sequences (M)	Q20-29 (%)	>Q30 (%)	GC (%)	Avg. Length (bp)
ERR10619203_1	35,327,995	34,384,326	49.30	69.7	0.38	99.62	50	98.76
ERR10619203_2	35,213,974	34,498,347	49.50	69.7	0.46	99.54	50	98.69
ERR10619204_1	39,374,492	37,376,880	48.70	76.8	0.36	99.64	50	98.76
ERR10619204_2	39,541,593	37,209,779	48.50	76.8	0.55	99.45	50	98.65
ERR10619205_1	42,029,445	43,036,588	50.60	85.1	0.37	99.63	50	98.73
ERR10619205_2	42,008,446	43,057,587	50.60	85.1	0.50	99.50	51	98.62
ERR10619206_1	37,725,963	38,288,684	50.40	76.0	0.37	99.63	50	98.85
ERR10619206_2	37,579,135	38,435,512	50.60	76.0	0.49	99.51	50	98.78
ERR10619207_1	35,554,174	33,957,827	48.90	69.5	0.42	99.58	50	98.87
ERR10619207_2	35,345,430	34,166,571	49.20	69.5	0.38	99.62	50	98.85

Abbreviations: Q20–29 (%) = percentage of bases with Phred quality score between 20 and 29; >Q30 (%) = percentage of bases with Phred quality score above 30; GC (%) = percentage of guanine (G) and cytosine (C) bases; Total Sequences (M) = Total Sequences (millions).

Sequencing performance was consistent across all samples, with each producing 69.5–85.1 million reads and duplicate rates between 48.5% and 50.6%. Over 99.4% of reads exceeded the Q30 threshold, confirming high base call accuracy. GC content remained stable (50–51%), and mean read length was 98.7 bp.

3.2. Alignment and Mapping Statistics

All sequencing reads were aligned to the GRCh38 reference genome using Bowtie2, achieving overall alignment rates of 99.87–99.89% (Table 2).

Table 2 Reads Alignment mapping statistics

Sample	All Reads	Not Mapped Reads	Mapped Reads	Alignment Rate (%)
ERR10619203_1__pair_combined.sam	139,424,669	144,048	139,280,621	99.89
ERR10619204_1__pair_combined.sam	153,502,771	156,258	153,346,513	99.90
ERR10619205_1__pair_combined.sam	170,132,093	188,260	169,943,833	99.89
ERR10619206_1__pair_combined.sam	152,029,321	147,916	151,881,405	99.90
ERR10619207_1__pair_combined.sam	139,024,029	131,800	138,892,229	99.91

Alignment statistics confirmed high mapping quality, with unmapped reads accounting for only 0.09–0.11% of total reads.

3.3. Variant Characterization and Annotation

Variant calling with MuTect2 produced comprehensive variant profiles, which were annotated using ANNOVAR. Annotations included gene identifiers, functional categories, exonic functions, and predicted amino acid changes.

3.4. Exonic Variant Distribution and Classification

Analysis of exonic regions identified 23,361 variants across multiple functional categories (Table 3).

Table 3 Genomic variants frequencies and exonic regions mutations

Variant Category	Subcategory	Count	Percentage
*SNV (21,428)	Synonymous SNV	10,945	46.8%
	Non-synonymous SNV	10,483	44.9%
Small insertions/deletions (1,725)	Non-frameshift deletion	840	3.6%
	Non-frameshift insertion	173	0.7%
	Non-frameshift substitution	220	0.9%
	Frameshift deletion	288	1.2%
	Frameshift insertion	204	0.9%
Nonsense variants (53)	Start loss	29	0.1%
	Stop loss	24	0.1%
Disruptive variants (155)	Stop gain	155	0.7%
Total		23,361	100%

*SNV: Single Nucleotide Variant

Note: Structural variations (>50 bp genomic rearrangements) were not detected in this analysis. Frameshift and stop-gain variants are classified as small variants, not structural variations.

