ISSN: 2755-9866

Journal of Health Statistics Reports

SCIENTIFIC Research and Community

Case Report

Open Access

The Importance of Early Diagnosis of Ehlers-Danlos Syndrome – A Case Study and Public Health Perspective

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ABSTRACT

Ehlers-Danlos Syndrome (EDS) is a rare, inherited connective tissue disorder, frequently diagnosed with significant delay due to fragmented care and lack of coordination among specialists. We present the case of a 35-year-old woman whose EDS diagnosis was established 17 years after the onset of symptoms. Despite numerous specialist consultations, the focus on isolated symptoms without a holistic approach led to prolonged diagnostic neglect. This case highlights the crucial role of the family physician as a care coordinator, integrating clinical data from various fields and adopting a holistic view of the patient. Early detection of rare diseases such as EDS is vital for public health—it shortens the time to diagnosis, improves patient quality of life, and optimizes healthcare resource utilization. Our analysis underscores the urgent need for systemic solutions: better care coordination, development of integrated medical data tools, and enhanced education of physicians in rare diseases, especially at the primary care level.

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Received: September 26, 2025; **Accepted:** September 30, 2025; **Published:** October 09, 2025

Keywords: Ehlers-Danlos Syndrome, Connective Tissue Disease, Skin Hyperelasticity, Joint Hyperextension, Hemorrhagic Spots, Petechiae, Vascular Fragility, Hemorrhagic Diathesis, Rare Diseases, Joint Hypermobility

Introduction

Ehlers-Danlos Syndrome (EDS) is a heterogeneous group of inherited connective tissue disorders, directly resulting from mutations in genes encoding collagen proteins. These disorders lead to significant structural and functional abnormalities of connective tissue, such as joint hypermobility, increased skin elasticity, vascular wall fragility, and heightened tissue susceptibility to injury [1].

The epidemiology of Ehlers-Danlos Syndrome (EDS) indicates a prevalence of approximately 1:5,000 live births, with the hypermobility type occurring at a frequency of 1:10,000 [1]. Advances in molecular genetics have enabled the identification of numerous mutations responsible for the clinical manifestations of the disease. Combined with the complex inheritance mechanisms, this makes the accurate diagnosis of EDS a significant diagnostic and therapeutic challenge.

Early diagnosis of EDS is crucial for preventing complications and improving patients' quality of life. As scientific reports indicate, it also optimizes healthcare costs [2]. Unfortunately, due to fragmented patient care, lack of collaboration between specialists, and low awareness of rare diseases, the time from the onset of initial symptoms to the correct diagnosis often spans several years [3].

It is estimated that in Poland, between 7,600 and 15,000 individuals may be affected by this disease. In practice, fewer than one thousand cases have been diagnosed, suggesting that many individuals remain undiagnosed. The prevalence of specific EDS types is as follows: Hypermobile type (hEDS): 1 in 5,000 to 1 in 20,000 individuals; Classic type: approximately 1 in 20,000; Vascular type: approximately 1 in 100,000.

The authors present the case of a 35-year-old female patient in whom the diagnosis of EDS was established 17 years after the onset of initial symptoms. The study analyzes the diagnostic process, complications, the impact of the disease on the patient's quality of life, and highlights the social and economic costs, as well as discusses the importance of early diagnosis from a public health perspective.

Case Presentation

General Data and Medical History

A 35-year-old female patient presented for her first visit to a Primary Care Physician (PCP), having transferred from another clinic. During the initial visit, she reported complaints that, despite the efforts of other specialists, had not been alleviated and had even intensified recently. These symptoms prevented the patient from functioning normally in both her family and professional life.

Documentation indicates that the patient underwent numerous specialist consultations, including orthopedics, endocrinology, gastroenterology, cardiology, rheumatology, and neurology. Despite extensive imaging and laboratory investigations, the cause of her complex symptoms remained unidentified.

Orthopedic Diagnostics and Treatment

2005: Orthopedic Outpatient Clinic

Patient reported knee joint pain, mainly during walking.
Diagnostics: X-ray of the knee joints.
Findings: Oval radiolucencies (fibrous dysplasia), at the femoral metaphysis an oval radiolucency with a sclerotic rim measuring approximately 20x11 mm (Defectus fibrosis cortilaris susp). Otherwise, bone structures without pathological changes.

