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Original Article

Recognizing Stuve–Wiedemann syndrome in childhood: clinical insights from 11 patients with founder and novel *LIFR* variants

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ABSTRACT

Background. Stuve–Wiedemann syndrome is a rare autosomal recessive bent-bone dysplasia characterized by dysautonomia and distinctive skeletal abnormalities, most commonly caused by biallelic *LIFR* variants.

Methods. Eleven patients from ten unrelated families with molecularly confirmed Stuve–Wiedemann syndrome were evaluated. Detailed demographic, perinatal, clinical, radiographic, and molecular data were collected. All patients underwent Sanger sequencing of *LIFR*.

Results. Respiratory problems and episodic hyperthermia were present in all patients, while feeding difficulties, hypotonia, growth failure, developmental delay, craniofacial dysmorphism, oral abnormalities, and ocular involvement were also common. All patients demonstrated long-bone bowing, cortical thickening, and flared metaphyses. A biallelic *LIFR* variant was identified in all patients, occurring in the homozygous state in nine patients and in the compound heterozygous state in two patients. The recurrent c.2074C>T; p.(Arg692Ter) variant was homozygous in seven patients and compound heterozygous in one patient. Three novel *LIFR* variants were also identified.

Conclusion. Stuve–Wiedemann syndrome is characterized by a high mortality rate during the first two years of life due to severe dysautonomia, and orthopedic complications worsen progressively with age; therefore, early diagnosis and lifelong multidisciplinary follow-up are essential. Furthermore, the high rate of consanguinity in Türkiye, together with the recurrent p.(Arg692Ter) variant in *LIFR* identified in multiple unrelated families, supports the possibility of a founder effect and suggests that the frequency of Stuve–Wiedemann syndrome in our population may be higher than expected. Our findings further characterize the previously reported clinical manifestations of Stuve–Wiedemann syndrome while expanding the molecular spectrum through the identification of three novel *LIFR* variants.

Key words: Stuve-Wiedemann syndrome, LIFR, dysautonomia, skeletal dysplasia.

Stuve–Wiedemann syndrome (SWS; OMIM #601559), also referred to as neonatal Schwartz–Jampel syndrome type 2 (SJS2), is a rare autosomal recessive disorder characterized by dysautonomia, episodic hyperthermia, respiratory insufficiency, feeding difficulties, and distinctive skeletal abnormalities. The disorder was first described in 1971 by Stüve and Wiedemann in two female siblings and a male cousin who exhibited congenital bowing of the long bones, hand anomalies, craniofacial abnormalities, and respiratory distress, suggesting autosomal recessive inheritance.[1]

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SJS, also known as chondrodystrophic myotonia, is primarily defined by myotonia and characteristic skeletal abnormalities. A distinct subgroup presenting at birth with prominent skeletal features, including fetal hypokinesia, congenital contractures, shortening and bowing of the long bones, undertubulation of the metaphyses, and osteoporosis, has been described as SJS type 2 (a neonatal-onset form with Pyle disease–like bone dysplasia).[2] Notably, beyond its clinical characteristics, this subgroup has been shown to lack genetic linkage to the classical SJS locus on 1p36–34.[2] Subsequent studies showed that SJS type 2 and SWS share essentially the same clinical spectrum with identical skeletal features and differ only by the presence of myotonia on electromyography in SJS, leading to the conclusion that these two conditions represent a single entity.[3]

In 2004, a study involving 19 affected families demonstrated that null mutations in the leukemia inhibitory factor receptor (LIFR) gene (OMIM *151443) located on 5p13.1 are responsible for the disorder, thereby providing molecular confirmation that the two entities are indeed the same condition.[4] Furthermore, SWS patients negative for *LIFR* mutations have been identified, and in this clinically and molecularly heterogeneous disorder, biallelic loss-of-function variants in *IL6ST* (OMIM *600694) have also been shown to cause the phenotype, leading to the designation of SWS type 2 (OMIM #619751).

It has been reported that while the risk of death due to dysautonomia is high during infancy, this risk decreases in childhood, during which orthopedic complications begin to predominate in the clinical course. Early recognition of SWS is crucial, as timely diagnosis enables appropriate management of dysautonomia and prevention of progressive orthopedic complications.