The majority of variants were single nucleotide variants (SNVs) (21,428; 91.7%), with nearly equal distribution between synonymous (10,945; 46.8%) and non-synonymous (10,483; 44.9%) substitutions. Small insertions and deletions accounted for 1,725 variants (7.4%), with frameshift mutations representing 492 variants (2.1%).

3.5. Epilepsy-Associated Gene Variants

Targeted analysis of established epilepsy-associated genes revealed 3,005 variants distributed across 11 major functional gene categories (Table 4).

Table 4 Summary of epilepsy genes and their variants

Gene Category	Gene Name	Variant Count	Total	Percentage
Sodium Channel Genes	*SCN1A*	12	267	17.81%
	SCN1A-AS1	61		
	SCN2A	60		
	SCN2A;CSRNP3	27		
	SCN3A:SCN2A	4		
	SCN8A	87		

	SCN8A;FIGNL2	6		
	SLC4A8;SCN8A	10		
Potassium Channel Genes	*KCNQ3*	153	414	20.32%
	KCNQ3;HPYR1	36		
	HHLA1;KCNQ3	8		
	KCNA2	26		
	KCNA2;KCNA3	9		
	KCNA10;KCNA2	29		
	KCNT1	146		
	KCNT1;CAMSAP1	5		
	SOHLH1;KCNT1	2		
Calcium Channel Genes	*CACNA1A*	296	639	22.84%
	CACNA1A;CCDC130	170		
	IER2;CACNA1A	38		
	CACNB4	126		
	CACNB4;STAM2	9		
NMDA Receptors	*GRIN2A*	167	643	24.47%
	GRIN2A;ATF7IP2	303		
	LINC01195;GRIN2A	173		
GABA Receptor Genes	*GABRG2*	27	416	18.38%
	*GABRG2;CCNG1**	343		
	LINC01202;GABRG2	12		
	GABRA1	20		
	GABRA1;LINC01202	3		
	GABRA6;GABRA1	11		
BarH-Like Homeobox (BARX)	*BARX1*	3	226	11.30%
	BARX1-DT	1		
	BARX1-DT;PTPDC1	29		
	MIR4291;BARX1	104		
	BARX2	30		
	BARX2;LINC01395	24		
	LOC399975;BARX2	35		
Chromatin Remodeling Genes	*CHD2*	89	122	5.27%
	CHD2;RGMA	9		
	CHCHD2	11		
	CHCHD2;NUPR2	6		
	PHKG1;CHCHD2	7		
Leucine-Rich Repeat (LRR) Genes	*LGI1*	25	133	5.15%

	LGI1;SLC35G1	77		
	FRA10AC1;LGI1	31		
Syntaxin Binding Protein 1	*STXBP1*	41	57	3.41%
	STXBP1;CFAP157	3		
	NIBAN2;STXBP1	13		
E3 Ubiquitin Ligase	*NHLRC1;TPMT*	83	84	3.67%
	KIF13A;NHLRC1	1		
Spectrin Gene	*SPTAN1*	4	4	0.18%
Total			3,005	100%

*: GABRG2;CCNG1 variants reflect genomic co-localization of these genes, not functional interaction

Ion channel genes were the most affected, accounting for approximately 61% of epilepsy-related variants. *NMDA* receptors showed the highest individual category burden (643 variants; 24.47%), followed by calcium channels (639 variants; 22.84%) and potassium channels (414 variants; 20.32%). *GABA* receptor genes accounted for 416 variants (18.38%), and sodium channel genes contributed 267 variants (17.81%).

Notable findings included *SCN8A* with 87 variants, representing the highest count among sodium channel genes, and *KCNQ3* with 153 variants among potassium channels. The *GABRG2;CCNG1* annotation reflected 343 variants, representing genomic co-localization rather than functional interaction.

