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Phenotypes of autism spectrum disorder and schizoaffective disorder associated with *SETD1B* gene but without intellectual disability and seizures

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Abstract

The *SETD1B* gene, located on chromosome 12q24, is one of the chromatin-modifying genes involved in epigenetic regulation of gene transcription. The phenotype of pathogenic variants in the *SETD1B* gene includes intellectual disability, seizures, and language delay (IDDSELD, OMIM 619000). In this study, we present a family consisting of consanguineous parents who died of cancer and their offspring. This family includes two cases diagnosed with autism spectrum disorder (ASD); six cases diagnosed with schizophrenia, bipolar disorder, or schizoaffective disorder; there cases diagnosed with cancer; and five cases who died of unknown causes in early childhood. Three affected members of this family agreed to genetic testing. We used whole exome sequencing. We report a novel in-frame deletion variant of the *SETD1B* gene in a family with cases diagnosed with schizoaffective disorder and ASD without seizures and intellectual disability. It was found that the phenotypic features were inherited for at least three generations in the family we presented, and it was shown that the pathogenic variant of the *SETD1B* gene was transmitted from the affected parent to his affected children. In addition, the father was diagnosed with both schizoaffective disorder and leukemia. We proposed an association between rare variants of *SETD1B* and phenotypes of ASD and schizoaffective disorder without seizures and intellectual disability. The *SETD1B* gene is included in both the ASD genetic database of SFARI (https://gene.sfari.org/) and the cancer database of COSMIC (https://cancer.sanger.ac.uk/cosmic). However, there are very few reports of *SETD1B* gene variants as clinical entities. To our knowledge, the *SETD1B* gene variant has not been previously reported in an individual diagnosed with both a neuropsychiatric disorder and cancer.

KEYWORDS
autism, cancer, schizoaffective disorder, SETD1B, WES

Abbreviations: ACMG, American College of Medical Genetics and Genomics; ASD, autism spectrum disorder; CADD, combined annotation dependent depletion; COSMIC, Catalogue of Somatic Mutations in Cancer; DNA, deoxyribonucleic acid; EEG, electroencephalography; ExAC, The Exome Aggregation Consortium; gnomAD, Genome Aggregation Database; HGMD, The Human Gene Mutation Database; IDDSELD, Intellectual Developmental Disorder, Seizures and Language Delay; MRI, magnetic resonance imaging; SD, standard deviation; SETD1A, SET Domain Containing 1A; SETD1B, SET Domain Containing 1B; SFARI, Simons Foundation Autism Research Initiative; WES, whole exome sequencing; WISC-R, Wechsler Intelligence Scale for Children– Revised.

1 | INTRODUCTION

SET domain-containing protein 1B (*SETD1B*) gene, located on chromosome 12q24, is one of the chromatin-modifying genes involved in epigenetic regulation of gene transcription (Hiraide et al., 2018; Lee et al., 2007). The *SETD1B* gene encodes a histone methyltransferase that plays a role in the production of trimethylated histone H3 at Lys4 (H3K4) (Lee et al., 2007). Loss of function of individual histone methyltransferases has been associated with human diseases such as neurodevelopmental disorders and cancer (Roston et al., 2021). Diseases associated with variants in the *SETD1B* gene have been identified particularly in recent years.

The phenotype of pathogenic variants in the *SETD1B* gene includes intellectual developmental disorder, seizures, and language delay (IDDSELD, OMIM 619000). Additional behavioral problems included attention-deficit/hyperactivity disorder, autism spectrum disorder (ASD), anxiety, self-mutilation, irritability, and sleep disturbance (Roston et al., 2021). IDDSELD syndrome has been described in eight cases carrying pathogenic variants of the *SETD1B* gene and four cases carrying microdeletions including the *SETD1B* gene (Den et al., 2019; Hiraide et al., 2018, 2019; Krzyzewska et al., 2019; Roston et al., 2021). Recently, Weerts et al. (2021) performed a study evaluating the phenotypes of 36 cases with *SETD1B* variants. The authors reported 14 pathogenic variants and 10 probable pathogenic variants. Only one of these variants was inherited from an affected parent. All previous pathogenic variants in 12 individual cases were reported as de novo variants. To date, most pathogenic variants have been reported as de novo and heterozygous. Inheritance of the neurodevelopmental disorder has rarely been reported in association with the *SETD1B* gene.

