BMC Medical Genomics

CASE REPORT

Open Access

Long-read sequencing identifies a novel de novo inversion in *SMARCC2* in a pediatric patient with Coffin-siris syndrome 8: a case report

Amal Abdulsalam Ibrahim[1,2], Waleed Aamer[3], Elbay Aliyev[3], Aljazi Al-Maraghi[3], Saba F. Elhag[3], Mona Abdi[3], Najeeb Syed[3], Sasirekha Palaniswamy[3], Ammira S. A. Akil[4], Madeeha Kamal[5] and Khalid A. Fakhro[1,3,6*]

Abstract

Background Coffin-Siris Syndrome 8 (CSS8; MIM# 618362) is a rare neurodevelopmental disorder caused by heterozygous variants in the *SMARCC2* gene. Patients with CSS8 present with variable phenotypic presentations, with speech abnormalities, behavioral issues, hypotonia, and dysmorphic features being the most common. Here, we report a novel de novo inversion involving *SMARCC2,* identified through long-read whole-genome sequencing, which highlights its added diagnostic value in uncovering complex structural variants.

Case presentation The patient is a 12-year-old male of Arab descent, born to healthy, non-consanguineous parents. He presented with speech difficulties and behavioral issues, including autism spectrum disorder (ASD), obsessive–compulsive disorder (OCD), and attention-deficit/hyperactivity disorder (ADHD). Long-read sequencing identified a 233 kb inversion on chromosome 12, with breakpoint 1 disrupting *SMARCC2* between exons 16 and 17. This inversion was confirmed by Sanger sequencing. Disruption of *SMARCC2* is predicted to cause loss of function, resulting in haploinsufficiency.

Conclusions This is the first report of a structural variant in a patient with CSS8. This finding expands our genetic understanding of CSS8 and demonstrates the diagnostic utility of long-read sequencing in uncovering clinically relevant structural variants that may be missed by conventional methods.

Keywords Long-read sequencing, Coffin-Siris syndrome 8, Autism spectrum disorder, Structural variant, Oxford nanopore technologies, Rare genetic disorders

*Correspondence:
Khalid A. Fakhro
kfakhro@sidra.org
[1]College of Health & Life Sciences, Hamad Bin Khalifa University, Qatar Foundation, Doha 5825, Qatar
[2]Department of Biomedical Sciences, College of Health Sciences, QU Health, Qatar University, Doha 2713, Qatar
[3]Human Genetics Section, Sidra Medicine, Doha, Qatar
[4]Precision Genomics and Translational Omics Lab, Metabolic and Mendelian Disorders Clinical Genomic Research Program, Sidra Medicine, Doha 26999, Qatar
[5]Department of Pediatrics, Sidra Medicine, Doha 26999, Qatar
[6]Department of Genetic Medicine, Weill Cornell Medicine, Doha 24144, Qatar

Background

Coffin-Siris syndrome (CSS; OMIM #135900) is a rare congenital anomaly characterized by intellectual disability, developmental delays, coarse facial features, hypertrichosis, absence/hypoplasia of the distal phalanx, and multiple organ anomalies [1]. Fewer than 200 cases have been molecularly confirmed with CSS, however, this number may be an underestimate due to variable disease presentation and genetic heterogeneity [2]. Multiple subtypes of CSS have been identified, each associated with distinct variants in the BAF complex genes that are involved in SWI/SNF chromatin remodeling. These genes include *ARID1A*, *ARID1B*, *ARID2*, *DPF2*, *SMARCA4*, *SMARCB1*, *SMARCC2*, *SMARCE1*, *SOX11*, and *SOX4* [1].