To date, more than 90 patients with SWS have been reported in the literature.[5-20] Here, we report 11 patients from 10 unrelated families diagnosed with SWS, providing detailed clinical, radiographic, and molecular characterization to further delineate the disorder.

Methods

A total of 11 patients with a molecularly confirmed diagnosis of SWS who were followed at a single tertiary referral center were included in this study. Detailed demographic and clinical data, including perinatal features, respiratory problems, feeding difficulties, dysautonomic episodes, growth and developmental outcomes, skeletal abnormalities, and other associated comorbidities, were collected.

Peripheral blood samples were obtained from all patients and available parents. Genomic DNA was extracted using the standard salt precipitation method after written informed consent had been obtained. All patients underwent Sanger sequencing of the LIFR gene (NM_001127671.2). Specific primers were used to amplify all coding exons and exon–intron boundaries by polymerase chain reaction (PCR). PCR products were subjected to capillary electrophoresis on an ABI 3500 Genetic Analyzer (Thermo Fisher Scientific, Waltham, Massachusetts, USA). Sequence chromatograms were evaluated using standard sequencing analysis software, and genotypes were determined accordingly. Primer pairs are available upon request.

Variants were classified according to the ACMG guidelines using available databases including ClinVar, gnomAD, HGMD, and in silico prediction tools such as MutationTaster, SIFT, and PolyPhen.[21] Segregation analysis was performed whenever parental samples were available.

Statistical analysis

Given the descriptive nature of the study, only descriptive statistical analyses were performed. Continuous variables are presented as median (range), whereas categorical variables are presented as numbers and percentages.

The study was approved by Hacettepe University Ethics Committee (SBA 25/1013, date: 17/12/2025).

Results

Demographic and clinical findings

A total of 11 patients from 10 families, including eight females and three males, were included in the study. The demographic, clinical, and molecular characteristics of the patients are summarized in Table I. Consanguinity was present in 6 of 10 families (60%), and two additional families originated from the same village. A history of sibling death was reported in five (50%) families. The median age at diagnosis was 12 months, ranging from postnatal day 1 to 4 years. Patient 4 died at 7 months of age due to sudden infant death, whereas Patient 6 died suddenly at 4 years of age, with a presumed cerebrovascular event as the cause of death. Prenatal abnormalities were documented in six patients, most commonly limb shortening/bowing and decreased fetal movements.

Respiratory problems and episodic hyperthermia were observed in all patients, while feeding difficulties were present in 10 of 11 patients (90.9%), hypotonia in 9 of 10 patients (90%), and growth failure in 9 of 10 patients (90%). Developmental delay was present in the majority of patients (80%), manifesting as isolated speech delay, motor delay, or global developmental delay. Characteristic craniofacial findings were variably observed and pursed lips were noted in all evaluated patients (10/10, 100%) (Fig. 1). Oral/dental abnormalities were observed in all evaluated patients (9/9, 100%), including oligodontia, multiple dental caries, dry mouth, and atrophy of the lingual papillae. Ocular involvement was frequent (9/10, 90%) and included keratitis, corneal opacity, dry eye, refractive errors, and strabismus.

Radiological and skeletal findings

Among the skeletal findings, bowing of the long bones, cortical thickening, and flared metaphyses were present in all patients, whereas camptodactyly was observed in 8 of 11 patients (72.7%), joint contractures in 9 of 11 (81.8%), scoliosis in 8 of 10 (80%), and bone fractures in 6 of 9 (66.6%) (Fig. 1 and Fig. 2).