We report a novel in-frame deletion variant of the *SETD1B* gene in a family with cases diagnosed with schizoaffective disorder and ASD without seizures and intellectual disability.

2 | METHODS

2.1 | Patients

In this study, we present a family consisting of consanguineous parents who died of cancer and their descendants. This family includes two cases diagnosed with ASD; six cases diagnosed with schizophrenia, bipolar disorder, or schizoaffective disorder; three cases diagnosed with cancer; and five cases who died of unknown causes in early childhood. Three affected members of this family agreed to genetic testing. Most of the remaining cases were not alive. After written informed consent, genomic DNA was extracted from blood leukocytes of two patients and their parents.

2.2 | Genetic testing

Genomic DNA was isolated from the patients' peripheral blood using the Roche High Pure FFPE DNA Isolation Kit according to the manufacturer's protocols. All coding regions of the patient's human genome were sequenced by whole exome sequencing analysis on the MGI DNBSEQ-G400 platform using the Twist Library Preparation EF Kit (Enzymatic Fragmentation) from Twist Bioscience (South San Francisco, USA).

2.3 | Variant analysis and classification

We analyzed the raw data using the Genomize® (https://seq.genomize.com) data analysis platform. We used filtering steps to identify pathogenic variants associated with clinical features as follows: (1) all missense, nonsense, frameshift, splice site, indel, in-frame, and synonymous variants and (2) variants with minor allele frequency < 1.0% in population studies (1000 Genome [1000G], ESP, ExAC, and Genome Aggregation Database [gnomAD]). The Genome Integrative Viewer was used to display the sequence data. We checked the novel variants in the databases of HGMD® and ClinVar (http://ncbi.nlm.nih.gov/clinvar). Pathogenicity of novel variants was interpreted using in silico analysis tools (Mutation Taster, Combined Annotation Dependent Depletion [CADD]). The American College of Medical Genetics and Genomics guidelines for pathogenicity classification of variants were followed (Richards et al., 2015). Segregation analysis was then performed on DNA samples from available family members.

3 | CASE REPORTS

3.1 | Case 1 (Generation III, Individual 5)

A 15-year-10-month-old boy presented to the Medical Genetics with a diagnosis of ASD and a family history of variable psychiatric disorders.

This case was born as a second child to nonconsanguineous Turkish parents after a 40-week pregnancy without any significant problems. His birth weight and length were 3000 g (− 0.89 standard deviation [SD]) and 49 cm (− 0.45 SD), respectively. He was hospitalized

in the neonatal intensive care unit for postnatal jaundice. He used his first word at 5 years of age and walked at 2.5 years of age. He was diagnosed with ASD at the age of 3 with symptoms of language delay, reluctance to social communication, decreased eye contact, stereotyped movements, and sensitivity to sounds and smells. After being diagnosed with ASD, he began receiving special education services and his symptoms improved with special education services. Case 1 (Generation III, Individual 5) presented with global developmental delay in early childhood. No causal explanation for the developmental delay was found in the patient's examinations, brain imaging, and electroencephalography (EEG). Developmental milestones were acquired during special education. No intelligence or learning problems developed during the follow-up of the case. Laboratory tests showed no abnormalities other than GH deficiency. MRI of the pituitary gland was normal. At the time of his application (2010), his height was 89 cm (SD − 2.45) and his bone age was 2 years behind. He has been on somatropin since 2010. He was also diagnosed with allergic asthma. There was no history of seizures in any period of his life.

At the last examination, he was continuing his high school education with a good grade. He had no difficulties related to learning difficulties in his school life. His active complaints consist of multiple psychiatric comorbidities. These psychiatric diagnoses and symptoms are as follows: symptoms of ASD (reluctance to social communication, intense sensitivity to sounds and smells, the behavior of closing his ears to some sounds, irritability when disturbed by smells and sounds, stereotypies such as clapping his hands), anxiety (social phobia and specific phobias), attention deficit and hyperactivity disorder, depressive symptoms, and suicidal thoughts and attempts.