Among the known CSS subtypes, Coffin-Siris syndrome-8 (CSS8; OMIM# 618362) presents with a broad spectrum of neurodevelopmental manifestations, including varying degrees of intellectual disability (HP:0001249), speech impairment (HP:0000750), hypotonia (HP:0001252), feeding difficulties (HP:0011968), and behavioral abnormalities such as hyperactivity (HP:0000752), self-injurious behavior (HP:0100716) and aggressive behavior (HP:0000718), and sleep abnormality (HP:0002360). Dysmorphic features can be variable, with some individuals exhibiting hypertrichosis (HP:0000998) or sparse scalp hair (HP:0002209), prominent eyebrows (HP:0000574), a thin upper vermilion (HP:0000219), and an upturned nose (HP:0000463), while others may not display noticeable craniofacial abnormalities [3]. Coffin-Siris Syndrome 8 is a phenotypically heterogeneous disorder inherited in an autosomal dominant mode, mainly through variants in *SMARCC2*, a gene located on chromosome 12q13.

In a study investigating *SMARCC2*-related neurodevelopmental disorders, 15 unrelated CSS8 patients were reported with a broad phenotypic spectrum, including speech abnormalities (13/15), behavioral issues (10/15), hypotonia (13/15), and dysmorphic features (11/15) [3]. More recently, a study with a large cohort of 65 individuals with *SMARCC2* variants revealed that, in addition to a wide range of neurodevelopmental features, 35% of cases exhibited autistic behaviors, suggesting a strong link between *SMARCC2* variants and Autism Spectrum Disorder (ASD) [4]. Notably, all previously reported variants in the literature were point mutations. Here, we present a pediatric case of CSS8, presenting with neurodevelopmental manifestations including autistic behaviors and speech delay, negative for routine microarray and short-read sequencing of the SMARCC2 gene. Using long-read sequencing, we identified and validated a novel de novo 233-kb inversion impacting exons 16 and 17 of the *SMARCC2* gene, among others. Our findings highlight the utility of long-read sequencing in diagnosing rare Mendelian diseases.

Case presentation

The patient is a 12-year-old male of Arab descent. He is the first child of non-consanguineous, healthy parents and has two younger unaffected sisters. He was born full-term via normal vaginal delivery, with a birth weight of 3.2 kg. His developmental milestones included sitting without support at the age of eight months, crawling at ten months, and walking at fourteen months.

The patient's developmental concerns were first noted at the age of two when his mother observed atypical behaviors, including not responding to his name, absence of eye contact, delayed speech development, social isolation, and difficulties in expressing his needs. He also had a limited two-word vocabulary at the age of two. Despite these challenges, the patient did not exhibit signs of intellectual disability, as assessed using the Wechsler Intelligence Scale for Children, Fifth Edition (WISC-V), resulting in a total IQ score of 88, which suggests low-average cognitive ability [5]. However, the initial physical assessment had described the intellectual level as average based on clinical judgment. The patient also had no history of significant head injuries, seizures, or surgical interventions. Also, dysmorphology evaluation was performed by a senior pediatric physician during the patient's initial clinical visit in May 2021, who did not note any apparent dysmorphic features at the time of assessment. However, the evaluation was not conducted by a clinical geneticist, and clinical photographs were not obtained due to a lack of parental consent. At three years old, the patient was diagnosed with Autism Spectrum Disorder (ASD) (HP:0000729) following the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-V). The patient also has comorbidities, including Obsessive–Compulsive Disorder (OCD). (diagnosed based on requesting things to be done in a specific way and performing repetitive checking). Also, according to his presentation of hyperactivity (HP:0000752), inattention, and being talkative, he was diagnosed with Attention-Deficit/Hyperactivity Disorder (ADHD). Other behavioral history included echolalia (HP:0010529), tiptoeing, self-hitting when frustrated (HP:0100716), and rigidity in routines (HP:0002063).

Between the ages of three and six, the patient received interventional therapy at the child development center, which included speech therapy, Applied Behavior Analysis (ABA), and occupational therapy. Through these therapeutic interventions, he has made significant improvements in reading, writing, and mathematical skills. However, he continues to struggle with social and communication skills, exhibiting traits such as excessive talkativeness, inattentiveness, poor eye contact, and an

intense fixation on gaming. He also faces difficulties in forming friendships and often forgets daily essentials. His social anxiety has gradually improved, yet he still experiences episodes of agitation and occasional anger, which have been managed with treatment using risperidone at a dose of 0.7 mg/day. Currently, he is also taking fluoxetine 20 mg daily. Due to considerable hyperactivity observed both at school and at home, he was recently prescribed methylphenidate at a dose of 5 mg twice daily. Previous medications included atomoxetine 18 mg per day and clonidine 0.1 mg daily in oral liquid form, both of which were discontinued after one year due to ineffectiveness.