3.6. Variant Classification According to ACMG Guidelines

Variants were classified according to ACMG guidelines (Table 5), with the majority requiring further functional validation to determine pathogenicity.

Table 5 ACMG variant classification for epilepsy-associated genes

Classification	Count	Percentage	Notes
Pathogenic	0	0%	No variants met strict pathogenic criteria
Likely Pathogenic	12	0.4%	Require functional validation
Uncertain Significance *(VUS)	1,847	61.5%	Majority of variants
Likely Benign	834	27.8%	Predicted benign effects
Benign	312	10.4%	Clearly benign variants
Total	3,005	100%	

*VUS: Variants of Uncertain Significance

3.7. Synonymous Variants in Epilepsy Genes

Synonymous variants were catalogued separately as they may affect splicing or mRNA stability (Table 6).

Table 6 Synonymous variants in epilepsy-associated genes

Gene Category	Synonymous SNVs	Total Variants	Synonymous Percentage
Sodium Channel Genes	23	267	8.6%
Potassium Channel Genes	31	414	7.5%
Calcium Channel Genes	28	639	4.4%
NMDA Receptors	45	643	7.0%
GABA Receptor Genes	19	416	4.6%

Other Categories	67	626	10.7%
Total	213	3,005	7.1%

3.8. Mutation Distribution Across Epilepsy-Associated Gene Families

Analysis of mutation distribution across epilepsy-associated gene families revealed distinct patterns (Fig. 1). Ion channel genes, particularly sodium, calcium, and potassium channels, exhibited the highest mutation counts and broadest diversity of variant types.

