JIG Journal of Interdisciplinary Genomics

https://doi.org/10.22742/JIG.2026.8.2.54
Journal of Interdisciplinary Genomics 2026;8(2):54-59
eISSN 2671-6771

Monogenic Causes of Short Stature in Children Initially Diagnosed with Idiopathic Short Stature

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Short stature is a common reason for referral to pediatric endocrinologists, yet its underlying cause often remains unidentified despite comprehensive clinical and biochemical evaluation. Recent advances in molecular genetic testing have revealed that a substantial proportion of children previously classified as having idiopathic short stature harbor pathogenic genetic variants. This review summarizes current genetic approaches to short stature and highlights major genes involved in growth plate regulation and the growth hormone–insulin-like growth factor 1 axis. Variants in genes such as SHOX, ACAN, NPR2, NPPC, IHH, and FGFR3 can disrupt growth plate chondrogenesis, whereas defects in GH1, GHR, IGF1, and IGF1R affect the GH–IGF-1 pathway. In addition, mild syndromic disorders, including those associated with PTPN11 variants, may present primarily as short stature. Phenotypic features, including body disproportion, bone age, biochemical findings, and family history, can provide important clues for selecting appropriate genetic tests. However, because many affected children lack distinctive clinical features, multigene panels and exome sequencing are increasingly important diagnostic tools. Establishing a molecular diagnosis may reduce unnecessary investigations, improve prognostic assessment, guide treatment decisions, and enable more individualized management. Genetic testing is therefore likely to become an increasingly integral component of the evaluation of children with unexplained short stature.

Key words: Short stature, Idiopathic short stature, Genetic testing, Next-generation sequencing, Growth disorders, High-Throughput Nucleotide Sequencing

REVIEW ARTICLE

Received: October 3, 2026
Revised: October 23, 2026
Accepted: October 28, 2026

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INTRODUCTION

Short stature (SS) stands as the leading reason children are referred to pediatric endocrinologists. It is conventionally defined as a height falling more than 2 standard deviations below the age-, sex-, and population-specific mean—a threshold that applies to approximately 2.3% of children [1]. Height is shaped by a range of genetic contributors, including chromosomal anomalies, pathogenic single-gene variants, and polygenic predisposition [2]. To pinpoint an underlying cause, children with SS are generally subjected to thorough clinical, biochemical, and imaging assessments. Nonetheless, a considerable number remain without a definitive diagnosis even after exhaustive evaluation [3]. This group spans both complex cases—marked by skeletal abnormalities, dysmorphic traits, neurodevelopmental delay, or congenital anomalies—and those whose short stature occurs in isolation, without any other notable clinical findings. An increasing body of research points to a genetic basis underlying many of these cases [4]. Monogenic etiologies of growth failure have been well documented for years, particularly among GH–IGF axis abnormalities, skeletal dysplasias, and syndromic disorders. Advances in molecular genetic techniques over the past three decades have markedly widened the scope of diagnostic evaluation, making genetic testing more accessible, affordable, and feasible for routine clinical use [5]. Early approaches centered on analyzing individual genes, but the advent of next-generation sequencing (NGS)—

encompassing exome sequencing (ES) and genome sequencing (GS)—has transformed genetic diagnosis across virtually every medical field. Notably, these technologies, previously confined largely to research applications, are now being incorporated with growing frequency into routine clinical care [6]. Against this backdrop, a considerable number of investigations have examined how effective genetic testing is at establishing a diagnosis in children presenting with short stature of unexplained origin [7,8]. This review aims to provide an overview of the genetic causes of short stature, with a particular focus on genes associated with growth disorders and their clinical phenotypes. In addition, we discuss the clinical implications of genetic testing in children with short stature to facilitate appropriate genetic evaluation and diagnosis (Fig. 1).

Exploring genetic approaches to short stature

Conventional diagnostic approaches to short stature in children have primarily emphasized hormonal abnormalities, chronic illness, and a limited set of genetic conditions. Earlier studies, however, suggest that routine extensive screening offers relatively low yield for identifying an underlying cause in children with short stature who otherwise appear asymptomatic [9]. Genetic causes of linear growth disorders identified to date are remarkably heterogeneous, involving a diverse array of cellular pathways; broadly speaking, these variants exert their effects by disrupting chondrogenesis at the growth plate [10].

