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A Case Report: When Family History Holds the Clue: A Prolonged Diagnostic Journey to Spinal Muscular Atrophy

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Case Report

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Abstract

Spinal muscular atrophy (SMA) is an autosomal recessive motor neuron disorder caused by biallelic abnormalities of the survival motor neuron 1 (SMN1) gene, with phenotype modified in part by SMN2 copy number. The estimated incidence is approximately 1 in 10,000 live births, although estimates vary between populations. SMA type 3 presents after independent walking has been achieved and may cause progressive proximal weakness, falls, difficulty rising from the floor, and gait abnormalities.

Delayed diagnosis remains clinically relevant despite the availability of molecular testing and disease-modifying therapies. Published reports have described children initially managed for hypotonia, gait abnormalities, orthopedic problems, or other neuromuscular diagnoses before SMA was recognized.

This case is publishable because it combines several educationally important features. The diagnostic delay following an initial orthopedic label of bilateral club feet, despite having a similarly affected older sibling, indicate the lack of knowledge and clinical correlation. It therefore illustrates a clinically relevant diagnostic pathway, particularly for resource-limited settings, rather than simply documenting a rare genetic result.

This article report a 12-year-old boy who had progressive difficulty walking since the age of 18 months, after achieving independent walking. He initially fell after walking short distances and developed difficulty rising from sitting, followed by difficulty climbing stairs, running, and jumping as his age progresses. At about 3 years he was reportedly diagnosed with bilateral club feet and treated with corrective shoes without improvement. Follow-up was later interrupted due to lack of improvement. The mother mentioned that the patient use his hands to crawl and stand. His 27-year-old brother had a similar early-onset progressive motor disorder and became wheelchair dependent. Examination showed exaggerated lumbar lordosis, generalized hypotonia, absent deep tendon reflexes, proximal lower-limb weakness with preserved distal strength, waddling gait, positive Gowers sign, and tongue fasciculation without atrophy. Sensation was intact. EMG showed mixed neurogenic and myogenic changes and NCS severe bilateral tibial axonal neuropathy. Molecular testing demonstrated homozygous deletion of SMN1 exon 7 and heterozygous deletion of exon 8, supporting 5q SMA, clinically consistent with SMA type 3.

This case report shows that physicians should consider SMA in patients presented with progressive gait difficulty after acquisition of independent walking, especially when accompanied by proximal weakness, hypotonia, areflexia, Gowers sign and preserved sensation. Early recognition and molecular confirmation are increasingly important because disease-modifying therapies and multidisciplinary care have changed SMA management.

Introduction

SMA is a genetically and clinically heterogeneous disorder characterized by degeneration of lower motor neurons and progressive weakness. Identification of SMN1 established the molecular basis of the

common 5q form (1). The disease is autosomal recessive, while SMN2 acts as an important phenotype modifier (2, 4).

Epidemiologic studies estimate an incidence around 1 in 10,000 live births and prevalence around 1–2 per 100,000 persons, with variation among populations (3). SMA type 3 is an ambulant phenotype in which symptoms develop after independent walking. Typical manifestations include falls, difficulty running or climbing stairs, difficulty rising from the floor, proximal lower-limb weakness, and waddling gait (4, 5).

The therapeutic landscape has changed substantially. International recommendations emphasize molecular confirmation, multidisciplinary care, and consideration of disease-modifying therapy (11, 12). Newer best-practice recommendations emphasize efficient diagnosis and newborn screening because early treatment can improve outcomes (5).

Diagnostic delay remains important. In a multicenter Italian cohort of 480 genetically confirmed patients with SMA types 1–3, the interval between recognition of clinical signs and genetic diagnosis remained clinically relevant (7). Iwayama et al. reported a 12-year-old girl whose SMA type 3b diagnosis was delayed seven years because initial SMN1 copy-number testing did not identify an intragenic mutation; subsequent sequencing established the diagnosis (8).