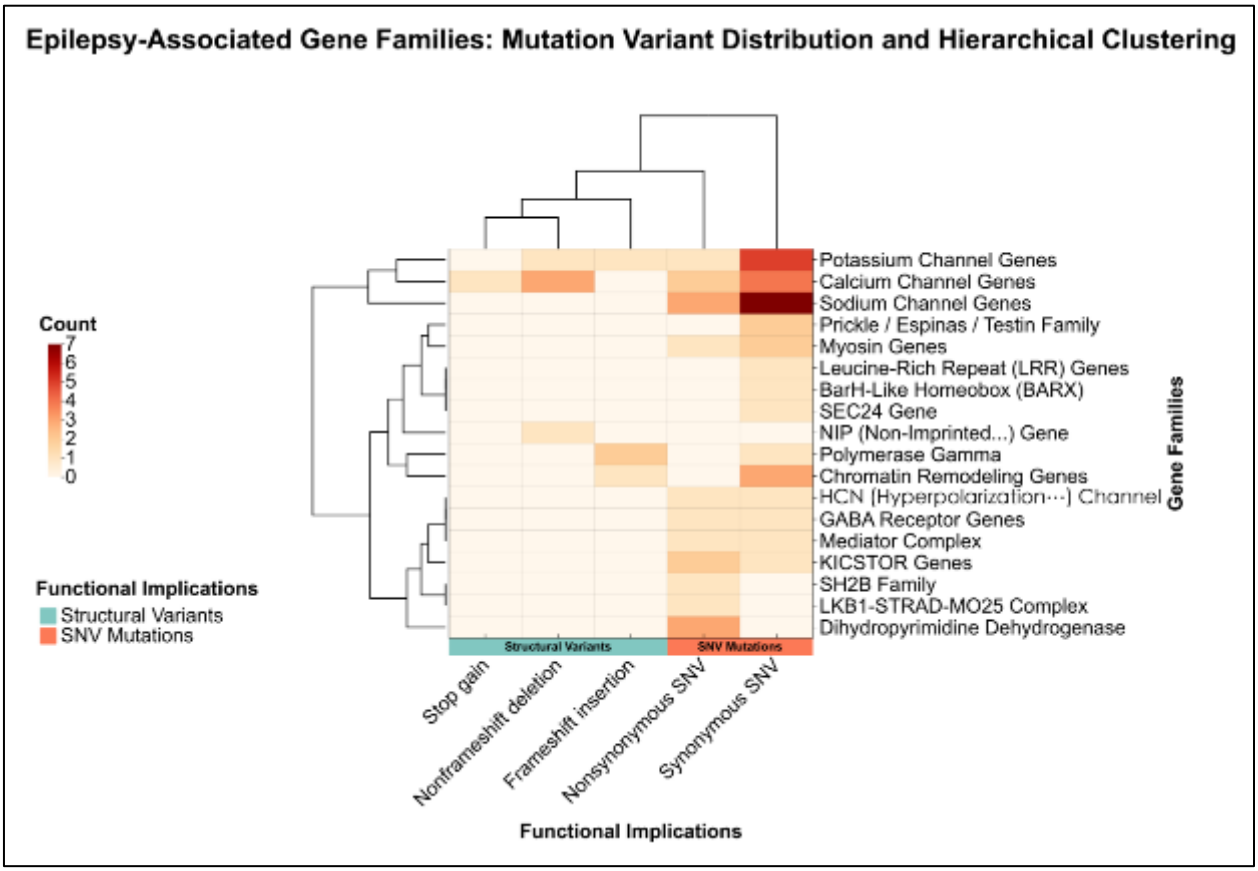

Figure 1 Heat Map Analysis of Epilepsy-Associated Gene Families: Mutation Variant Distribution and Hierarchical Clustering

Hierarchical clustering of variant types and gene families identified patterns driven primarily by variant type rather than functional categories, with single nucleotide variants clustering separately from small insertions and deletions.

4. Discussion

This preliminary analysis provides descriptive insights into the genetic landscape of epilepsy in the Saudi Arabian population, but several limitations must be acknowledged. The small sample size (n=5) significantly restricts statistical power and generalizability of findings. The study should be viewed as hypothesis-generating rather than conclusive, with findings requiring validation in larger cohorts. Our analysis identified 23,361 exonic variants across all genes, with 3,005 variants in established epilepsy-associated genes. The distribution pattern showed ion channel genes as predominant contributors (61% of epilepsy-related variants), consistent with the established role of ion channel dysfunction in epilepsy pathogenesis [7,8].

Variants in sodium, calcium, and potassium channels accounted for the majority of epilepsy-related variants. Notably, *SCN8A* variants (n=87) exceeded *SCN1A* variants (n=12), contrasting with patterns reported in other populations where *SCN1A* predominates [19]. This finding, while preliminary, suggests potential population-specific differences that warrant investigation in larger cohorts. Potassium channel genes, particularly *KCNQ3* (153 variants) and *KCNT1* (146

variants), showed substantial variant burdens, consistent with their established roles in epilepsy syndromes [20]. Calcium channel variants, with *CACNA1A* harboring 296 variants, highlight the importance of calcium signaling dysregulation in epilepsy [21].

GABA receptor variants accounted for 18.38% of epilepsy-related variants, with *GABRG2* showing 27 direct variants. The annotation of 343 variants as "*GABRG2;CCNG1*" reflects genomic co-localization of these genes rather than functional interaction, as previously clarified [22]. This finding emphasizes the importance of careful interpretation of genomic annotations.

NMDA receptor variants represented the largest category (643 variants, 24.47%), with *GRIN2A* showing 167 variants. This substantial burden suggests that excitatory neurotransmission dysregulation contributes significantly to the genetic landscape in this population [8].

The ACMG classification revealed that 61.5% of variants remain of uncertain significance (VUS), highlighting the need for functional validation studies. Only 0.4% of variants were classified as likely pathogenic, emphasizing the challenges in variant interpretation and the importance of population-specific databases for improved classification. Future studies with larger cohorts are essential to establish whether the observed variant distributions represent true population-specific features or sampling artifacts.