2005: Orthopedic Department

Diagnosis: Fibrous dysplasia of the left femur, lateralization of the left patella.
Treatment: Arthroscopy of the left knee joint; lateral retinacular release and resection of the plica anterior to the lateral cruciate ligament.
Discharge Summary: Mild anemia (hemoglobin 11.2 g/dL).
Recommendations: Further treatment at the Orthopedic Outpatient Clinic.

2006: Trauma and Orthopedic Surgery Outpatient Clinic

Patient reported persistent pain symptoms unresponsive to conservative treatment (NSAIDs).
Diagnostics: MRI of the left knee joint in SE (T1 - sagittal and coronal) and TSE (T2 - sagittal, coronal, and axial) sequences without contrast enhancement.
Conclusions: Postoperative state of the left femur after fibrous dysplasia, postoperative state of the patellar retinaculum after lateral release, chondromalacia of the left patella.

2006: Trauma and Orthopedic Surgery Outpatient Clinic

Patient reported knee joint pain, especially in the left knee.
Diagnostics: Ultrasound of the left knee joint.
Findings: Chondromalacia of the patella, grade I-II cartilage defects, osteophytic deformities of the tibiofemoral joint, and abnormalities of the distal femoral metaphysis.

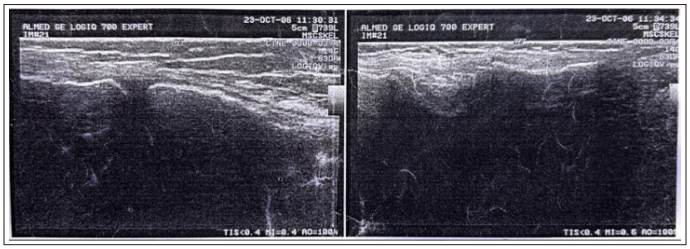

Figure 1: Ultrasound Examination of the Left Knee Joint Demonstrated Chondromalacia of the Patella, Grade I–II Cartilage Defects, Osteophytic Deformities of the Tibiofemoral Joint and Abnormalities of the Distal Femoral Metaphysis

2005: Department of Trauma and Orthopedic Surgery

The patient was scheduled for arthroscopy of the left knee joint due to exacerbation of symptoms following a twisting injury.
Procedures Performed: Second arthroscopy of the left knee joint; shaving of patellar chondromalacia and resection of the infrapatellar plica, joint lavage and drainage. After drain removal and resolution of pain, the patient was discharged home.
Diagnosis: Status post left knee joint injury, focus of patellar chondromalacia.
Recommendations: Non-weight bearing on the left lower limb. Medications prescribed: Fraxiparine (nadroparin) 0.3 ml once daily, Ketonal (ketoprofen) 50 mg as needed for pain, Zinnat (cefuroxime) 500 mg twice daily, dietary supplements.

2007: Department of Radiology and Imaging Diagnostics

The patient reported chest discomfort.
Procedure Performed: MRI of the costal arches.
Findings: Discrete asymmetry (localized protrusion) of the left costal arch.

2008: Department of Trauma and Orthopedic Surgery

The patient was admitted for a second arthroscopy of the left knee joint.
Procedures Performed: Third arthroscopy of the left knee joint; shaving of left patellar chondromalacia, damage to the upper-lateral pole, excision of the plica anterior to the anterior cruciate ligament.
Diagnosis: Chondromalacia of the left patella.

2008: Department of Trauma and Orthopedic Surgery

The patient was admitted for excision of a contractile scar (keloid) in the region of the right knee joint.
Procedure performed: Excision of a contractile keloid in the region of the left knee joint; radical excision of the lesion.

2009: Department of Trauma and Orthopedic Surgery

The patient was electively admitted for knee arthroscopy due to knee pain and progressive patellar chondromalacia.
Procedures Performed: First Arthroscopy of the right Knee Joint.
Findings: Foci of Patellar Chondromalacia.
Treatment: Excision of the Plica Anterior to the Anterior Cruciate Ligament, Synovial Biopsy.