The patient underwent clinical microarray and exome testing, but no diagnostic findings were reported. As such, he was referred to the research department and enrolled in the BARAKA study (Building a Resource for the Advancement of Knowledge of Autism in Qatar). Whole genome sequencing was performed as previously described [6, 7]. Initial analysis identified a homozygous missense variant in the *NBN* gene (NM_002485.5; c.671G > A; p.Gly224Glu), associated with Nijmegen breakage syndrome. The variant was classified as a Variant of Uncertain Significance (VUS) based on ACMG guidelines (2015) [8], and the in silico pathogenicity scores of the variant were as follows: CADD = 25.2, PolyPhen2 = 1, and M-CAP = 0.138698. Given that the clinical phenotype did not fully align with the characteristic features of this syndrome, it was not considered clinically relevant and was not reported to the patient's family. In contrast, structural variant analysis identified a potentially complex rearrangement involving the *SMARCC2* gene, which has a Simons Foundation Autism Research Initiative (SFARI) score of 1S (High Confidence, Syndromic), indicating a stronger correlation with the patient's phenotype. Given the complexity and relevance of this finding, further investigation was necessary to confirm it.

We proceeded with whole-genome long-read sequencing on DNA extracted from whole blood as described previously [6, 7]. High-molecular-weight DNA was sheared using the Megaruptor 3 (Diagenode, USA) to generate fragments with an average length of 15 kb. Libraries were then prepared with the Ligation Sequencing Kit (SQK-LSK114-XL; Oxford Nanopore Technologies, UK) according to the manufacturer's instructions. Sequencing was performed on the PromethION P48 platform (Oxford Nanopore Technologies, UK) using R10.4.1 flow cells, with a minimum target of 20 × coverage per sample. Raw data were aligned to the GRCh38 human reference genome using Minimap2, and resulting Binary Alignment Map (BAM) files were processed using the wf-human-variation workflow (v2.2.5) from Epi2me Labs (https://github.com/epi2me-labs/wf-human-variation), which includes Sniffles2 for structural variant calling (version 2.0.7) [9] with standard key parameters recommended in the workflow. Sequencing achieved an average read coverage of 23.07 × and an N50 of 17.1 kb.

Analysis revealed a 233-kb inversion supported by 22 reads and a local read depth profile of (upstream: 13; start: 9; center: 22: end: 7; downstream: 13). The first breakpoint (chr12:56173943) occurs within the middle of the *SMARCC2* gene, between exons 16 and 17, while the second breakpoint (chr12:56407078) is located within the intergenic region between the *TIMELESS* and *APOF* genes (Fig. 1B). The variant description according to Human Genome Variation Society (HGVS) nomenclature is at the genomic level as NC_000012.12:g.56173943_56407078inv and at the transcript level as NM_001330288.2:c.–217617_1497-94inv. To confirm the inversion, we performed Sanger sequencing using specific primers designed to flank the breakpoints, which revealed the exact breakpoint sequence and confirmed the de novo origin (Fig. 1C and D, and Table 1). Long-read breakpoint visualizations are provided in Fig. 2.

Discussion and conclusion

In this report, we describe a pediatric case of CSS8 with a novel de novo inversion involving the *SMARCC2* gene, further expanding our understanding of the genetic landscape of CSS8 syndrome. The variant was identified through long-read sequencing in a patient who had previously undergone chromosomal microarray and short-read sequencing, both of which were inconclusive. In resolving the diagnostic odyssey of this patient, our findings support the growing diagnostic utility of long-read sequencing in resolving complex structural variants that are typically missed by standard technologies [10–13]. For example, long-read sequencing enabled the detection of different type of SVs, such as large deletions in *PRKAR1A* in Carney complex and *G6PC* in Glycogen Storage Disease type IA, as well as insertion within *TAF1*, a gene associated with X-linked Parkinsonism, and a chromosomal translocation linked to developmental disorders [14–17].