The study employed standard bioinformatics workflows and quality control measures, ensuring reliable variant detection. The use of publicly available data and established analysis pipelines facilitates reproducibility. However, the absence of appropriate control groups limits the ability to distinguish population-specific patterns from general genetic variation. These preliminary findings suggest potential directions for future investigation, which includes: larger cohort studies to validate observed patterns, functional validation of variants of uncertain significance, comparative analyses across different populations, and integration with detailed phenotypic data.

While the current findings are preliminary, they contribute to the growing understanding of geographical diversity in epilepsy genetics. The predominance of ion channel variants supports the continued relevance of channel-targeting therapies, though personalized approaches will require much larger datasets and functional validation.

5. Conclusion

This preliminary analysis provides descriptive insights into the genetic landscape of epilepsy in the Saudi Arabian population. The findings highlight the importance of ion channel gene variants and suggest potential population-specific patterns that warrant further investigation. However, the small sample size and exploratory nature of this study necessitate cautious interpretation and emphasize the need for larger-scale investigations to establish definitive population-specific genetic architectures.

Future research should focus on expanding cohort sizes, implementing functional validation studies, and developing population-specific variant databases to improve the clinical utility of genomic findings in epilepsy. These efforts will be essential for advancing precision medicine approaches that account for geographical and population-specific genetic diversity.

5.1. Limitations

This study presents some limitations. With only five patients included, the statistical power is severely limited, and the results cannot be extrapolated to the broader Saudi Arabian population. It however does provide an exploratory perspective to the subject. The research is primarily descriptive in nature and lacks comparative statistical analysis with the ability to distinguish disease-associated variants from population-specific ones affected by the absence of a control group restricts. Furthermore, 61.5% of the identified variants remain of uncertain significance and require functional validation. Finally, the small sample size may not adequately capture the genetic diversity within the Saudi Arabian population.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare that there are no conflicts of interest associated with this work.

Statement of ethical approval

The present research work does not contain any studies performed on animals or human subjects by any of the authors. The study utilized publicly available, de-identified data obtained from the National Center for Biotechnology Information (NCBI) Sequence Read Archive (SRA)

Statement of informed consent

Not applicable. The study utilized publicly available de-identified datasets.

Author's Contribution

Katherine David, Adekunle O. Adeluwoye, and Opeyemi B. Ogunsuyi conceptualized the study. Methodology, software, and formal analysis were conducted by Ayanfeoluwa Alabetutu, Katherine David, and Adekunle O. Adeluwoye. Investigation was performed by Katherine David and Ayanfeoluwa Alabetutu, while data curation, validation, and project administration were handled by Ayanfeoluwa Alabetutu and Adekunle O. Adeluwoye. Katherine David wrote the original draft, which was reviewed and edited by Adekunle O. Adeluwoye and Ayanfeoluwa Alabetutu. Supervision was provided by Ayanfeoluwa Alabetutu and Opeyemi B. Ogunsuyi, with Ayanfeoluwa Alabetutu assuming accountability for data integrity. All authors approved the final manuscript.

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Data Availability Statement

The datasets analyzed during the current study are available in the National Center for Biotechnology Information (NCBI) Sequence Read Archive (SRA) repository under accession numbers ERR10619203–ERR10619207.

References

[1] Bayanova M, Bolatov AK, Bazenova A, Nazarova L, Nauryzbayeva A, Tanko NM, et al. Whole-genome sequencing among Kazakhstani children with early-onset epilepsy revealed new gene variants and phenotypic variability. Mol Neurobiol. 2023;60(8):4324–35. doi:org/10.1007/s12035-023-03346-3

[2] American Association of Neurological Surgeons. Epilepsy [Internet]. Rolling Meadows (IL): American Association of Neurological Surgeons; 2023 [cited 2023 Oct 10]. Available from: https://www.aans.org/Patients/Neurosurgical-Conditions-and-Treatments/Epilepsy