3.2 | Case 2 (Generation III, Individual 4)

Case 2 was the older sister of Case 1. She was 24 years old and had a diagnosis of learning disability and schizoaffective disorder. Case 2 was born by cesarean section at 36 weeks gestation. No problems were described in the prenatal period. Her birth weight was 3200 g, and her birth length was 51 cm. At 18 months of age, she walked and used her first words. When she started primary school, she had problems with reading and writing. She was referred to the Child and Adolescent Psychiatry Department with complaints of learning difficulties, inattention, and hyperactivity. When she was 7 years old, she underwent brain MRI and EEG. All of these tests were within normal limits. At the same time, the Wechsler Intelligence Scale for Children-Revised (WISC-R) was administered. The verbal IQ score was 92, the performance IQ score was 82, and the total IQ score was 82. She was diagnosed with specific learning disability and attention-deficit/hyperactivity disorder and was referred for special education. And she was able to learn to read and write 3 years after starting special education. When she was 14 years old, she had an episode of mania with psychotic features, including delusions and visual and auditory hallucinations. Although her mood components decreased with treatment, her psychotic features persisted. Her treatment was continued with a diagnosis of schizoaffective disorder. In this episode, the brain MRI was again performed and no pathological findings were obtained. Her cognitive impairment worsened after the diagnosis of schizoaffective disorder. When the WISC-R was administered again at age 16, the verbal IQ score was 74, the performance IQ score was 62, and the total IQ score was 68.

3.3 | Case 3 (Generation II, Individual 13)

Case 3, 68 years old, is the father of Case 1 and Case 2. He is being treated with a diagnosis of schizoaffective disorder. His medications include quetiapine, risperidone, carbamazepine, and aripiprazole. He had no history of epileptic seizures in his life. He was diagnosed with leukemia. Besides Case 1 and Case 2, he has two other children from his first marriage. It has been reported that his son from this marriage has been diagnosed with schizoaffective disorder. And one of his two granddaughters was diagnosed with ASD and the other with bipolar disorder. When the family history was reviewed, it was reported that Case 3's mother died of leukemia and his father died of a brain tumor. It was reported that there was consanguinity between the parents of Case 3.

It was reported that five siblings of Case 3 died of unknown causes between the ages of 2 and 5. In addition, it was reported that two of his siblings were diagnosed with schizophrenia and one of his siblings was diagnosed with leukemia before they died. Only two of his siblings had no diagnosis of psychiatric disorders or cancer. The pedigree of this large family is shown in Figure 1.

3.4 | Genetic results

Given the predominance of psychiatric disorders in the family and the five siblings who died of unknown causes between the ages of 2 and 5 years, the family was

screened for balanced translocations or copy number variants by karyotype and microarray analysis prior to sequencing, and no clinically relevant pathogenic variants were detected.

We identified a heterozygous in-frame deletion variant on chromosome 12, within the *SETD1B* gene c.2755_2778del (NM_001353345.1) in two siblings. This heterozygous likely pathogenic variant resulted in an in-frame deletion (p.Glu919_Asp926del) and is not present in the gnomAD database. We checked the novel variants in the databases of HGMD® and ClinVar (http://ncbi.nlm.nih.gov/clinvar). This variant is considered likely to be pathogenic based on the criteria of Richards et al. (2015)). Parental samples were also analyzed specifically for the variants detected in the patients. The father was heterozygous for the same variant.

The detected *SETD1B* variant was confirmed by Sanger sequencing in the healthy mother and in the cases with the disease. Sanger sequencing images are shown in Figure 2.