Molecular findings

Molecular analysis identified biallelic *LIFR* variants in all patients. The recurrent pathogenic variant c.2074C>T; p.(Arg692Ter) was identified in the homozygous state in patients from seven families, whereas in one patient it was detected in the compound heterozygous state in combination with an additional variant. Overall, the c.2074C>T; p.(Arg692Ter) variant accounted for 15 of 22 disease-associated alleles (68.2%) in the cohort. Moreover, three previously unreported variants in the *LIFR* gene were identified, comprising one nonsense variant, one missense variant, and one frameshift

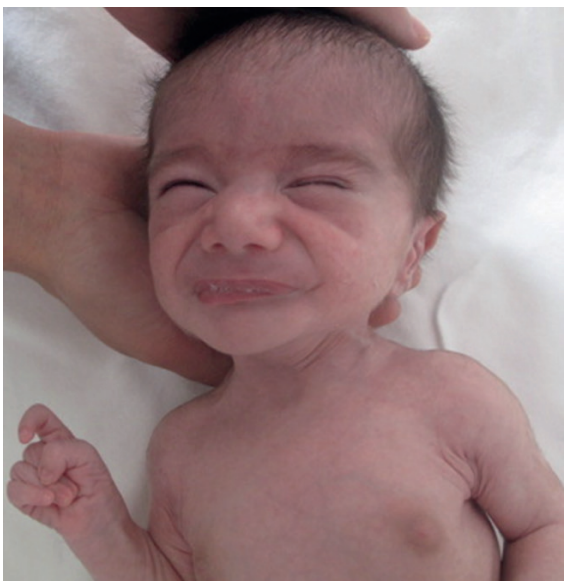

Fig. 1. Clinical image of Patient 2 at 1 month of age showing pursed lips and camptodactyly.

Table I. Demographic, clinical, and molecular characteristics of patients with Stuve-Wiedemann syndrome.

	Family	F1		F2	F3	F4	F5	F6	F7	F8	F9	F10
	Patient ID	P1	P2	P3	P4	P5	P6	P7	P8	P9	P10	P11
Demographic features	Sex	F	M	F	F	F	F	F	M	F	M	F
	Consanguinity	+	+	+	-	- (Same village)	+	+	+	+	-	- (Same village)
	History of sibling death	-	-	+	+	+	-	+	-	-	+	-
	Age at diagnosis	12 months	3 years	1.5 years	Day 1	4 years	6 months	3 months	3 years	4 months	4 months	2 years
	Survival status	Alive	Alive	Alive	Died at 7 months	Alive	Died at 4 years	Alive	Alive	Alive	n.a.	Alive
Clinical findings	Perinatal features	Increased nuchal translucency, and lower limb bowing and shortening	Limb shortening, and decreased fetal movements	Oligohydramnios, limb shortening, and decreased fetal movements	Limb bowing	- (No prenatal care)	Femoral shortening, and decreased fetal movements	Oligohydramnios, short femoral neck, and decreased fetal movements	-	- (No prenatal care)	- (No prenatal care)	-
	Hypotonia	+	+	+	+	+	+	+	+	-	n.a.	+
	Respiratory problems	+	+	+	+	+	+	+	+	+	+	+
	Feeding difficulty	+	+	+	+	+	+	+	-	+	+	+
	Episodic hyperthermia	+	+	+	+	+	+	+	+	+	+	+
	Growth failure	+	+	+	+	+	+	+	-	+	n.a.	+
	Developmental delay	-	Motor delay	Motor delay	Motor delay	-	Global delay	Global delay	Speech delay	Motor delay	n.a.	Motor delay
	Facial dysmorphism	Bitemporal narrowing, depressed nasal bridge, microretrognathia, and short neck	Depressed nasal bridge and microretrognathia	Bitemporal narrowing, depressed nasal bridge, and short neck	-	Narrow forehead with frontal hypertrichosis	Microretrognathia, prominent metopic suture, frontal bossing, depressed and broad nasal bridge, everted lower lip, high-arched palate	Mild frontal bossing, midfacial hypoplasia, incomplete eyelid closure, bulbous nasal tip, prognathism, short neck	Frontal bossing, hypertrichosis, synophrys, deep-set eyes, bulbous nasal tip, high and narrow palate	Micrognathia	Dolichocephaly	Blepharophimosis, microstomia, micrognathia
	Pursed lips	+	+	+	+	+	+	+	+	+	n.a.	+
	Oral/dental findings	Oligodontia, early dental caries, and atrophy of the lingual papillae	Oligodontia, premature tooth loss, dry mouth, and atrophy of the lingual papillae	Oligodontia, and atrophy of the lingual papillae	n.a.	Premature tooth loss, orthodontic problems and atrophy of the lingual papillae	Malocclusion, atrophy of the lingual papillae	Premature tooth loss, malocclusion, dry mouth, and atrophy of the lingual papillae	Multiple dental caries, dry mouth and atrophy of the lingual papillae	Multiple dental caries, premature tooth loss, and dry mouth	n.a.	Multiple dental caries and premature tooth loss
	Ocular findings	Keratitis, corneal opacity and dry eye	Keratitis, corneal opacity and dry eye	Hypermetropia, keratitis, corneal opacity, dry eye, and strabismus	Dry eye	-	Keratitis, corneal opacity and dry eye	Myopia, keratitis, corneal opacity and dry eye	Keratitis, corneal opacity, and strabismus	Keratitis, corneal opacity, and dry eye	n.a.	Keratitis, corneal opacity, and dry eye