2010: Outpatient Trauma and Orthopedic Surgery Clinic

The patient reported severe pain in the left shoulder joint.
Procedure Performed: MRI of the left shoulder joint.
Findings: Edema of the superior periarticular structures, heterogeneous labrum.

2013: Department of General Surgery

The patient was admitted for excision of a melanocytic skin lesion.
Histopathology: Benign lesion, Located on the Right Side of the Neck, Keloid formation after Healing.

2014: Department of Trauma and Orthopedic Surgery

The patient was admitted for a second arthroscopy of the right knee joint, reporting persistent knee pain, a sense of instability, and discomfort while walking.
Procedures Performed: Second Arthroscopy of the Right Knee Joint.
Findings: Large focus of grade III Chondromalacia in the Upper-Medial part of the Patella.
Treatment: Shaving of the Lesion.

2014: Department of Trauma and Orthopedic Surgery

The patient was referred for Arthroscopy of the Right Knee Joint due to Increasing Pain and Discomfort while Walking.
Procedures Performed: Second arthroscopy of the right knee joint – grade III chondromalacia of the right patella – shaving.
Clinical Diagnosis: Grade III Chondromalacia of the Right Patella.

2014: Department of Trauma and Orthopedic Surgery

The patient was referred for arthroscopy due to persistent joint pain, worsening when climbing stairs.
Procedures Performed: Fourth arthroscopy of the right knee joint; removal of small synovial fragments from the suprapatellar recess.
Diagnosis: Grade III chondromalacia of the right patella (M22.4).

2019: Department of Trauma and Orthopedic Surgery
The patient was admitted for surgical management of a left knee sprain with ligament injury.
Diagnosis: Sprain of the left knee joint, anterior cruciate ligament injury, acute patellar cartilage injury.
Procedures Performed: Fourth arthroscopy of the left knee joint – cruciate ligament reconstruction.

Rheumatological Diagnostics and Treatment
2007: Department of Pediatric Diseases
The patient reported joint pain.
Excluded: Lyme Disease, Chlamydia, Syphilis, Hyperparathyroidism.
Findings: Negative Antinuclear Antibodies, Normal Complement Levels, MRI of the Left Knee Showed Postoperative Changes and Metallic Bodies Hindering Precise Assessment, Positive ASO Titer, Diagnosis of Past Streptococcal Infection.
Additional tests: Echocardiography, Abdominal Ultrasound, Scintigraphy – all within Normal Limits.
Consultations: ENT, Ophthalmology, and Cardiology – No Abnormalities Detected.

2008: Department of Rheumatology
The patient was admitted for diagnostic evaluation due to polyarticular pain with mild limitation of wrist mobility and limited extension in the right knee joint.
History of multiple right knee arthroscopies due to diagnosed fibrous dysplasia of the femur.
Diagnosis: Polyarthritis, Dental Caries, Status Post Left Knee Surgery.
Tests Performed: Anti-dsDNA Antibodies Negative, ANA1 Negative, WR Negative, Inflammatory Markers Negative, X-ray Showed mild Osteoporosis.
Planned: Initiation of Disease-Modifying Antirheumatic Drugs (for arthritis), Postponed Pending RF and Sterile Urine Culture Results.
Discharge Summary: Previous attempt to start Arechina (chloroquine) in outpatient setting, discontinued due to ophthalmologic reasons – no further details.
Recommendations: Ibuprofen 200 mg three times daily, PPI 20 mg once daily, amoxicillin with clavulanic acid, tetrazepam, dental sanitation, urine culture after treatment, continued rheumatology follow-up.

2014: Department of Rheumatology
The patient was admitted due to generalized pain in the hip and wrist joints, with mild morning stiffness.
Findings: Tenderness of the hip, knee, and right shoulder joints, hypertrophy in the knee, elbow, and wrist joints, keloids after surgical procedures.
Tests Performed: Abdominal ultrasound, chest X-ray, laboratory tests (basic, RF, ANA) – all within normal limits.
Diagnosis: Joint hypermobility syndrome, patellar chondromalacia, status post arthroscopy of both knee joints.
Treatment: Lornoxicam, Rehabilitation.

