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Case Report

Autosomal Dominant Spondyloepiphyseal Dysplasia (COL2A1 Type) in a Young Adult: Multidisciplinary Management According to Latest Recommendations – A Familial Case Report

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Abstract: We present the case of a 33-year-old patient who has been followed since the age of 16 for autosomal dominant spondyloepiphyseal dysplasia (SED), in an evocative familial context (his sister has the same clinical picture). The disease began in adolescence with disabling mechanical pain in the hips and knees, with a walking distance limited to 500 metres. Clinical examination showed bilateral knee flexion contracture, generalised stiffness of the hips, shoulders, elbows, wrists and fingers, girdle muscle wasting, and a BMI of 16 (47 kg for 1.67 m). The patient walks with a cane. Symptomatic treatment combining analgesics, NSAIDs and physiotherapy was initiated. The article details the diagnostic arguments, indications for bilateral hip arthroplasty, anaesthetic precautions and the role of genetic counselling. Medium-term outcome was favourable, with improvement in pain and walking distance after total hip replacement.

Keywords: Autosomal Dominant Spondyloepiphyseal Dysplasia, COL2A1, Early Osteoarthritis, Total Hip Arthroplasty, Multidisciplinary Management.

INTRODUCTION

Spondyloepiphyseal dysplasias (SED) are a heterogeneous group of genetic skeletal disorders affecting the vertebrae and epiphyses, with an overall prevalence estimated at approximately 1 in 100,000 births [1]. The autosomal dominant form, linked to mutations in the COL2A1 gene encoding type II collagen, represents the most frequent entity in adults after the congenital form [2]. Unlike the X-linked late-onset form (TRAPPC2), autosomal dominant SED can present at any age, from childhood to adulthood, with complete penetrance but highly variable expressivity [3]. Early joint manifestations, particularly severe osteoarthritis of the hips, knees and shoulders, often occur as early as the third decade, leading to major functional disability [4]. Recent recommendations advocate a multidisciplinary approach involving rheumatology, orthopaedics, physiotherapy, genetics and sometimes cardiology, due to an increased risk of extra-articular complications (myopia, deafness, aortic aneurysm) [5]. We report the case of a 33-year-old patient followed for autosomal dominant SED probably related to COL2A1, with severe functional impairment, and discuss current therapeutic management [6].

CASE REPORT

The patient is a 33-year-old right-handed man with no relevant past medical history, from a sibship of two children (one sister affected by the same condition). The diagnosis of autosomal dominant SED was made at the age of 16 on the basis of slowed statural growth (final adult height 1.67 m) and chronic hip pain that had started around 10-12 years of age. Standard radiographs performed during adolescence showed diffuse

platyspondyly, irregularities and deformities of the epiphyses of all joints without metaphyseal abnormality. Genetic testing was not performed at that time; the diagnosis was based on the combination of clinical, radiological and familial features.

At the age of 33, the patient consulted for worsening mechanical pain in the lower limbs, mechanical in timing, relieved by rest but sometimes nocturnal. Walking distance was limited to 500 metres, requiring the use of a unilateral elbow crutch. Clinical examination revealed an asthenic patient with a body mass index of 16 (weight 47 kg, height 1.67 m), suggesting relative undernutrition related to chronic pain and disability (Figure 1). Osteoarticular examination showed:

- Bilateral knee flexion contracture estimated at 20° (complete extension impossible), with maximal flexion of 100° bilaterally.
- Quadriceps and gluteal muscle wasting.
- Severely reduced hip ranges of motion (scored using the French Society of Rheumatology method):
- Right hip: internal rotation 0°, external rotation 0°, flexion 45°, extension -5°, abduction 20°, adduction 10°.
- Left hip: internal rotation 0°, external rotation 10°, flexion 60°, extension -5°, abduction 20°, adduction 20°.
- Generalised stiffness of the upper limbs: limited anterior elevation of the shoulders to 90°, elbow flexion to 100°, stiff wrists and fingers with reduced grip strength.

Inflammatory markers (CRP, erythrocyte sedimentation rate) were normal, excluding inflammatory rheumatism. Radiographs of the pelvis and hips showed bilateral joint space narrowing, subchondral osteocondensation and geodes.