ASD: Atrial septal defect, AR: Aortic regurgitation, F: Female, M: Male, MR: Mitral regurgitation, MVP: Mitral valve prolapse, n.a.:Not available, PAH: Pulmonary arterial hypertension, PEV: Pes equinovarus, PFO: Patent foramen ovale, TR: Tricuspid regurgitation, VUS: Variant of Uncertain Significance. The novel variants are indicated in bold.

Table I. Continued.

	Family	F1		F2	F3	F4	F5	F6	F7	F8	F9	F10
	Patient ID	P1	P2	P3	P4	P5	P6	P7	P8	P9	P10	P11
Skeletal features	Camptodactyly	+	+	-	+	+	-	+	-	+	+	+
	Joint contracture	+	+	+	+	+	-	+	-	+	+	+
	Scoliosis	+	+	+	-	+	+	+	-	+	n.a.	+
	Osteoporosis/ Bone fracture	+/+	n.a. / +	+ / -	n.a. / n.a.	+/+	+ / +	+ / +	+ / -	+ / -	n.a. / n.a.	+/+
	Bowing of the long bones	+	+	+	+	+	+	+	+	+	+	+
	Cortical thickening of the long bones	+	+	+	+	+	+	+	+	+	+	+
	Wide, flared metaphyses	+	+	+	+	+	+	+	+	+	+	+
	Anesthetic complications	-	-	-	-	-	Respiratory distress after anesthesia	-	-	-	n.a.	-
	Other clinical findings	PEV	Morgagni hernia	Moderate TR, papular skin lesions	-	Papular skin lesions	Microcephaly, mildly thin corpus callosum, PAH, MVP, mild-to-moderate MR, PFO and PEV	Hearing loss, minimal AR, bicuspid aortic valve, and aortic root dilatation	Microcephaly, small ASD, PFO	-	Seizures	-
Molecular findings	LIFR variant NM_001127671.2	c.2074C>T, p.(Arg692Ter)	c.2074C>T, p.(Arg692Ter)	**c.823G>T, p.(Glu275Ter)**	c.274C>T, p.(Gln92Ter) / **c.1429delC, p.(Gln477LysfsTer11)**	c.2074C>T, p.(Arg692Ter)	c.2074C>T, p.(Arg692Ter)	c.2074C>T, p.(Arg692Ter) / c.1105_1106insT, p.(Tyr369LeufsTer5)	**c.890T>A, p.(Val297Asp)**	c.2074C>T, p.(Arg692Ter)	c.2074C>T, p.(Arg692Ter)	c.2074C>T, p.(Arg692Ter)
	Zygosity	Homozygous	Homozygous	Homozygous	Compound heterozygous	Homozygous	Homozygous	Compound heterozygous	Homozygous	Homozygous	Homozygous	Homozygous
	ACMG Classification	Pathogenic	Pathogenic	Likely pathogenic	Likely pathogenic / Likely pathogenic	Pathogenic	Pathogenic	Pathogenic / Likely pathogenic	VUS	Pathogenic	Pathogenic	Pathogenic

ASD: Atrial septal defect, AR: Aortic regurgitation, F: Female, M: Male, MR: Mitral regurgitation, MVP: Mitral valve prolapse, n.a.:Not available, PAH: Pulmonary arterial hypertension, PEV: Pes equinovarus, PFO: Patent foramen ovale, TR: Tricuspid regurgitation, VUS: Variant of Uncertain Significance. The novel variants are indicated in bold.