When biochemical workup proves unremarkable and no distinctive phenotype is apparent, pinpointing the cause of short stature through history and physical examination alone becomes challenging. Under these circumstances, clinicians often turn to genetic testing—karyotyping to evaluate for Turner syndrome, chromosomal microarray to detect structural chromosomal abnormalities, targeted gene panels when specific genes or associated features are suspected, and ES for more extensive genetic evaluation. Beyond parental height measurements, evaluation of the extended family pedigree can assist in differentiating monogenic from oligogenic or polygenic etiologies, particularly in cases where one parent is also short-statured or where short stature follows an X-linked inheritance pattern among male relatives.

- When both parental heights fall in a low percentile and the patient's height deficit mirrors parental height (or predict-

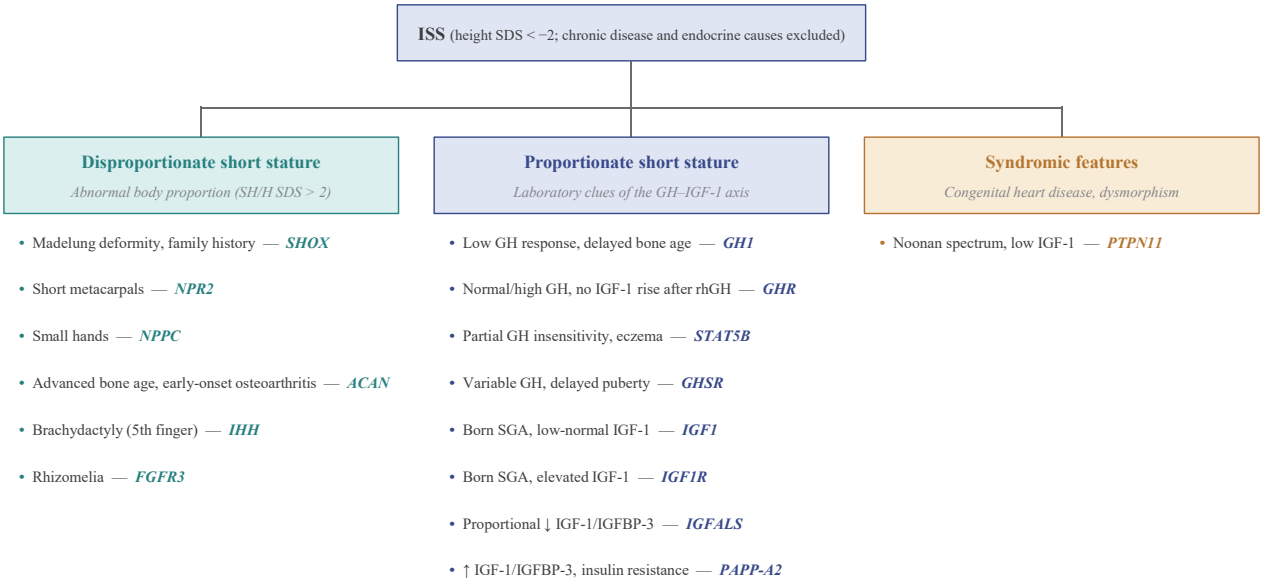


Fig. 1. Proposed diagnostic approach to the genetic causes of isolated short stature (ISS), based on associated clinical and laboratory clues. Children with ISS (height SDS <−2 after chronic systemic and endocrine causes are excluded) are grouped into three phenotypic categories that guide genetic testing. (Left) Disproportionate short stature (sitting height/height SDS >+2) suggests a growth plate disorder: (Middle) Proportionate short stature is evaluated with laboratory markers of the GH–IGF-1 axis: (Right) Syndromic features, such as congenital heart disease and dysmorphism, suggest a Noonan spectrum disorder. *ACAN*, aggrecan; *FGFR3*, fibroblast growth factor receptor 3; GH, growth hormone; *GHR*, GH receptor; *GHSR*, GH secretagogue receptor; IGF-1, insulin-like growth factor 1; *IGF1R*, IGF-1 receptor; *IGFALS*, IGF-binding protein acid-labile subunit; IGFBP-3, IGF-binding protein 3; *IHH*, Indian hedgehog; *NPPC*, natriuretic peptide precursor C; NPR2, natriuretic peptide receptor 2; *PAPPA2*, pregnancy-associated plasma protein A2; *PTPN11*, protein tyrosine phosphatase non-receptor type 11; rhGH, recombinant human GH; SDS, standard deviation score; SGA, small for gestational age; *SHOX*, short stature homeobox; SH/H, sitting height/height.

ed adult height (PAH) approximates midparental height (MPH)), familial short stature (FSS) is likely—though its genetic basis is often elusive, as it typically reflects the cumulative, incremental effects of multiple genes rather than a single variant. By contrast, a more pronounced deficit (< −2.5 SDS) despite similarly short parents raises suspicion for an underlying monogenic cause [11].

- A height deficit matching one parent's suggests autosomal dominant inheritance of the causal variant, often with multiple affected relatives across the extended family—as seen with *ACAN* or *SHOX* mutations [12,13].