The main clinical features of CSS8 are neurodevelopmental and craniofacial abnormalities. These characteristics were established in a cohort of 15 unrelated individuals with CSS8, where 13/15 had speech abnormalities and 10/15 showed behavioral difficulties, including aggression, hyperactivity, self-injury, and obsessive or rigid behavior [3]. Additionally, the study also reported hypotonia (13/15) and dysmorphic features (11/15). Although the patient presented with speech delay and behavioral issues, subtle facial or syndrome-specific features could not be fully evaluated, as no facial photographs were obtained during clinical assessment. Further, the patient had severely limited social skills, leading to the diagnosis of ASD. This is supported by a

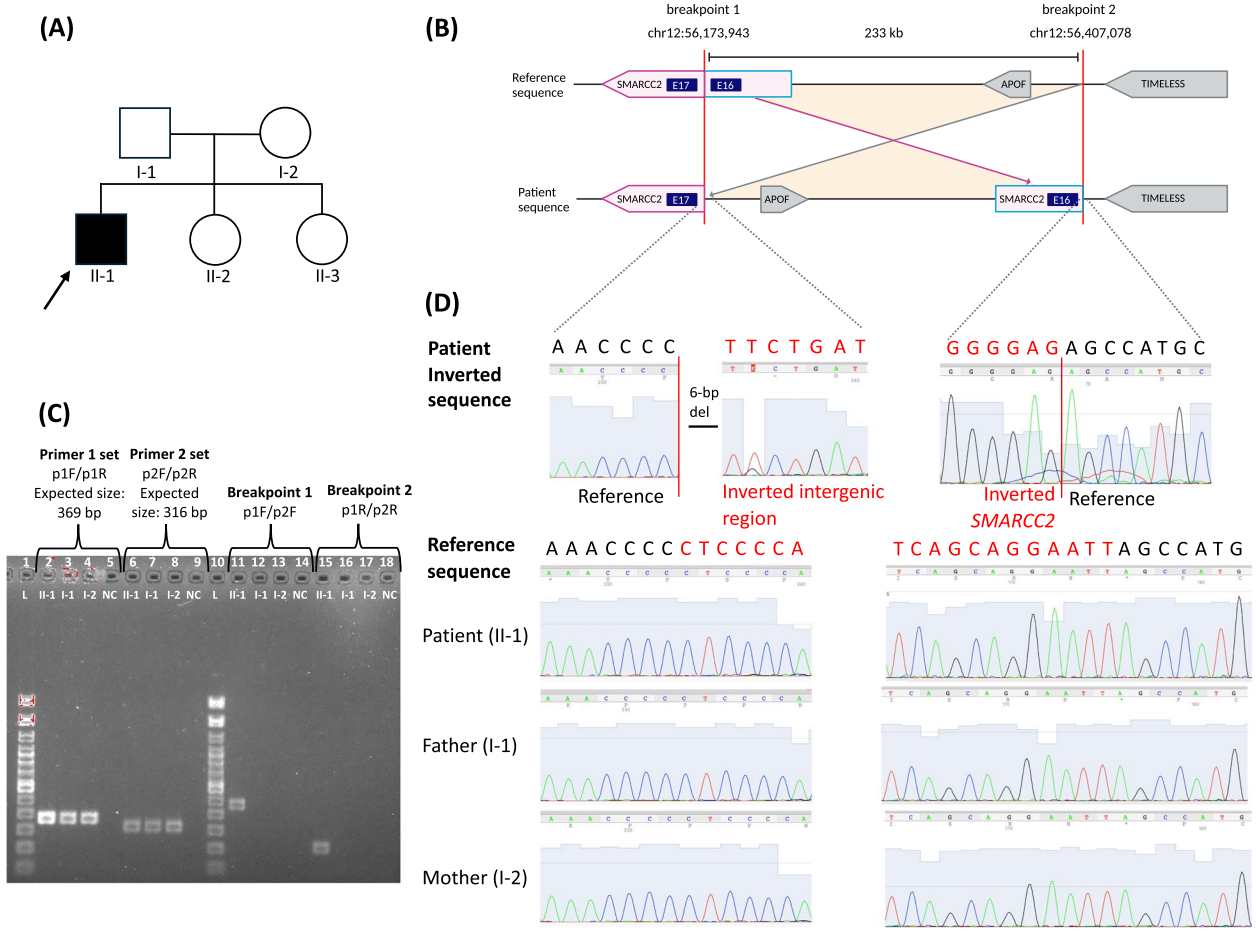

Fig. 1 Clinical data and the results of genetic analysis of the patient. **A** Pedigree of the family members, with the arrow indicating the proband (II-1). **B** Diagram of patient's DNA, highlighting inverted segments and their impact on *SMARCC2* and surrounding regions. **C** Gel electrophoresis results using two primer sets confirming the inversion. **D** Sanger sequencing chromatograms showing the precise sequences at both breakpoints in the patient compared to the reference sequence. Facial images were not obtained due to a lack of parental consent

Table 1 Sequence of forward and reverse primers used around the breakpoints

Primer set	Name	Sequence	Product size
P1	Forward	CCATAGCACCTCACCCTACC	369
	Reverse	CTTGGGTGTCACTGGCATTT	
P2	Forward	AGAGAAGCCAGAAGTCCCAC	316
	Reverse	TGAGGCATCTTCTTGTCCCA	