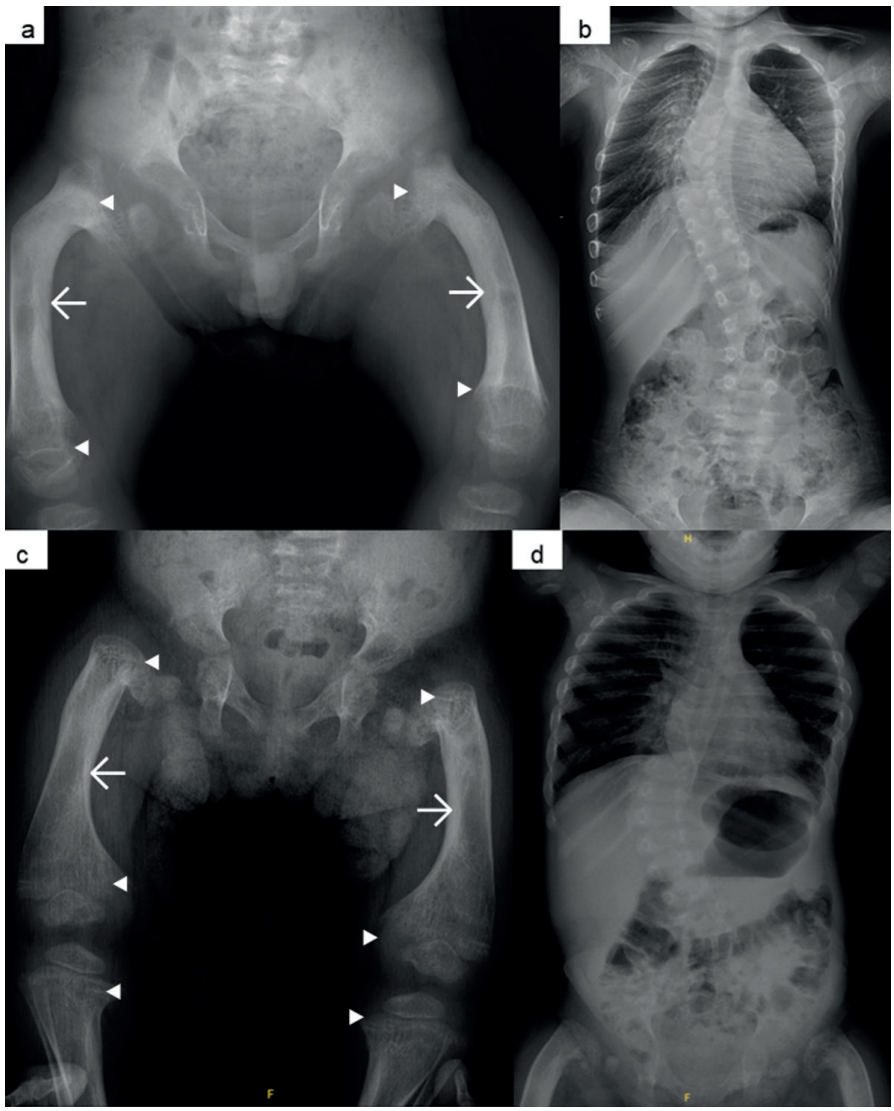

Fig. 2. Radiographs of Patients 2 and 3 demonstrate bowing of the long tubular bones, cortical thickening (arrows), and wide, flared metaphyses (arrowheads) (a: Patient 2 at 5 years; c: Patient 3 at 2 years), as well as scoliosis (b: Patient 2 at 6 years; d: Patient 3 at 2 years).

variant. Among the novel variants detected in our cohort, c.890T>A, p.(Val297Asp), was classified as a variant of uncertain significance (VUS) based on ACMG guidelines; however, several lines of evidence raised the possibility of pathogenicity. This variant is not present in the gnomAD database (PM2_Supporting), and in silico prediction tools suggest a deleterious impact on protein function, as supported by AlphaMissense (score: 0.754; PP3_Supporting) and PolyPhen-2, which classifies it as probably damaging (score: 1.000). In addition, segregation analysis supported an autosomal recessive inheritance pattern, and the patient's phenotype was strongly consistent with SWS, with no alternative molecular diagnosis identified. The novel variants were submitted to the ClinVar database (SUB16043420, SUB16045977, SUB16046006), and all identified variants were marked on the schematic representation of the LIFR protein (Fig. 3).