Previously reported case reports demonstrate the clinical variability of SMA. Kibtiar et al. described a 5-year-old boy with SMA type 3, proximal weakness, generalized hypotonia, absent reflexes, normal muscle enzymes, and a similarly affected sibling; the diagnosis was ultimately supported by muscle biopsy (6). Sharma et al. reported an 11-year-old boy with difficulty walking and rising from sitting, proximal weakness, hypotonia, absent reflexes, and severe hyperlordosis, highlighting hyperlordosis as a clinical clue (9). Bregvadze et al. reported a case series focused on diagnostic difficulties and molecular challenges in SMA (10). These reports closely relate to the present case because our patient also had progressive proximal weakness, hyperlordosis, areflexia, gait impairment, family recurrence, and a prolonged diagnostic pathway.

The present case adds a specific diagnostic lesson: symptoms began at 18 months after independent walking, yet the gait disorder was initially attributed to bilateral club feet. The combination of a similarly affected sibling, lower-motor-neuron signs, mixed electrodiagnostic findings, and SMN1 molecular confirmation demonstrates how an inherited motor neuron disorder can remain unrecognized when an orthopedic explanation is assigned early.

The case is publishable because its value lies in the diagnostic and educational message in addition to the rarity of SMA itself. It documents prolonged diagnostic delay, orthopedic misattribution, familial recurrence, mixed electrodiagnostic findings, and molecular confirmation in a setting where access to specialist and genetic services may be limited. These features provide a practical lesson for pediatricians, orthopedists, physiotherapists, and neurologists.

Case presentation

A 12-year-old boy from Ethiopia presented for pediatric neurological evaluation because of progressive difficulty walking. Symptoms began at the age of 1 year and 6 months (18 months). His mother noticed falling after short-distance walking, despite achiving independent walking prior to the onset of symptomes. He subsequently developed difficulty standing from sitting, requiring use of his hands, and later was unable to climb stairs, jump, and run.At approximately 3 years, he was evaluated at local hospital where he was reportedly diagnosed with bilateral club feet. Corrective shoes were used without improvement. Follow-up was discontinued because of no improvement. There was no reported history of abnormal movements, loss of consciousness, dysphagia, numbness, tingling, pain, hearing or visual impairment, trauma, urinary or bowel incontinence, fever, cough, shortness of breath, or other chronic illness.

He has similar family history. His 27-year-old brother developed difficulty standing from sitting at approximately 1 year of age and never walked without support. He later became wheelchair dependent but retained writing and electronics-maintenance abilities. No sensory or bowel/bladder symptoms were reported.

The patient had normal developmental milestone before the onset of syptomes. He smiled socially at 3 months, sat unsupported at 6 months, crawled at 8 months, and said “mama” and “dada” at 11 months. He began standing and walking at approximately 1 year and developed progressive difficulty at 18 months. He is currently a grade 4 student with good academic performance. He was born at term after an uncomplicated pregnancy and spontaneous vaginal delivery, with birth weight 3 kg. He cried immediately and breastfed within the first hour; no NICU admission was reported. In addition, he is fully vaccinated according to the national EPI schedule.

On physical presentation, he is well appearing. Vital signs are in the normal range (BP 105/60 mmHg, pulse 98/min, RR 24/min, SpO2 95% on room air and temperature 36.5°C). Antropometry is in the normal range (weight 34 kg, height 135 cm, BMI 18.7 kg/m²). Head and neck, respiratory, cardiovascular, abdominal and genitourinary examinations were unremarkable. Musculoskeletal examination showed exaggerated lumbar lordosis without scoliosis or kyphosis.

On neurologcal examination, mental status, attention, memory, speech and language were intact. No sensory and coordination abnormality noted. On motor examination, There is visible muscle wasting on the lower extrimity. Power was 3/5 in lower limb, particularly proximal part. Tone is reduced in all four extrimity. Deep tendon reflexes were absent on knee and ankle bilaterally. He had tongue fasciculation. Gait was waddling and Gowers sign was positive. Motor exam on upper limb is less severe compaired to lower limb.

On investigation, complete blood count, organ function test, electrolyte was normal. Creatinine kinase was also normal (Table 1).