recent cohort study that reported that 35% of *SMARCC2* patients exhibited autistic traits, highlighting the phenotypic variability in neurodevelopmental and behavioral features [4]. Supporting this, research on the role of *SMARCC2* in autism using an in-silico protein analysis (DAPPLE) has revealed that *SMARCC2* is directly associated with about 11 genes that harbor de novo mutations linked to autism. [18].

Genetic variation in *SMARCC2* has been associated with CSS8. Here, we identified a 233 kb inversion on chromosome 12, with breakpoint 1 located in intron 16 of SMARCC2, leading to the inversion of exons 1–16 of *SMARCC2*, a dosage-sensitive gene with pLI = 1.0 and LOEUF = 0.17 (ACMG criteria 2C-1; score 0.90) [19] (Fig. 1B). The structural variant has been confirmed as de novo, and the reported phenotype is consistent with *SMARCC2*-related phenotypes (Coffin-Siris syndrome-8 OMIM #618,362), albeit with some genetic heterogeneity (criteria 5 A; score 0.15) [19]. Based on these criteria, the variant was classified as pathogenic (total score 1.05) [19]. Thus, inversion of the *SMARCC2* sequence is predicted to cause loss of function (LoF), resulting in haploinsufficiency [20]. Currently, there are no reports of structural variants (variants > 50 bp) affecting only the *SMARCC2 gene* in public databases as ClinVar, DECIPHER, and Human Gene Mutation Database (HGMD). However, several pathogenic LoF variants have been reported in CSS8 cases across exons 1–16 of *SMARCC2*. These variants include splice-site, nonsense, and frameshift mutations (examples listed in Table 2). Among these, a de novo splice-site variant in intron 16 of *SMARCC2* (NM_001330288.2:c.1496 + 1G > T) was reported in monozygotic