2014: Department of Rheumatology
The patient was admitted due to generalized pain in the hip and wrist joints, with mild morning stiffness.
Findings: Tenderness of the hip, knee, and right shoulder joints, hypertrophy in the knee, elbow, and wrist joints, keloids after surgical procedures.
Tests Performed: Abdominal ultrasound, chest X-ray, laboratory tests (basic, RF, ANA) – all within normal limits.
Diagnosis: Joint hypermobility syndrome, patellar chondromalacia, status post arthroscopy of both knee joints.
Treatment: Lornoxicam, Rehabilitation.

Neurological Diagnostics and Treatment
2014: Neurology Outpatient Clinic
The patient reported sharp, piercing headaches, worsening in the morning, localized in the occipital region, accompanied by visual disturbances (loss of visual acuity in one eye).
Procedure Performed: MRI of the head.
Result: Normal.

2015: Department of Neurology
The patient was admitted due to recurrent and chronic spinal joint pain.
Tests Performed: Laboratory tests (within normal limits), spinal MRI (trace L5/S1 discopathy), otherwise unremarkable.
Clinical Diagnosis: Lumbar spondylosis with radiculopathy.
Recommendations: Baclofen 10 mg, Ketonal 50 mg as needed for pain.

2016: Neurology Outpatient Clinic
The patient was under regular care for back and head pain.
Findings/Procedures: X-ray of the cervical and thoracic spine: S-shaped scoliosis, Th7/8 and Th8/9 discopathy.
Head CT: No significant brain pathology.
Cervical Spine MRI: Normal.

2023: Neurology Outpatient Clinic
The patient reported lumbar spine pain.
Procedure Performed: MRI.
Findings: Abnormal lumbar-sacral spine alignment with acute sacral positioning, two-level discopathy (central protrusions with bulging) at L4-L5 and L5-S1, early lumbar spondylosis.

Obstetric Complications
2022: Outpatient Obstetrics and Gynecology Clinic
The patient reported spotting and was admitted for a follow-up ultrasound. One week prior, a gestational sac without embryonic echo was visualized.
Ultrasound Findings: Embryonic echo corresponding to 5 weeks gestation, FHR (+).
Recommendations: Complete blood count, BOM, TSH, glucose, and follow-up in 3 weeks.

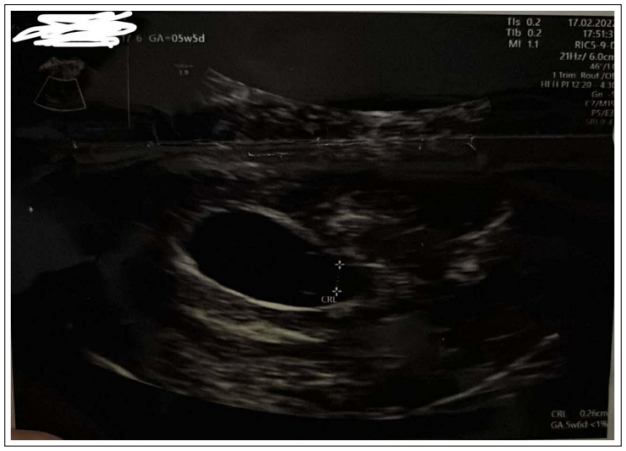

Figure 2: Anteverted Uterus. A Single Gestational Sac is Present within the Uterine Cavity, Containing a Yolk Sac and an Embryonic Echo

2022: Emergency Department, Clinical Center of Gynecology, Obstetrics and Neonatology
Patient presented with vaginal spotting (10 weeks of gestation).
Gynecological Examination: Mucous discharge tinged with blood in the vagina. Transvaginal ultrasound (TVUS) revealed a single gestational sac with visible embryonic echo, CRL 2.48 cm (corresponding to 9 weeks 1 day), FHR 171 bpm, chorion properly implanted without signs of detachment.

Diagnosis: Threatened miscarriage.