4 | DISCUSSION

Up to now, 20 different variants in the *SETD1B* gene (missense/nonsense, splice site, small deletion, gross deletion, small insertion/duplication) have been reported in Human Gene Mutation Database® (HGMD®) Professional (https://portal.biobase-international.com/hgmd/pro/all.php). In the previous literature, most of the reports presenting pathogenic variants of the *SETD1B* gene found similar phenotypic characteristics including

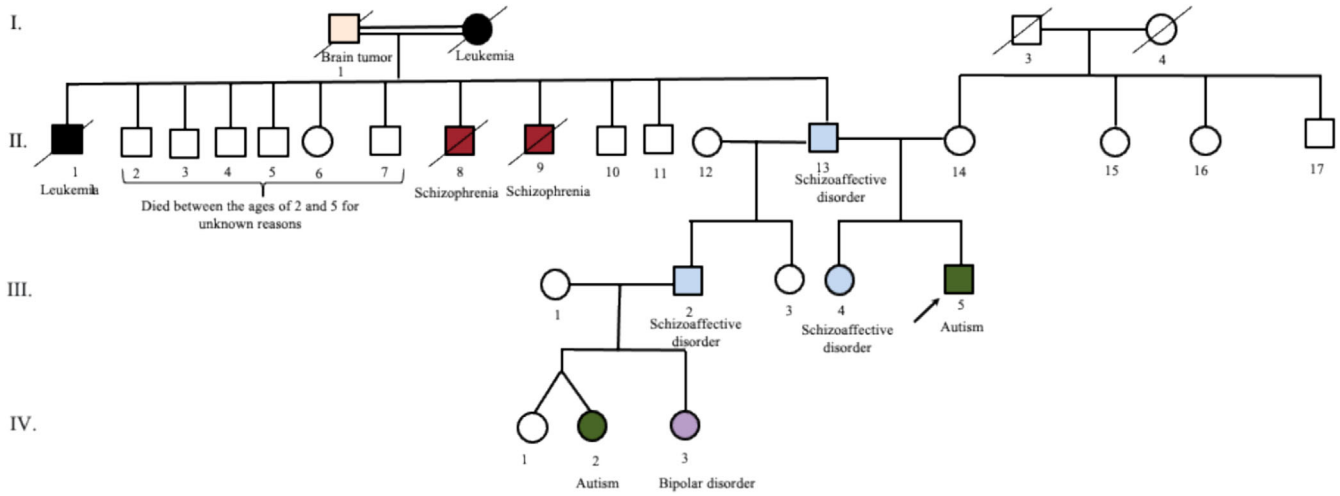

FIGURE 1 Pedigree of the cases carrying *SETD1B* variant (cases performed genetic test and detected *SETD1B* (NM:001353345.1: c.2755_2778del) variant: Generation III, Individual 4; Generation III, Individual 5, Generation II, Individual 13.

FIGURE 2 Sanger sequencing for *SETD1B* (NM:001353345.1:c.2755_2778del) variant.

intellectual developmental disorder with seizures and language delay. In this study, we report an in-frame deletion variant of the *SETD1B* gene in a family including cases diagnosed with schizoaffective disorder and ASD without seizures and intellectual disability. In addition, this family consisted of descendants of consanguineous parents who died of cancer. This family included two cases diagnosed with ASD; six cases diagnosed with schizophrenia, bipolar disorder, or schizoaffective disorder; three cases diagnosed with cancers; and five cases died of unknown reasons in early childhood. This *SETD1B* gene variant was presented in only nine individuals in the gnomAD v4 database. To our knowledge, our cases are first cases carrying this *SETD1B* gene variant reported in the Turkish population.

Our cases have not presented seizures or EEG and brain MRI abnormalities. To date, most cases were presented with seizures starting before 4 years of age and difficulty responding to treatment despite multiple medications (Roston et al., 2021). Prior to the study conducted by Weerts et al., all reported cases carrying the pathogenic variant of *SETD1B* gene suffered from seizures (Weerts et al., 2021). In this recent study, the seizure rate was reported to be 78% (28/36) with a median age of seizure onset of 3 years. Consistent with our findings, cases without seizures were found in this cohort. Similar to previous literature, brain MRI findings in our cases were unremarkable. Individual pathogenic *SETD1B* variants have been associated with varying degrees of intellectual disability, often including language delay. Weerts et al. (2021) reported intellectual disability in 28 cases (28/32, 88%) (Weerts et al., 2021).