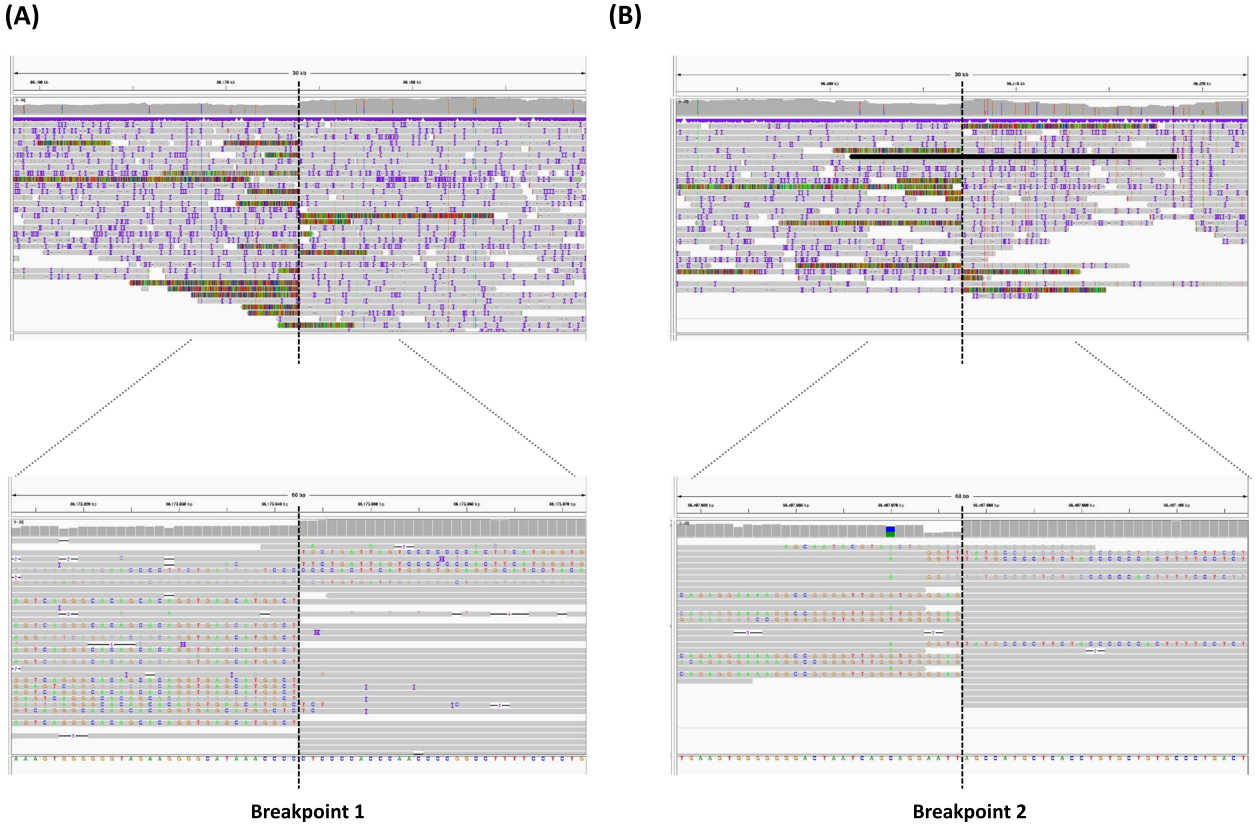

Fig. 2 IGV visualization of inversion breakpoints. **A** IGV visualization at breakpoint 1 (chr12:56,173,943). **B** IGV visualization at breakpoint 2 (chr12:56,407,078). In both panels, soft-clipped reads (colored segments) indicate sequences that do not align with the reference genome, pointing to the exact locations of the inversion

Table 2 ClinVar-reported pathogenic/likely pathogenic *SMARCC2* variants associated with CSS8

Nucleotide	Variant type	Genomic location (GRCh38)	Exon/ Intron Affected	Origin	Molecular consequence	Classification	Submission Accession	Literature Reference
NM_001330288.2: c.1496 + 1G > T	SNV	12:56174650	Intron 16	De novo	Splice donor	Pathogenic	SCV005414383	ClinVar submission; [21]
NM_001330288.2: c.574C > T (p.Arg192Ter)	SNV	12:56183919	Exon 7	Maternal	Nonsense	Pathogenic/Likely pathogenic	SCV001985052	[4]
NM_001330288.2: c.805C > T (p.Arg269Ter)	SNV	12:56181739	Exon 9	Germline	Nonsense	Pathogenic/Likely pathogenic	SCV002061496	[4]
NM_001330288.2: c.723G > A (p.Trp241Ter)	SNV	12:56181821	Exon 9	Germline	Nonsense	Pathogenic	SCV000891367	[3]
NM_001330288.2: c.1311-3C > G	SNV	12:56178096	Intron 14	Germline	Intron variant	Pathogenic	SCV000891365	[3]
NM_001330288.2: c.1091del (p.Lys364fs)	Indel (deletion 1 bp)	12:56179047	Exon 12	De novo	Frameshift	Pathogenic	SCV003935120	ClinVar submission; [22]
NM_001330288.2: c.1094_1097del (p.Lys365fs)	Microsatellite, (4 bp)	12: 56179041–56179044	Exon 12	De novo	Frameshift	Pathogenic	SCV003035419	[4]