- When both parents are of normal stature, yet the child has ISS, several possibilities merit consideration. Marked bone age delay, a family history of delayed puberty, and a PAH approximating MPH point toward CDGP. Otherwise, a genetic cause—potentially *de novo*, autosomal recessive, or X-linked (in boys)—should be considered [14].

Genes involved in growth plate regulation

Endochondral ossification is an intricate process occurring within the growth plate that drives bone elongation and the resulting increase in stature. It encompasses chondrocyte proliferation, hypertrophy, and senescence, along with synthesis of the cartilage matrix [15].

SHOX

SHOX, located in the pseudoautosomal region (PAR1) on the short arms of the X and Y chromosomes, regulates growth plate chondrogenesis and represents the most common monogenic cause of isolated short stature (~2.6% of nonsyndromic cases). Phenotype ranges from isolated short stature to Léri-Weill dyschondrosteosis in heterozygotes and Langer mesomelic dysplasia in homozygotes, typically due to gene or enhancer deletions. Disproportionate body segments (elevated sitting height/height ratio) are a key clinical clue warranting *SHOX* screening [16].

ACAN

ACAN encodes aggrecan, a key structural proteoglycan in growth plate and articular cartilage. Heterozygous mutations cause isolated short stature marked by advanced (rather than delayed) bone age, proportionate short stature, and growth impairment evident even prenatally. A blunted pubertal spurt and early growth cessation further compromise final height, often with body disproportion in adulthood. Additional features include brachydactyly, mild midface hypoplasia, and early-onset osteoarthritis affecting the knees by the fourth decade. Advanced bone age—contrasting with the delayed bone age typical of ISS—serves as a key diagnostic clue prompting *ACAN* testing [17].

NPR2 and NPPC

CNP and its receptor NPR-B (encoded by *NPPC* and *NPR2*) regulate endochondral ossification by stimulating chondrocyte proliferation, differentiation, and matrix synthesis, acting partly through inhibition of the *FGFR3* pathway. While biallelic *NPR2* loss-of-function causes acromesomelic dysplasia type Maroteaux, heterozygous *NPR2* mutations—first identified in ISS in 2013—account for 1.8%–13.6% of cases, producing variable short stature, abnormal body proportions, and nonspecific skeletal findings without a clear selection criterion for screening. Heterozygous *NPPC* mutations, more recently described in two families (six carriers), were associated with height SDS between -4.3 and -2.3 and a consistent small-hands phenotype [18,19].