Table 1
Laboratory investigations

CBC		**RFT**	**LFT (u/l)**	**Electrolytes**	**Creatinine kinase**
WBC(×10³/μL)	5.1	Cr-0.2 mg/dl	AST-26	**Na – 139 mmol/l**	**144 U/L (Normal)**
Neut(%)	65.5	Urea-22 mg/dl	ALT-20	K – 4.5 **mmol/l**	
Lymph(%)	25		ALP-209	Cl – 104.9 **mmol/l**	
HGB(g/dl)	13.5			Ca – 10 mg/dl	
RBC(×10^6/μL)	5.06			Mg – 2 mg/dl	
PLT(×10³/μL)	222,000				
MCHC(g/dl)	33.5				
HCT(%)	41.4				
RDW(%)	14.2				
MCV(fl)	82.0				
MCH(pg)	27.4				

Electromyography demonstrated mixed neurogenic and myogenic changes, including mild myogenic changes with short-duration, low-amplitude MUAPs and early recruitment in bilateral vastus medialis, with chronic neurogenic changes elsewhere. Few fibrillations and positive sharp waves were reported. Nerve conduction test showed severe bilateral tibial axonal neuropathy. The differential included SMA type 3 and chronic myopathy such as Becker muscular dystrophy.

Molecular genetic testing demonstrated homozygous deletion of SMN1 exon 7 and heterozygous deletion of exon 8. SMN2 exons 7 and 8 did not show a significant increase in copy number. DMD copy-number testing did not show abnormalities.

The combination of onset at 18 months after independent walking, progressive proximal weakness, difficulty rising, waddling gait, Gowers sign, hypotonia, generalized areflexia, tongue fasciculation, preserved sensation, and normal-range CK favored a lower motor neuron disorder. The SMN1 abnormality provided molecular support for SMA, clinically consistent with SMA type 3.

Management of spinal muscular atrophy (SMA) requires a proactive, multidisciplinary approach aimed at preserving motor function, preventing respiratory and nutritional complications, maintaining skeletal and functional status, and improving quality of life. Current standards of care recommend regular assessment and coordinated follow-up involving pediatric neurology, pulmonology, nutrition and gastroenterology, physiotherapy and rehabilitation, orthopedics, and other relevant specialists. Respiratory care is particularly important because respiratory muscle weakness, ineffective cough, recurrent respiratory infections, and sleep-disordered breathing are major causes of morbidity and

mortality in severe SMA. Management includes regular respiratory assessment, immunization and prevention of respiratory infections, airway-clearance techniques, assisted coughing, and non-invasive ventilatory support when indicated. Nutritional management should include monitoring of weight and growth, assessment of swallowing and aspiration risk, treatment of gastroesophageal reflux and constipation, and consideration of enteral feeding when oral intake is inadequate or unsafe. Physiotherapy and rehabilitation are directed toward maintaining joint range of motion, preventing contractures, optimizing positioning and mobility, and maximizing functional independence. Orthopedic surveillance is required for scoliosis, hip displacement, contractures, and other skeletal complications. Genetic counseling and reproductive counseling should also be offered to affected families.(15, 16)

Disease-modifying therapies have substantially changed the natural history of SMA. Currently available therapies include nusinersen, an antisense oligonucleotide that promotes production of full-length SMN protein from the SMN2 gene; risdiplam, an orally administered SMN2 splicing modifier; and onasemnogene abeparvovec, an adeno-associated virus-based gene-replacement therapy designed to deliver a functional copy of SMN1. Early treatment, particularly before substantial motor-neuron loss, is associated with better outcomes. (17) However, these therapies were not available to our patient because of limited access and resource constraints. Consequently, management in this case focused on optimal supportive multidisciplinary care and the use of salbutamol as an off-label adjunctive treatment.

Because disease-modifying therapies were unavailable, the patient was started on **oral salbutamol at a dose of 0.1 mg/kg three times daily**. This treatment was selected as an adjunctive, rather than disease-modifying, therapy. Salbutamol (albuterol) is a selective β2-adrenergic receptor agonist that has been investigated in SMA because β2-adrenergic signaling may influence both neuromuscular function and SMN protein biology. Activation of β2-adrenergic receptors increases intracellular cyclic adenosine monophosphate (cAMP), which activates protein kinase A (PKA) and may enhance transcription of the **SMN2** gene. Experimental and clinical evidence suggests that β2-adrenergic stimulation may increase the production of full-length SMN2 transcripts and functional SMN protein. In addition, β2-adrenergic stimulation may have effects on skeletal muscle through increased protein synthesis and reduced muscle protein breakdown, while potentially improving neuromuscular transmission and muscle contractility.(18, 19)

The proposed molecular mechanism is therefore complementary to, but distinct from, established SMN2-directed disease-modifying therapies. Recent evidence suggests that salbutamol may enhance full-length SMN2 transcript expression through cAMP-dependent signaling and may increase SMN protein levels through the β2-adrenergic receptor–PKA pathway, including possible reduction of ubiquitin-mediated SMN degradation.[4] Nevertheless, the available evidence remains insufficient to consider salbutamol a replacement for nusinersen, risdiplam, or onasemnogene abeparvovec. Its use in this patient should therefore be interpreted as an off-label therapeutic strategy in a resource-limited setting where approved disease-modifying therapies were inaccessible.