SNV Single Nucleotide Variant, *Indel* Insetion-deletion

twins with CSS8. Functional mRNA studies of a clone derived from the patient sample revealed aberrant splicing that disrupts the SWIRM domain of *SMARCC2,* supporting a loss-of-function mechanism [21].

This is the first report of a structural variant in a patient with CSS8. This novelty highlights the added diagnostic value of long-read sequencing in resolving complex structural variants. Diagnosis of rare genetic disorders with inconclusive findings could be improved by integrating long-read sequencing into diagnostic pipelines. Future studies should aim to uncover the mechanisms by which this inversion impacts *SMARCC2* function and contributes to the patient's phenotype.

Abbreviations

CSS	Coffin-Siris syndrome
CSS8	Coffin-Siris syndrome-8
ASD	Autism Spectrum Disorder
OCD	Obsessive–Compulsive Disorder
ADHD	Attention-Deficit/Hyperactivity Disorder
DSM-V	Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition
WISC-V	Wechsler Intelligence Scale for Children, Fifth Edition
BARAKA	Building a Resource for the Advancement of Knowledge of Autism
IRB	Institutional Review Board
VUS	Variant of Uncertain Significance
OMIM	Online Mendelian Inheritance in Man
SFARI	Simons Foundation Autism Research Initiative
GRCh38	Genome Reference Consortium Human Build 38
HGVS	Human Genome Variation Society
BAM	Binary Alignment Map
SNV	Single-Nucleotide Variant
HGMD	Human Gene Mutation Database
LoF	Loss of function

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12920-025-02269-3.

Supplementary Material 1

Acknowledgements

We are grateful to Qatar National Research Fund (QNRF) for funding this research (BARAKA-Qatar project: NPRP10-0202-170320). We express our gratitude to all individuals with Autism Spectrum Disorder (ASD) and their families who took part in this cohort. We would also like to acknowledge Aamira Saed for being involved in patient recruitment.

Authors' contributions

Conceptualization: A.A.I, W.A, E.A, K.A.F. Methodology: A.A.I, E.A, W.A, N.S. Software: A.A.I, E.A, W.A, N.S. Formal analysis: A.A.I, E.A, M.A. Data curation: A.A.I, E.A, M.A. Investigation: A.A.I, E.A, W.A, S.F.E, M.A, N.S. Project administration: S.P, A.S.A.A. Resources: A.A, S.F.E, K.A.F. Validation: M.K. Visualization: A.A.I, E.A. Writing—original draft: A.A.I, W.A. Writing—review and editing: A.A.I, W.A, E.A, A.A, S.F.E, M.A, N.S, S.P, A.S.A.A, M.K, K.A.F. Funding acquisition: K.A.F. Supervision: A.A, M.K, S.P, A.S.A.A, K.A.F. All authors read and approved the final manuscript.

Funding

This work was supported by Qatar Foundation, Qatar National Research Fund (QNRF) (BARAKA-Qatar project: NPRP10-0202–170320).

Data availability

The datasets analyzed during the current study are available in the Genome Sequence Archive at Sidra Medicine, Qatar. WGS data of the family in this manuscript in the BARAKA study has been submitted to the MSSNG project. Access to the data can be requested by completing the data access agreement at https://research.mss.ng. The identified variant has been submitted to ClinVar (Accession: VCV004280325.1). A VCF file with the detected structural variant is provided in the supplementary materials.