[3] Yin YH, Ahmad N, Makmor-Bakry M. Pathogenesis of epilepsy: Challenges in animal models. Iran J Basic Med Sci. 2013;16(11):1119–32. doi:10.22038/ijbms.2013.1928

[4] Lawal M, Omobayo H, Lawal K. Epilepsy: Pathophysiology, clinical manifestations and treatment options. Br J Neurosci Nurs. 2018;14(2):58–72. doi:10.12968/bjnn.2018.14.2.58

[5] World Health Organization. Epilepsy [Internet]. Geneva: World Health Organization; 2017 [cited 2023 Oct 8]. Available from: https://www.who.int/news-room/fact-sheets/detail/epilepsy

[6] Thakran S, Guin D, Singh P, Kukal S, Rawat C, Yadav S. Genetic landscape of common epilepsies: Advancing towards precision in treatment. Int J Mol Sci. 2020;21(20):7784. doi:10.3390/ijms21207784

[7] Al Anazi AH, Ammar AS, Al-Hajj M, Cyrus C, Aljaafari D, Khoda I, et al. Whole-exome sequencing of a Saudi epilepsy cohort reveals association signals in known and potentially novel loci. Hum Genomics. 2022;16(1):71. doi:10.1186/s40246-022-00444-6

[8] CD Genomics. Bioinformatics analysis [Internet]. Shirley (NY): CD Genomics; 2023 [cited 2023 Nov 3]. Available from: https://bioinfo.cd-genomics.com/comparative-genomics-analysis.html

[9] Ma R, Duan Y, Zhang L, Qi X, Zhang L, et al. SCN1A-related epilepsy: Novel mutations and rare phenotypes. Front Mol Neurosci. 2022;15:826183. doi:10.3389/fnmol.2022.826183

[10] Veltra D, Theodorou V, Katsalouli M, Vorgia P, Niotakis G, et al. SCN1A channels a wide range of epileptic phenotypes: Report of novel and known variants with variable presentations. Int J Mol Sci. 2024;25(11):5644. doi:10.3390/ijms25115644

[11] Metzker ML. Sequencing technologies: The next generation. Nat Rev Genet. 2010;11(1):31–46. doi:10.1038/nrg2626

[12] Ewels P, Magnusson M, Lundin S, Käller M. MultiQC: Summarize analysis results for multiple tools and samples in a single report. Bioinformatics. 2016;32(19):3047–48. doi:10.1093/bioinformatics/btw354

[13] Bolger AM, Lohse M, Usadel B. Trimmomatic: A flexible trimmer for Illumina sequence data. Bioinformatics. 2014;30(15):2114–20. doi:10.1093/bioinformatics/btu170

[14] Langmead B, Salzberg SL. Fast gapped-read alignment with Bowtie 2. Nat Methods. 2012;9(4):357–59. doi:10.1038/nmeth.1923

[15] Li H, Durbin R. Fast and accurate short read alignment with Burrows–Wheeler transform. Bioinformatics. 2009;25(14):1754–60. doi:10.1093/bioinformatics/btp324

[16] McKenna A, Hanna M, Banks E, Sivachenko A, Cibulskis K, Kernytsky A, et al. The Genome Analysis Toolkit: A MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res. 2010;20(9):1297–1303. doi:10.1101/gr.107524.110

[17] Benjamin D, Sato T, Cibulskis K, Getz G, Stewart C, Lichtenstein L. Calling somatic SNVs and indels with Mutect2. bioRxiv. 2019. doi:10.1101/861054

[18] Wang K, Li M, Hakonarson H. ANNOVAR: Functional annotation of genetic variants from high-throughput sequencing data. Nucleic Acids Res. 2010;38(16):e164. doi:10.1093/nar/gkq603

[19] Wickham H, Averick M, Bryan J, Chang W, McGowan LD, François R, et al. Welcome to the tidyverse. J Open Source Softw. 2019;4(43):1686. doi:10.21105/joss.01686