The most recent systematic review and meta-analysis specifically evaluating salbutamol in 5q SMA was published by Xing et al. in 2025.(18) The authors included eight clinical studies involving 154 patients and evaluated motor function, respiratory function, SMN2 transcript levels, musculoskeletal outcomes, patient-reported outcomes, and adverse events. The meta-analysis demonstrated a statistically significant improvement in Revised Upper Limb Module (RULM) scores among patients younger than 6 years in two studies, with a mean difference of 3.89 points (95% CI 0.35–7.43; P = 0.03). Salbutamol was also associated with a significant increase in peripheral full-length SMN2 transcripts, with a mean difference of 25.13 (95% CI 16.12–34.13; P < 0.00001) at 6 months, with the effect maintained at 12 months. Some studies additionally reported subjective improvement, respiratory benefits, and weight gain in younger patients. Overall, salbutamol appeared to be relatively well tolerated, although the authors emphasized that the available evidence is limited by the small number of participants, heterogeneity between studies, and the lack of adequately powered double-blind randomized controlled trials. They concluded that salbutamol may represent a potentially useful and relatively accessible adjunctive treatment, particularly for younger patients and in settings where disease-modifying therapies are unavailable, but that definitive randomized controlled trials are required before its efficacy can be established.(18)

In our patient, salbutamol was therefore used as an accessible off-label adjunctive therapy in the context of unavailable disease-modifying treatment, while maintaining multidisciplinary supportive care and follow-up. The patient was referred for continued pediatric pulmonology, cardiology, and orthopedic assessment, and genetic counseling was planned, consistent with the multidisciplinary approach recommended for SMA. It has been two weeks since started on the management, no significant improvement is noted yet.

Discussion

This case illustrates a clinically important diagnostic pathway in pediatric neuromuscular disease. Symptoms began at 18 months after independent walking and progressively affected proximal lower-limb function. The initial attribution to bilateral club feet delayed recognition of the motor neuron disorder. Gait disturbance, falls, and lower-limb deformity can lead families to seek orthopedic care before neurological assessment.

The principal clinical clues were proximal weakness, hypotonia, areflexia, Gowers sign, waddling gait, preserved sensation, and tongue fasciculation. This constellation is compatible with SMA (4, 11). The normal-range CK supported the differential against muscular dystrophy, although CK alone cannot diagnose or exclude SMA. Another differential can be CIDP. Even though lower extrimity hypotonia and areflexia might mimic CIDP, the patient didn’t have any sensory deficit. In addition, NCS shows axonal problem instead of demyelination which is not expected in CIDP. The family history is particularly informative. An older brother had a strikingly similar early-onset progressive motor phenotype and became wheelchair dependent. Because classic 5q SMA is autosomal recessive, recurrence among siblings is a strong clue to inherited disease and supports genetic counseling (1, 4, 11).

The electrodiagnostic study showed mixed neurogenic and myogenic changes and severe bilateral tibial axonal neuropathy. Such mixed findings can complicate initial classification, but electrodiagnostic results should be interpreted alongside the clinical phenotype and molecular data. In this case, the SMN1 finding and clinical syndrome were decisive. In addition, long standing hypotonia might result secondary myogenic changes like in our patient.

Kibtiar et al. reported a 5-year-old boy with SMA type 3 who had proximal weakness, generalized hypotonia, absent reflexes, normal muscle enzymes, and a similarly affected sibling (6). Their diagnosis was supported by muscle biopsy after initial consideration of myopathy. Our patient shares the clinical phenotype and family recurrence but was diagnosed later and had molecular confirmation of SMN1 exon 7 deletion.