IHH

IHH, expressed in prehypertrophic chondrocytes, is a key paracrine regulator of chondrocyte proliferation and differentiation; homozygous mutations cause acrocapitofemoral dysplasia, while heterozygous mutations cause brachydactyly type A1 (BDA1), though short stature is not a consistent BDA1 feature. In 2018, heterozygous *IHH* mutations were identified as a cause of growth impairment in 3.4% of an ISS cohort, without classical BDA1 findings. The predominant radiographic feature was shortening of the middle phalanx of the fifth finger (brachymesophalangia-V), present in 64.3% of affected hands—markedly higher than the 12.1% population prevalence. Affected individuals typically showed mild, progressively disproportionate short stature, with some born small for gestational age by birth length [20].

FGFR3

FGFR3 negatively regulates growth plate chondrogenesis; heterozygous gain-of-function mutations cause achondroplasia (the most common skeletal dysplasia, marked by rhizomelic disproportionate short stature) and the milder, more variable hypochondroplasia. While *FGFR3* alterations are rare among ISS-classified patients, some mild cases may go undiagnosed initially, and a previous recent study identified an *FG-*

FR3-activating mutation causing autosomal dominant proportionate short stature without other distinguishing features [21].

Genetic regulators of the GH-IGH1 axis regulation

Since GH and insulin-like growth factor 1 (IGF-1) were first identified as the primary regulators of longitudinal growth, early genetic studies of short stature focused on identifying defects within the GH–IGF-1–bone axis. GH, secreted by the pituitary gland, promotes bone elongation chiefly by regulating circulating and local IGF-1 synthesis. Growth failure can arise from defects at multiple points along this cascade, spanning from GH synthesis to IGF-1 action. Notably, affected individuals may lack the classical features of GH/IGF-1 deficiency or resistance, in which case they are often classified as having ISS.

GH1

GH1 defects—typically deletions or truncating mutations, though missense and promoter variants also occur—classically cause isolated GH deficiency (IGHD), presenting with proportionate short stature, low growth velocity, low IGF-1, and inadequate GH stimulation response; heterozygous/homozygous variants can also produce biologically inactive GH, phenotypically similar but with normal or elevated GH levels. However, milder *GH1* defects may lack these typical diagnostic features, as illustrated by a reported promoter mutation associated with ISS, leaving the underlying etiology undefined after standard workup [22].

GHR

GH acts via the GH receptor (*GHR*); homozygous or compound heterozygous *GHR* mutations cause complete GH insensitivity (Laron syndrome), marked by extreme short stature, normal/high GH, markedly low IGF-1 and IGFBP-3, and no IGF-1 rise after GH stimulation. Heterozygous *GHR* mutations produce a milder, more variable spectrum of GH insensitivity—only seven cases with a dominant-negative effect and recognizable partial GHI features have been reported, while broader ISS cohort studies report heterozygous *GHR* variants at 5%–15.5% frequency, though causality for many remains unproven. Low GHBP levels have been proposed as a marker of extracellular *GHR* domain defects in these patients. Given the subjective nature of clinical characterization, less deleterious heterozygous *GHR* mutations may underlie growth failure in a subset of ISS children with laboratory findings suggestive of partial GH insensitivity [23].

IGF1 and IGF1R

IGF-1, the principal growth factor in both prenatal and postnatal growth, signals through its cell-surface receptor *IGF1R*; homozygous *IGF1* deletions or loss-of-function mutations cause severe pre- and postnatal growth failure, microcephaly, and intellectual disability, with extremely low IGF-1 despite normal/high GH [24]. Heterozygous *IGF1* carriers show milder but significant short stature and reduced head circumference relative to unaffected relatives, with maternal transmission associated with greater height compromise, possibly reflecting placental dysfunction—as illustrated by a 2014 case of heterozygous *IGF1* deletion presenting as ISS with postnatal growth failure and low-normal IGF-1. Heterozygous *IGF1R* defects produce a comparable phenotype but with relatively elevated IGF-1 levels; although rare, presentation can be subtle, with short stature sometimes the sole clinical finding [25].