2022: Outpatient Obstetrics and Gynecology Clinic
Patient presented with recurrent episodes of spotting.
Investigations: Ultrasound showed a fetus in breech presentation, size corresponding to 20 weeks of gestation. Amniotic fluid volume was normal, fetal organs assessed by ultrasound were normal, and cervical length was measured by TVUS.

2022: Department of Obstetrics and Gynecology
Patient admitted due to threatened preterm labor and cervical insufficiency.
Examination: No premature uterine contractions observed; fetus showed no signs of distress.
Diagnosis: Cervical insufficiency at 24 weeks 0 days of gestation, Breech Presentation, Cervical Incompetence – for further Observation.

2022: Outpatient Obstetrics and Gynecology Clinic
Patient presented with spotting within the last 24 hours.
Examination: Transvaginal ultrasound showed cervical length of 21 mm. The patient was referred to the Department of Obstetrics and Gynecology.

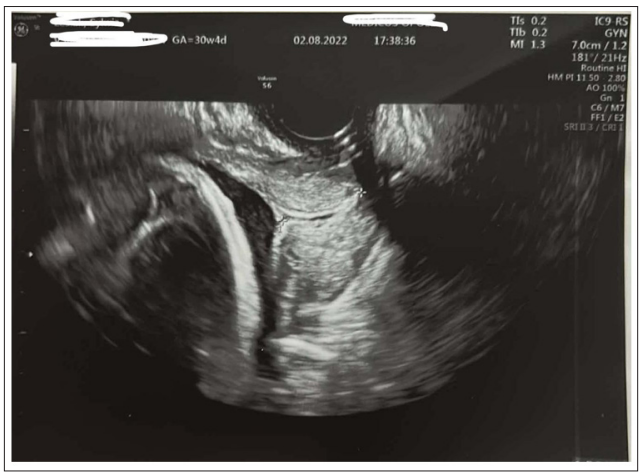

Figure 3: Transvaginal Ultrasound of Cervix. (Cervical Lenght) 21 mm (GA 30w0d)

2022: Department of Obstetrics and Gynecology
Patient at 30 weeks 0 days of gestation, admitted due to threatened preterm labor. Procedures performed: Fetal ultrasound, cardiotocography, urinalysis, urine sediment examination, complete blood count (CBC).
Findings: Cervical length 18 mm, internal os closed.
Management: Appropriate treatment initiated.

2022: Department of Obstetrics and Gynecology
Patient admitted at 38 weeks 4 days of gestation, vaginal delivery, live-born infant, complicated by rupture of the left labium majus.

Diagnostic Breakthrough
In 2024, the patient presented to her primary care physician due to persistent pain in the spine and knees, chronic fatigue, and easy bruising. A detailed medical history and thorough physical examination were performed.

Physical Examination Findings
Skin: Normal Color and Warmth, Small Yellow-Violet Ecchymoses (~1 cm) on the Lower Legs
Oral Mucosa: Normal
Thyroid: Not Enlarged on Palpation
Right Sternocleidomastoid Region: Keloid Scar after Excision of a Pigmented Skin Lesion
Lungs: Normal Vesicular Breath Sounds
Heart Rate: 80/min
Blood Pressure: 135/80 mmHg,
Oxygen Saturation: 98%,
Temperature: 36.6°C

Abdomen: Level with Chest, Soft, Non-Tender
Lower Legs: No Edema
Right Knee: 4 Scars, One Linear,
Left Knee: 6 Scars above the Joint;
Neck (Right Side): Keloid Scar;
Left Thigh: Fibroma ~7 mm in Diameter;
Posterior Left Thigh: Irregular Distribution of Adipose Tissue (Lipoma), Joint Hypermobility, Kyphoscoliosis, Scapular Asymmetry, Increased Skin Elasticity in the Elbow Region, Peripheral Pulses: Present