Sharma et al. described an 11-year-old boy with difficulty walking and rising from sitting, proximal weakness, hypotonia, absent reflexes, and severe hyperlordosis (9). Our patient also had exaggerated lumbar lordosis, although not described as severe. Both cases demonstrate that abnormal lumbar posture may be a clue to chronic proximal weakness and should not automatically be treated as an isolated orthopedic condition.

Iwayama et al. reported a 12-year-old girl with a seven-year diagnostic delay; initial SMN1 copy-number analysis was nondiagnostic, while later sequencing identified an intragenic SMN1 variant and established SMA type 3b (8). That report highlights the limitation of copy-number testing alone. In contrast, our patient's exon 7 deletion was detectable by copy-number analysis, but the clinical diagnosis was nevertheless delayed because the presentation was initially interpreted as orthopedic.

Bregvadze et al. described a series centered on diagnostic difficulties in SMA, emphasizing phenotypic variability and molecular diagnostic challenges (10). Their findings reinforce the need to maintain SMA in the differential diagnosis when a child has unexplained progressive motor weakness, even when the presentation is not stereotypical.

Pera et al. studied 480 genetically confirmed patients with SMA types 1–3 and demonstrated that the diagnostic journey can remain prolonged, supporting the clinical importance of early recognition and genetic confirmation (7). Our patient provides a concrete example of this problem, with symptoms beginning at 18 months and diagnosis only after years of interrupted follow-up.

Rizvi et al. reported a fatal delayed-diagnosis case of SMA type 1 and emphasized early detection, genetic testing, and multidisciplinary care (13). Although type 1 is clinically more severe than the present case, the report supports the general principle that delayed recognition can have important consequences across the SMA spectrum.

The therapeutic landscape has changed substantially. Current best-practice recommendations emphasize rapid molecular diagnosis and newborn screening, while disease-modifying therapies have changed the natural history of SMA (5, 11, 12). Consequently, recognizing an ambulant child with

progressive SMA is increasingly important because diagnosis may open access to disease-modifying treatment, rehabilitation, respiratory and nutritional surveillance, orthopedic management, and genetic counseling.

The case should be presented as educationally valuable rather than as proof that SMA type 3 is exceptionally rare or that its individual features are unprecedented. Previous reports have already described type 3 SMA, hyperlordosis, diagnostic delay, and molecular diagnostic difficulty (6, 8–10). The contribution of the present case is the combination of 18-month onset after independent walking, initial orthopedic labeling as bilateral club feet, familial recurrence, mixed electrodiagnostic findings, and molecular confirmation in a resource-limited setting. This combination provides a practical lesson for clinicians evaluating children with progressive gait abnormalities.

Strength of this case report include a detailed developmental and longitudinal history, an affected sibling, neurological examination, electrodiagnostic testing, and molecular confirmation. Limitations include the absence in the available record of standardized SMA motor-function scores, detailed respiratory function testing, formal nutritional/swallowing assessment, longitudinal treatment response, and a definitive SMN2 copy-number value. Inaddition, disease-modifying therapies for SMA are currently not available in Ethiopia. Therefore, due to the limited availability of these therapies, the patient was managed with the off-label use of salbutamol, based on evidence from previous studies suggesting potential benefits in patients with SMA. (18)

Conclusions

SMA should be considered in children with progressive proximal weakness, falls, difficulty rising from sitting, waddling gait, Gowers sign, hypotonia, and generalized areflexia, particularly when sensation is preserved. Orthopedic findings such as abnormal gait or lumbar lordosis should not prematurely exclude a neuromuscular diagnosis.

In this patient, symptoms began at 18 months after independent walking, the initial diagnosis was bilateral club feet, and a similarly affected sibling suggested an inherited disorder. The clinical phenotype, electrodiagnostic abnormalities, and SMN1 exon 7 deletion supported 5q SMA, clinically consistent with type 3. Early recognition and molecular confirmation are important for timely counseling, multidisciplinary care, and consideration of disease-modifying treatment.