Fig. 3. Schematic representation of the LIFR protein domains and the distribution of pathogenic variants identified in our cohort. The LIFR protein consists of a signal peptide (SP), cytokine receptor homology domains (CRH1 and CRH2), an immunoglobulin-like domain (Ig), fibronectin type III domains (FNIII), a transmembrane domain (TM), and a cytoplasmic domain (CD). The positions of the variants identified in this study are indicated above the protein schematic. Novel variants identified in the present study are indicated in bold and marked with an asterisk (*).

Discussion

SWS is a rare autosomal recessive disorder characterized by skeletal abnormalities and severe dysautonomic manifestations, particularly during early childhood, whereas orthopedic complications become more prominent with age.[5,9] All patients in our cohort demonstrated the characteristic skeletal phenotype of SWS, including universal bowing of the long bones and frequent camptodactyly, accompanied by varying degrees of autonomic dysfunction (Table I).

Short and bowed limbs have been reported in all patients with SWS, and camptodactyly has been described in approximately 80% of cases.[5] Consistent with these observations, bowing of the long bones was present in all of our patients, while camptodactyly was observed in 72.7% of the cohort. Characteristic radiological findings, including bowed long bones with cortical thickening, enlarged metaphyses, and an abnormal trabecular pattern, as well as scoliosis and osteoporosis-related fractures observed in our patients were consistent with the previously reported clinical spectrum and natural course of the disorder.[5]

Prenatal warning signs include growth restriction, oligohydramnios, reduced fetal movements, and characteristic skeletal findings such as short and bowed long bones, pronounced tibial bowing (greater than femoral involvement), relative sparing of the fibula and upper limb bones, camptodactyly, and talipes.[8,22] It has been emphasized that, in the prenatal differentiation of SWS from other bent-bone disorders, the presence of camptodactyly together with a normal scapula is an important distinguishing feature.[22] In our cohort, six prenatally monitored patients showed findings such as increased nuchal translucency, limb shortening, decreased fetal movements, and oligohydramnios. However, none of the patients received a molecularly confirmed diagnosis of SWS during the prenatal period. This may be attributed to the relatively nonspecific prenatal skeletal findings of SWS, the absence of severe abnormalities suggestive of a lethal skeletal dysplasia leading to limited prenatal recognition, restricted access to prenatal genetic testing, and the lack of a previously diagnosed affected sibling in the family, which may have reduced the likelihood of recommending molecular analysis.

A narrow thorax, one of the main prenatal indicators of lethality in skeletal dysplasias, is typically absent in SWS. Nevertheless, infant mortality remains high because of dysautonomic complications.[8] The disorder is considered potentially fatal before the age of 2 years, primarily because of respiratory distress and episodic hyperthermia. Sudden death and anesthesia-related malignant hyperthermia have also been reported. Mortality is notably high, estimated at approximately 46% overall,

reaching 42% in patients younger than 2 years and 10% in older individuals.[5] In our cohort, two patients died, one following a presumed cerebrovascular event and one because of sudden death. In addition, Patient 4 had two siblings who had previously died with similar clinical features. Anesthetic complications were also documented in one of our patients who subsequently died (Table I, Patient 6). Additionally, pulmonary arterial hypertension, which has been suggested to be associated with poor prognosis, was present in one of our patients, who died at 4 years of age.[5]

Additional features associated with the disorder include corneal involvement, oral and dental abnormalities, and dysmorphic facial features, particularly the characteristic pursed-mouth appearance, as well as neurological manifestations such as hypotonia, seizures, and delayed motor development.[5] Ectodermal findings such as milia, multiple eruptive vellus hair cysts, and dystrophic nails have been reported, and similarly, papular skin lesions were noted in two of our patients.[7] One of our patients was found to have a Morgagni hernia; to the best of our knowledge, this association has not previously been reported and may represent a coincidental finding.