[20] Brunklaus A, Dorris L, Ellis R, Reavey E, Lee E, Forbes G, et al. The clinical utility of an SCN1A genetic diagnosis in infantile-onset epilepsy. Dev Med Child Neurol. 2013;55(2):154–61. doi:10.1111/dmcn.12030

[21] Veeramah KR, O'Brien JE, Meisler MH, Cheng X, Dib-Hajj SD, Waxman SG, et al. De novo pathogenic SCN8A mutations causing early infantile epileptic encephalopathy and SUDEP. Am J Hum Genet. 2012;90(3):502–510. doi:10.1016/j.ajhg.2012.01.006

[22] Pietrobon D. Calcium channels and channelopathies of the central nervous system. Mol Neurobiol. 2002;25(1):31–50. doi:10.1385/mn:25:1:031

Authors short biography

Katherine David is a First-Class graduate of Biomedical Technology from the Federal University of Technology, Akure (FUTA), with training in bioinformatics and data analytics. She is a Technical Research Officer at UREKA Biotec (OmicsLogic Africa/OmicsLogic, USA). Her work centers on human genomics and precision medicine, focusing on the identification and interpretation of disease-associated genetic variants. She is driven by the goal of translating genomic discoveries into clinically actionable insights, particularly in precision oncology and targeted therapeutic development. Her approach integrates molecular biology with bioinformatics to advance personalized healthcare.

Ayanfeoluwa Alabetutu is a molecular scientist, computational biology professional, and social entrepreneur specializing in Next-Generation Sequencing (NGS) and data-driven molecular insights. He is proficient in diverse molecular assays and bioinformatics applications, including genome assembly, microbiome analysis, and machine learning in oncology and virology. Currently, he serves as the Co-Founder and COO of the health-tech startup SKRIND, and as a Bioinformatics Scientist and Community Engagement Manager with UREKA Biotec (OmicsLogic Africa/OmicsLogic, USA). Based in Novosibirsk, Russia, he is a Bioinformatics

	Scientist at the Federal Research Centre of Translational Medicine and a Senior Research Fellow at the University of Ilorin, Nigeria. An active mentor and trainer, Ayanfeoluwa is dedicated to advancing health informatics, biomedical data science, and interdisciplinary collaboration to address global social and research challenges.]
	**Opeyemi B. Ogunsuyi** (PhD) is a Lecturer in Medical Biochemistry at the Federal University of Technology, Akure, and a Research Fellow at the University of Birmingham, UK. His research specializes in neurochemistry and neurophytotherapy, focusing on functional foods and gender-based natural therapies for neurodegenerative diseases using Drosophila melanogaster models. He is a member of the International Society for Neurochemistry (ISN), the African Society of Drosophilists, and the Neuroscience Society of Nigeria (NSN).
	**Adeluwoye Adekunle Oluwatosin** is a medical laboratory scientist, certified cytotechnologist, and entrepreneur specializing in molecular diagnostics, cancer genomics, and AI-powered healthcare solutions. He is proficient in advanced histopathology, cytopathology, next-generation sequencing (NGS), and bioinformatics applications, including genomic data analysis, machine learning in diagnostic pathology, and SARS-CoV-2 genomic surveillance. Currently, he serves as Program Director and Faculty with UREKA Biotec (OmicsLogic Africa/OmicsLogic, USA). Based in Ibadan, Nigeria, he is a PhD candidate in Molecular Biology and Genomics at Lead City University and holds international certification as a Cytotechnologist (CT(IAC)) from the International Academy of Cytology. An accomplished author of two academic textbooks and multiple peer-reviewed publications, Adeluwoye combines technical expertise with entrepreneurial vision, leading innovation in digital diagnostics, bioinformatics education, and strategic public-private partnerships, he is dedicated to advancing laboratory medicine, precision diagnostics, and healthcare infrastructure development across Sub-Saharan Africa.