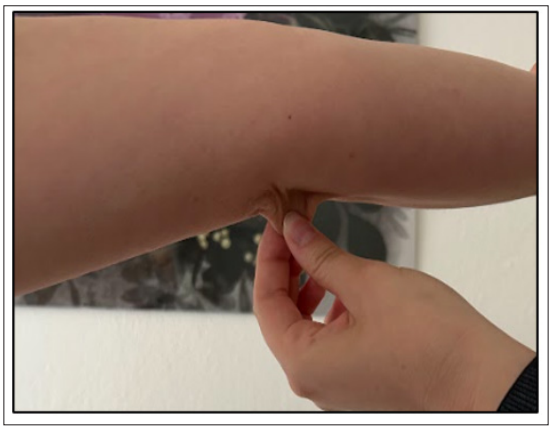

Figure 4: Excessive skin elasticity in the elbow area

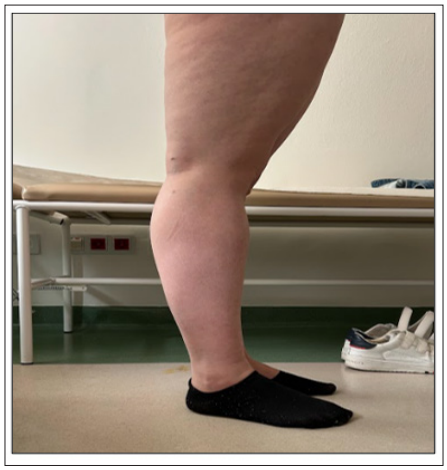

Figure 5: Hyperextension of the Knee Joints

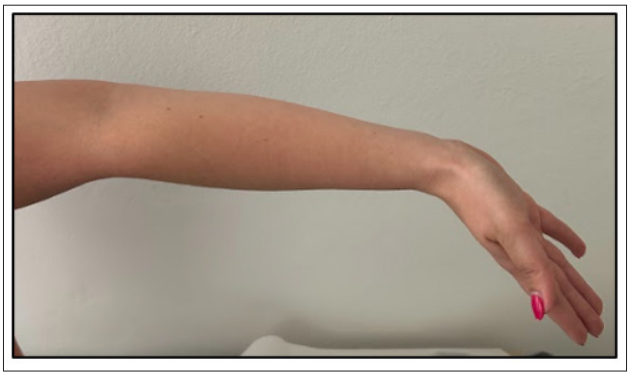

Figure 6: Hyperextension of the Elbow Joints

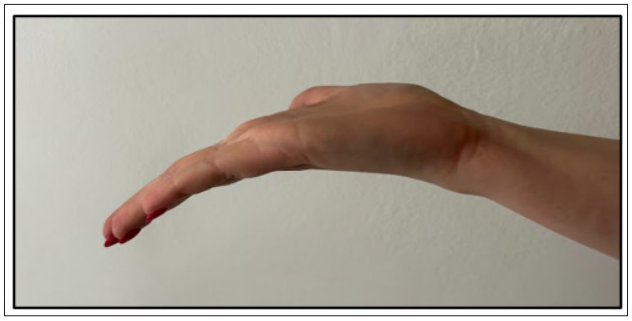

Figure 7: Hyperextension of the Wrist Joints

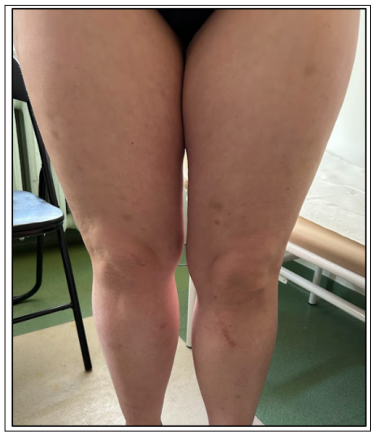

Figure 8: Petechie, Signs of Haemorrhagic Diathesis

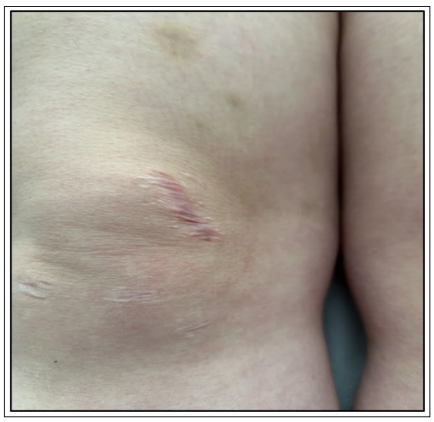

Figure 9: Scars Post Multiple Arthroscopies

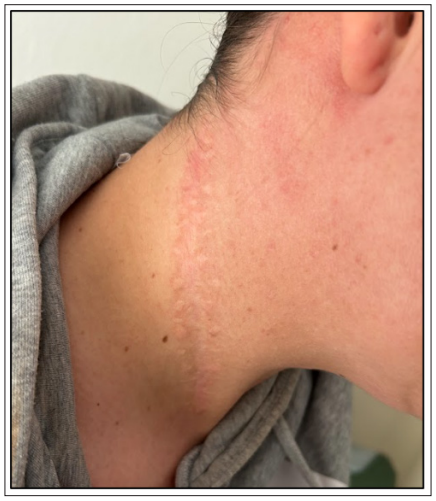

Figure 10: Keloid Scar on the Neck

Table 1: Beighton Criteria for Joint Hypermobility

Joint/Finding	Negative	Unilateral	Bilateral
Passive dorsiflexion of the 5th finger >90°	0	1	2
Passive flexion of thumbs to the forearm	0	1	2
Hyperextension of the elbows beyond 10°	0	1	2
Hyperextension of the knees beyond 10°	0	1	2
Forward flexion of the trunk with knees fully extended and palms resting on the floor	0	1	

A total score of ≥5 is considered positive for the presence of generalized joint hypermobility.

The patient's history revealed multiple episodes of joint dislocations and sprains, chondromalacia of articular cartilage, numerous arthroscopic procedures, recurrent generalized joint pain, vascular fragility, and a sense of generalized fatigue. Importantly, a diagnosis of joint hypermobility syndrome had previously been made in the rheumatology department, but no further diagnostics were performed.

The physical findings—joint hypermobility, vascular fragility, hyperextension of the elbows, knees, and wrists—together with a detailed personal and family history (similar symptoms in the patient's father and paternal grandmother) led to the suspicion of Ehlers-Danlos syndrome. The preliminary diagnosis was based solely on history and physical examination.

The patient was referred to a genetics clinic, where genetic testing confirmed the classical form of Ehlers-Danlos syndrome, with the presence of a VUS-type mutation in the COL5A1 gene [4].

2025: Genetics Clinic – Final Diagnosis of Ehlers-Danlos Syndrome