Although the disorder has been reported more frequently in Arab populations, with an estimated prevalence of 0.52 per 10,000 births, recent reports suggest that it may not be as rare in Türkiye as previously assumed.[23,24] This distribution may be related to the high rate of consanguinity in these populations, which increases the prevalence of autosomal recessive disorders. A systematic review published in 2022 reported consanguinity in 65% of families with SWS.[5] In our cohort, the corresponding rate was 60%, further supporting the contribution of consanguinity to disease occurrence. In addition, a founder variant in the Turkish population has been proposed in the *LIFR* gene, and this variant, c.2074C>T; p.(Arg692Ter), was homozygous in seven of our patients, while one patient carried it in trans with another pathogenic *LIFR* variant, consistent with a compound heterozygous genotype.[23] Notably, the fact that this variant accounted for the majority of disease-associated alleles in our cohort and was identified in apparently unrelated families further supports the possibility of a founder effect in the Turkish population. Although this variant has also been reported in other populations, it has been predominantly described in Turkish patients.[19,23,25,26] Moreover, a previous study demonstrated a shared haplotype spanning the *LIFR* locus among affected individuals, supporting a possible founder effect.[23] However, future haplotype analyses would be required to confirm a shared ancestral origin.

Null mutations in *LIFR* disrupt the JAK/STAT3 signaling pathway, and studies in Lifr−/− mice have demonstrated impaired bone remodeling, reduced astrocyte numbers, and perinatal lethality.[4,27] Similar to *CRLF1*-related cold-induced sweating syndrome 1 (Crisponi syndrome; OMIM #272430), SWS belongs to the spectrum of ciliary neurotrophic factor receptor (CNTFR) pathway–related disorders characterized by autonomic nervous system dysfunction.[11,27,28] In the differential diagnosis of SWS, Crisponi syndrome is an important consideration, as it shares multiple clinical features, such as episodic hyperthermia, feeding difficulties, respiratory problems, facial trismus, and camptodactyly. However, bowing of the long bones is highly characteristic of SWS and serves as a key distinguishing feature. Owing to this hallmark feature, SWS is classified within Group 20, the bent-bones dysplasia group, in the current Nosology of genetic skeletal disorders.[29] Nevertheless, the presence of dysautonomic features distinguishes SWS from other entities within this group.

This study has several limitations. First, the sample size was small, which is expected for a rare disorder but limits the generalizability of our findings. Second, the retrospective design resulted in incomplete data for some variables, as reflected by the variable denominators across clinical features. Third, the cohort was drawn from a population with a high rate of consanguinity, which may have influenced

both the frequency of homozygous variants and the apparent predominance of the recurrent p.(Arg692Ter) allele. Finally, although three previously unreported *LIFR* variants were identified, no functional studies were performed; therefore, their pathogenicity relies on ACMG-based interpretation, segregation data, and phenotype correlation.

In conclusion, SWS should be considered in children with dysautonomic manifestations accompanied by long-bone bowing, camptodactyly, and characteristic craniofacial features. Early recognition is essential for appropriate orthopedic surveillance, timely physical therapy, and surgical interventions when necessary. Early calcium and vitamin D supplementation may also help prevent osteoporosis-related fractures. Equally important is the careful assessment of dysautonomia-related symptoms and the prevention of anesthesia-associated complications, including malignant hyperthermia, which may be life-threatening. Our cohort further delineates the phenotypic and molecular spectrum of the disorder and underscores the recurrent p.(Arg692Ter) *LIFR* variant in the Turkish population.

Ethical approval

The study was approved by Hacettepe University Ethics Committee (date: December 17, 2025, number: SBA 25/1013).

Author contribution

The authors confirm contribution to the paper as follows: Study conception and design: GÜD, GEU, PÖŞK; data collection: GÜD, NBA; analysis and interpretation of results: GÜD, NBA, GEU, PÖŞK; draft manuscript preparation: GÜD, NBA. All authors reviewed the results and approved the final version of the manuscript.

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The authors declare the study received no funding.

Conflict of interest

The authors declare that there is no conflict of interest.

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