Declarations

Ethics approval and consent to participate

This study adheres to the principles of the Declaration of Helsinki and was approved by the Institutional Review Board (IRB) of Sidra Medicine (IRB No. 1500767). Written informed consent was obtained from all participants, including the patient's parents for themselves and as legal guardians on behalf of the patient, who is under the age of 16.

Consent for publication

Written consent for publication was obtained from parents to publish medical and genomic data related to the patient and family members.

Competing interests

The authors declare no competing interests.

Received: 8 July 2025 / Accepted: 3 November 2025
Published online: 25 November 2025

References

1. Vasko A, Drivas TG, Schrier Vergano SA. Genotype-phenotype correlations in 208 individuals with Coffin-Siris syndrome. Genes (Basel). 2021;12(6):937.
2. Vergano, S.S., et al., Coffin-siris syndrome. GeneReviews®, 2021.
3. Machol K, et al. Expanding the spectrum of BAF-related disorders: de novo variants in SMARCC2 cause a syndrome with intellectual disability and developmental delay. Am J Hum Genet. 2019;104(1):164–78.
4. Bosch E, et al. Elucidating the clinical and molecular spectrum of SMARCC2-associated NDD in a cohort of 65 affected individuals. Genet Med. 2023;25(11):100950.
5. Flanagan, D.P. and V.C. Alfonso, Essentials of WISC-V assessment. First edition. ed. Essentials of Psychological Assessment. 2017, Hoboken: Wiley.
6. Abdi M, et al. Genomic architecture of autism spectrum disorder in Qatar: the BARAKA-Qatar study. Genome Med. 2023;15(1):81.
7. Al-Maraghi A, et al. A loss-of-function AGTR1 variant in a critically-ill infant with renal tubular dysgenesis: case presentation and literature review. BMC Nephrol. 2024;25(1):139.
8. Richards S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17(5):405–24.
9. Smolka M, et al. Publisher correction: detection of mosaic and population-level structural variants with Sniffles2. Nat Biotechnol. 2024;42(10):1616.
10. AlAbdi L, et al. Beyond the exome: utility of long-read whole genome sequencing in exome-negative autosomal recessive diseases. Genome Med. 2023;15(1):114.
11. De Clercq G, et al. Full characterization of unresolved structural variation through long-read sequencing and optical genome mapping. Sci Rep. 2024;14(1):29142.
12. Hiatt SM, et al. Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. HGG Adv. 2021;2:2.
13. Negi S, et al. Advancing long-read nanopore genome assembly and accurate variant calling for rare disease detection. Am J Hum Genet. 2025;112(2):428–49.
14. Dutta UR, et al. Breakpoint mapping of a novel de novo translocation t(X;20)(q11.1;p13) by positional cloning and long read sequencing. Genomics. 2019;111(5):1108–14.
15. Merker JD, et al. Long-read genome sequencing identifies causal structural variation in a Mendelian disease. Genet Med. 2018;20(1):159–63.
16. Miao H, et al. Long-read sequencing identified a causal structural variant in an exome-negative case and enabled preimplantation genetic diagnosis. Hereditas. 2018;155:32.

17. Aneichyk T, et al. Dissecting the causal mechanism of X-linked dystonia-parkinsonism by integrating genome and transcriptome assembly. Cell. 2018;172(5):897-909 e21.
18. Neale BM, et al. Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature. 2012;485(7397):242–5.
19. Riggs ER, et al. Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen). Genet Med. 2020;22(2):245–57.
20. Gerasimavicius L, Livesey BJ, Marsh JA. Loss-of-function, gain-of-function and dominant-negative mutations have profoundly different effects on protein structure. Nat Commun. 2022;13(1):3895.
21. ClinVar. NM_001330288.2(SMARCC2):c.1496+1G>T. ClinVar; [VCV0003381186.2]. https://www.ncbi.nlm.nih.gov/clinvar/variation/3381186/. Accessed 17 May 2025.
22. ClinVar. NM_001330288.2:c.1091del (p.Lys364fs). ClinVar; [VCV0002506488.2]. https://www.ncbi.nlm.nih.gov/clinvar/variation/2506488/. Accessed 17 May 2025.

Publisher's Note