Molecular Diagnostics

Panel testing of genes associated with elastopathies revealed a variant in the COL5A1 gene: c.17G>T (p.Arg6Leu). This is a missense variant caused by the substitution of guanine with thymine at nucleotide position 17, resulting in the replacement of arginine with leucine at position 6 of the encoded protein. The variant has been classified as a VUS (Variant of Uncertain Significance).

The c.17G>T (p.Arg6Leu) variant has been classified as a VUS. Parental testing is necessary to determine whether this variant segregates with the clinical features of Ehlers-Danlos syndrome (EDS). Only after such family studies can a final consultation report be issued.

After family history analysis, the patient's relatives were also tested, and Ehlers-Danlos syndrome was confirmed in the patient's father and paternal grandmother.

Discussion

Delayed Diagnosis – A Global Problem

Delayed diagnosis of Ehlers-Danlos Syndrome (EDS) is a problem not only in Poland but worldwide. Studies conducted in Australia have shown that more than half of EDS patients reported their first symptoms over 15 years before receiving the correct diagnosis, and three-quarters received other diagnoses before the correct one was established [3].

Numerous, seemingly unrelated symptoms and complaints are treated separately, without a holistic view of the overall clinical picture. The complexity of the syndrome, with features such as hyperextension of the wrists, knees, and elbows, impaired wound healing with keloid formation, vascular fragility manifesting as easy bruising, gynecological and obstetric complications (heavy menstrual bleeding, preterm labor, miscarriages), as well as a multitude of non-specific symptoms (knee and elbow pain, fatigue, headaches, segmental spinal pain), makes diagnosis challenging. The difficulty in linking all symptoms leads to referrals to inappropriate specialists and repeated, costly imaging and laboratory tests—multiple MRIs, CT scans, and laboratory investigations—which, without comprehensive interpretation, prolong the diagnostic process and increase costs.