Other gene

PTPN11

PTPN11 encodes SHP2, a phosphatase central to several intracellular signaling pathways, including Ras/MAPK; activating heterozygous mutations cause Noonan syndrome (NS), featuring postnatal growth impairment, congenital heart disease, and facial dysmorphism, often with low IGF-1 reflecting partial GH insensitivity. As with other growth-related genes, phenotypic expression is highly variable—even within the same family—and craniofacial features evolve with age, making clinical diagnosis of NS challenging at times. Notably, pathogenic *PTPN11* variants have also been identified in children classified within ISS cohorts, underscoring the diagnostic overlap between mild NS and idiopathic short stature [26]. In addition to *PTPN11*, other genes involved in the RAS/MAPK signaling pathway, including *SOS1*, *RAF1*, and *KRAS*, contribute to growth impairment in patients with RASopathies. Furthermore, genetic defects affecting chromatin remodeling (e.g., *ARID1B* and *CHD7*), epigenetic regulation (e.g., *KMT2D* and *KDM6A*), DNA repair (e.g., *BLM*), and endosomal trafficking (e.g., *VPS33B* and *VIPAS39*) have been associated with short stature. Although these disorders often present with additional syndromic features, short stature may be an early or prominent clinical manifestation, highlighting the diverse molecular mechanisms underlying genetic growth disorders [27].

CONCLUSION

Childhood linear growth is governed by a complex array of

factors, and a substantial proportion of children classified as having ISS in fact harbor an underlying genetic cause.

Advances in molecular technology—particularly next-generation sequencing—are increasingly enabling identification of pathogenic variants even in mild phenotypes historically mistaken for ISS, despite the still-limited catalog of known causative genes. Establishing a genetic diagnosis carries practical value: it can spare children unnecessary testing and inform expected response to recombinant human GH (rhGH)—for instance, *SHOX*-deficient patients generally respond well to rhGH, whereas those with milder *GHR* defects tend to respond less robustly at standard doses. Emerging therapies may further personalize treatment, as the CNP analog vosoritide—already explored in achondroplasia—holds potential for children with *NPR2* or *NPPC* mutations. Because these conditions typically lack distinctive clinical features, candidate-gene testing alone is often inadequate, making multigene NGS panels or exome sequencing (prioritizing genes already linked to ISS) the preferred diagnostic strategy. A recent systematic review of 134 studies reported that the diagnostic yield of genetic testing in children with short stature varied according to the testing method, ranging from 4.7% for single-gene testing to 16.3% for chromosomal microarray, 21.6% for targeted gene panels, and 33.3% for exome sequencing. Notably, the diagnostic yield of exome sequencing was substantially higher in children with syndromic short stature (50.8%) than in those with isolated short stature (15.1%). Among children with isolated short stature, the most frequently identified genes included *PTPN11*, *ACAN*, *NPR2*, *COL2A1*, *SHOX*, *FGFR3*, *NF1*, *IGF1R*, *IHH*, and *COL11A1*, collectively accounting for 53.5% of genetically diagnosed cases. These findings highlight the importance of selecting appropriate genetic testing strategies based on clinical phenotypes [28].

According to the recent international guideline on genetic testing in children with short stature [29], genetic evaluation should be considered when clinical findings suggest an underlying genetic etiology. These include severe short stature (height <−3 SDS), disproportionate body proportions, skeletal abnormalities, dysmorphic features, neurodevelopmental disorders, and a family history suggestive of Mendelian inheritance. Genetic testing is also recommended in children born small for gestational age with persistent unexplained short stature when growth hormone treatment is being considered. Importantly, the absence of a family history does not exclude a genetic cause, as *de novo* variants may also contribute to growth disorders. Therefore, the selection of patients for genetic testing should be based on a comprehensive assessment of growth patterns, family history, physical findings, and associated clinical features rather than family history alone.

NOTES

Conflicts of interest

No potential conflict of interest relevant to this article was reported.

Ethical statement

Not applicable. This review article did not involve human participants or animals.

Use of artificial intelligence

During the preparation of this manuscript, the authors used ChatGPT (OpenAI) to assist with English translation, language editing, and improving the clarity and readability of the manuscript. The tool was not used to generate scientific content or data. The authors carefully reviewed and verified the final manuscript and take full responsibility for its content.

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