Abbreviations

BMD
Becker muscular dystrophy
CIDP
chronic inflammatory demyelinating polyneuropathy
CK
creatine kinase

DMD
Duchenne muscular dystrophy
EMG
electromyography
MUAP
motor unit action potential
NCS
nerve conduction study
SMA
spinal muscular atrophy
SMN
survival motor neuron

Declarations

Ethics approval and consent to participate

According to the institutional review board policy of our institution, ethical approval was not required for a single case report. Written informed consent was obtained from the patient's parents for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Consent for publication

According to the review board policy of our institution, ethics approval was not necessary. Written informed consent was obtained from the patient's parents for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Availability of data and materials

All data generated or analyzed during this study are included in this manuscript. Additional information is available from the corresponding author upon reasonable request.

Competing interests and Funding

No competing interests were declared. No funding was received for this work.

Authors' contributions

TZA (Corresponding author) collected the clinical data, reviewed the literature and performed original draft preparation, TA is the clinician who diagnose and manage the patient. BZ and EFM reviewed and edited the final draft. All authors read and approved the final manuscript.

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During the preparation of this manuscript, the authors used ChatGPT (OpenAI, GPT-5.5-mini, OpenAI) to assist with language refinement, improving readability, and editing of selected sections of the manuscript. The AI tool was not used for patient data generation, data analysis, clinical decision-making, or interpretation of clinical findings. All AI-assisted content was reviewed and revised by the authors, who take full responsibility for the accuracy, originality, and integrity of the manuscript.

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Figures

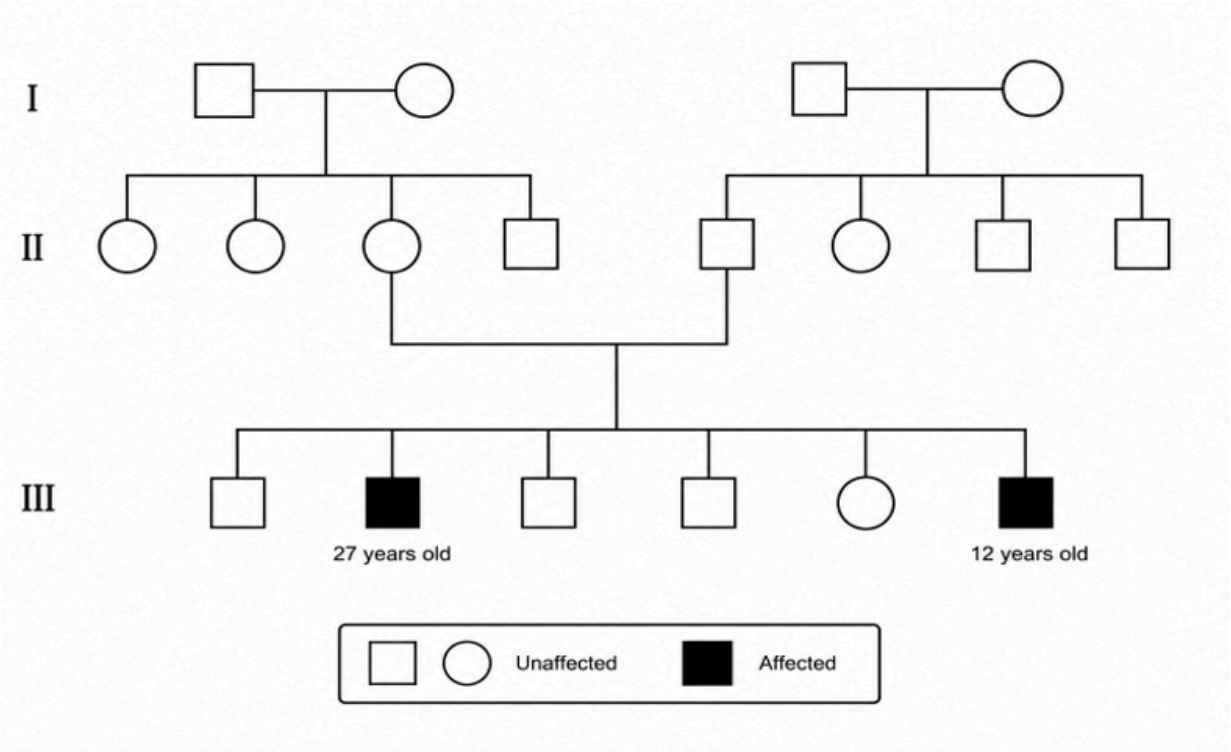


Figure 1

Pedigree of the family