Symptoms of ASD were reported in approximately 75% of cases carrying pathogenic *SETD1B* variants in the literature (Roston et al., 2021). In addition to ASD, we found schizoaffective disorder in two cases carrying variants of the *SETD1B* gene. SETD1A is another member of the histone methyltransferase family. SETD1B is paralogous to SETD1A, and they share phenotypic similarities, including neurodevelopmental disorders (Kummeling et al., 2021). Rare variants in the SETD1A gene have been reported in large cohorts of schizophrenia with a rate of 0.13% (Singh et al., 2016). The authors also suggested that chromatin modification, including histone H3 methylation, plays an important role in the etiology of schizophrenia. In one study evaluating patients with schizophrenia, a *SETD1B* variant was reported, but phenotypic features were lacking (Wang et al., 2015).

In the previous literature, only one of the variants was inherited from an affected parent. Most of the previous pathogenic variants were reported as de novo variants. In our study, the phenotypic features were found to be inherited for at least three generations in the family we presented, and it was shown that the pathogenic variant of the *SETD1B* gene was transmitted from the affected parent to his affected children.

Pathogenic variants of H3K4 methyltransferases have been reported to play role in different types of malignancies. Although there are limited reports including *SETD1B* variants, it was associated with colon cancer, oesophageal squamous cell carcinoma, endometrial carcinoma and polycythemia vera (Choi et al., 2014; García-Sanz et al., 2017; Song et al., 2014; Tiziana Storlazzi et al., 2014). Functional studies have also provided important information on the role of *SETD1B* in hematopoiesis and its possible contribution to etiology of leukemia (Schmidt et al., 2018; Yang & Ernst, 2017). Further studies are needed to demonstrate the clinical impact of *SETD1B* similar to other H3K4 methyltransferases. In our study, genetic analysis was not possible for family members who died from malignancies.

However, it is noteworthy that three individuals in this family were diagnosed with malignancies and two of them were diagnosed with leukemia. In our study, the father carrying the *SETD1B* variant was diagnosed with both schizoaffective disorder and leukemia.

Case 3 (Generation II, Individual 13), for whom genetic testing was performed in the family, was diagnosed with leukemia at the age of 66. There were also other cases diagnosed with cancer in the family (Generation I, Individual 1; Generation I, Individual 2; Generation II, Individual 1). However, it was not possible to obtain the age at cancer diagnosis and other detailed clinical information of these cases and to perform genetic testing.

We proposed an association between rare variants of *SETD1B* and phenotypes of ASD and schizoaffective disorder without seizures and intellectual disability. The *SETD1B* gene is included in both the ASD genetic database of SFARI (https://gene.sfari.org/) and the cancer database of COSMIC (https://cancer.sanger.ac.uk/cosmic). However, there are very few reports of *SETD1B* gene variants as clinical entities. To our knowledge, the *SETD1B* gene variant has not been previously reported in an individual diagnosed with both a neuropsychiatric disorder and cancer. We suggest that it should be considered as a risk factor for leukemia and other cancers when examining the distribution of disease in this family and previous animal studies. Further studies are needed to investigate this association.

Our study has several limitations. We identified a large family with complex phenotypes, including autism, psychiatric illness, and cancer in the pedigree. However, we were able to include four family members (two children and their two parents) in this study because other family members did not agree to undergo genetic testing.

For this reason, although there are several cases with the disease in the extended family, the genetic data are limited to the four cases from the family.

AUTHOR CONTRIBUTIONS

Hilmi Bolat and Gül Ünsel-Bolat wrote the main part of the manuscript. Hilmi Bolat performed and interpreted the genetic tests. Gül Ünsel-Bolat collected and interpreted the clinical data. Both authors critically revised and approved the final version of the manuscript.

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CONFLICT OF INTEREST STATEMENT

The authors have no competing interests to declare.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

ETHICS STATEMENT

Ethics committee approval was obtained from the Balıkesir University Faculty of Medicine Ethics Committee (2021/98-14/01/2021).

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