An additional problem is that approximately 90% of EDS cases lack a defined genetic cause, and diagnosis is based on symptoms and clinical presentation, as reported by patients and found on physical examination.

A study by R. Trudgian and T. Flood (2024) involving over 150 EDS patients in Australia showed that more than half noticed and reported their first symptoms over 15 years before the correct diagnosis. Additionally, three-quarters received other diagnoses before the correct one. Surveyed patients reported consulting many specialists, but in addition to the lack of a correct diagnosis, they also reported not being listened to and insufficient specialist knowledge about their condition [3].

Quality of Life and Social Functioning

Patients with Ehlers-Danlos syndrome, due to their symptoms—spinal pain, persistent fatigue, frequent injuries to the knees and elbows—often experience occupational and social exclusion.

Norwegian studies have shown that over 40% of surveyed patients reported their symptoms as an obstacle to work, and half retired early due to deteriorating health [5]. More than one-third of study participants declared that they could have continued working if their workplace had been adapted to their needs.

A study in Norway showed that individuals diagnosed during their student years, compared to adults, reported fatigue less frequently and had higher life satisfaction [5]. Over 40% of surveyed patients reported their symptoms as an obstacle to work, and half stated that their retirement was due to worsening health. More than one-third said they would like to continue working if their workplace was adapted.

Individual, Social, and Economic Costs

Scientific reports indicate that early diagnosis of Ehlers-Danlos syndrome allows for the avoidance of excessive, costly, and ineffective diagnostics, reduces unnecessary treatment costs, and prevents patients from taking ineffective medications, which generates additional financial burden.

Delayed diagnosis generates global healthcare costs, as timely and accurate diagnosis enables early rehabilitation, appropriate care, and better control of this incurable disease. Early and accurate diagnosis also reduces treatment costs, and good disease control leads to less absenteeism among affected patients.

Furthermore, early implementation of rehabilitation and appropriate treatment can reduce complications that result in exclusion from professional work, generating additional costs related to sick leave, disability, and sickness benefits.

Early diagnosis is particularly important for women—obstetric care for pregnant women with EDS is challenging, but earlier diagnosis allows for better preparation of the obstetric team to manage pregnancy in this group of patients [2].

In our patient's case, a significant portion of resources was consumed by numerous consultations, tests, and procedures that did not lead to improvement but only delayed the correct diagnosis. Additionally, the lack of a proper diagnosis resulted in frequent sick leave due to progressive complications of the undiagnosed disease, contributing to increased social and healthcare costs.

Summary and Conclusions

The presented case illustrates the typical diagnostic pathway of an EDS patient—years of searching for the cause of symptoms, numerous specialist consultations, fragmented care, and lack of coordination. The key role in establishing the diagnosis was played by the primary care physician, who took a holistic view and integrated data from various medical fields.

Based on the history of frequent joint dislocations, joint hypermobility, chronic fatigue and non-specific pain, recurrent vascular fragility, obstetric complications, and physical examination findings (increased skin elasticity, vascular fragility, joint hyperextension, keloids), the physician made a preliminary diagnosis of a collagenopathy, which was confirmed by genetic testing.

This case also prompts reflection on the approach to the patient—in the authors' opinion, only a holistic approach, with thorough data analysis, patient and attentive listening to all reported symptoms, as well as professional diligence and inquisitiveness, allows for the implementation of appropriate management and diagnostics.

Early detection of rare diseases such as EDS is crucial for public health—it shortens the time to diagnosis, improves patients' quality of life, and optimizes the use of healthcare resources. Our analysis highlights the urgent need for systemic solutions: better care coordination and increased awareness and education of physicians regarding rare diseases, especially at the level of primary healthcare.

Promoting a holistic approach to the patient is key to making the right diagnoses at the right time, which limits the patient's journey through the healthcare system, allows for better use of healthcare funding, and improves both the quality of life and the cost of care. Teaching proper communication between physician and patient is the path to improving quality in medicine [6-12].

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