Updated standard radiographs of the large joints systematically showed: enlargement of the epiphyses of the long bones (humerus, radius, ulna, femur, tibia, fibula) with a blown-out, irregular appearance, scattered with multiple subchondral geodes (clear lacunar images without a sclerotic rim), creating a pseudocystic structure. There was no aggressive osteolysis or cortical breach. Joint spaces were generally narrowed, with moderate osteocondensation. Multiple osteochondromas were present at both knees and the right shoulder. No associated metaphyseal abnormalities were found (Figures 2-3-4-5).

Radiographic evaluation of the spine, especially dynamic views to search for atlantoaxial instability, as well as cardiac and aortic ultrasound to look for possible aortic aneurysm, were requested but not performed by the patient. Genetic counselling was offered to the patient and his sister: after confirmation of the COL2A1 mutation by Sanger sequencing, prenatal or preimplantation diagnosis could be discussed for the sister (and for the patient's partner) if they have a child project, since the risk of transmission to offspring is 50%.

MRI Was Not Requested

Technetium-99m bone scan: described polyostotic involvement of spondyloepiphyseal dysplasia, with signs of activity (peri-articular ossifications and bony bridges) predominating in the hips and knees (Figure 6).

Current Treatment Combines:

- Step I (paracetamol 3 g/day) and step II (tramadol 100 mg/day if needed) analgesics.
- Non-steroidal anti-inflammatory drugs (ibuprofen 400 mg) on demand, with limited duration to avoid digestive and renal side effects.
- Physiotherapy twice a week: stretching of the posterior muscle chains, isometric strengthening of the quadriceps and gluteal muscles, proprioceptive training.

Bilateral total hip arthroplasty was proposed to the patient, but he preferred to defer any surgical procedure until an older age.

Figure 1: Clinical frontal and lateral images showing the patient in a standing position

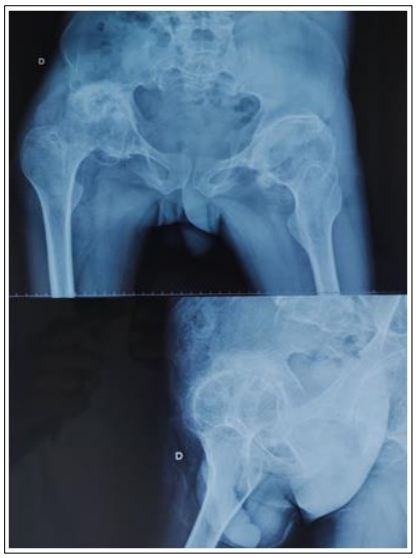

Figure 2: Globular femoral heads, increased in volume, with a mottled structure made of numerous confluent geodes, enlarged and shallow acetabula, narrowing of the overall joint line.

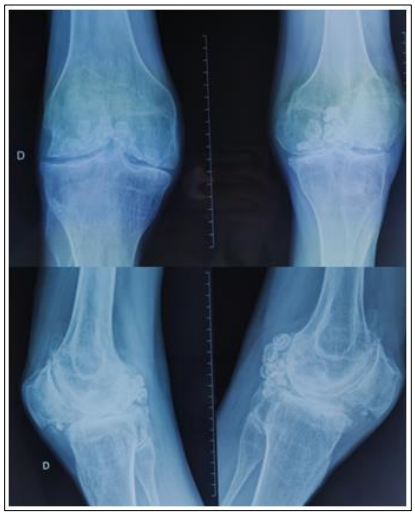

Figure 3: Distal femoral epiphyses and tibial plateaus are very wide, with bumpy contours, multiple subchondral geodes (bubble appearance), and medial and lateral femorotibial narrowing. Multiple osteochondromas are also present

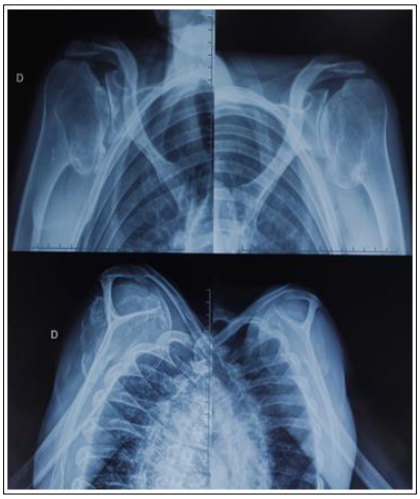

Figure 4: Proximal epiphyses of the humeral heads increased in volume, irregular contours, multiple subchondral geodes, narrowed glenohumeral interspaces, with moderate osteosclerosis, osteochondromas of the right shoulder

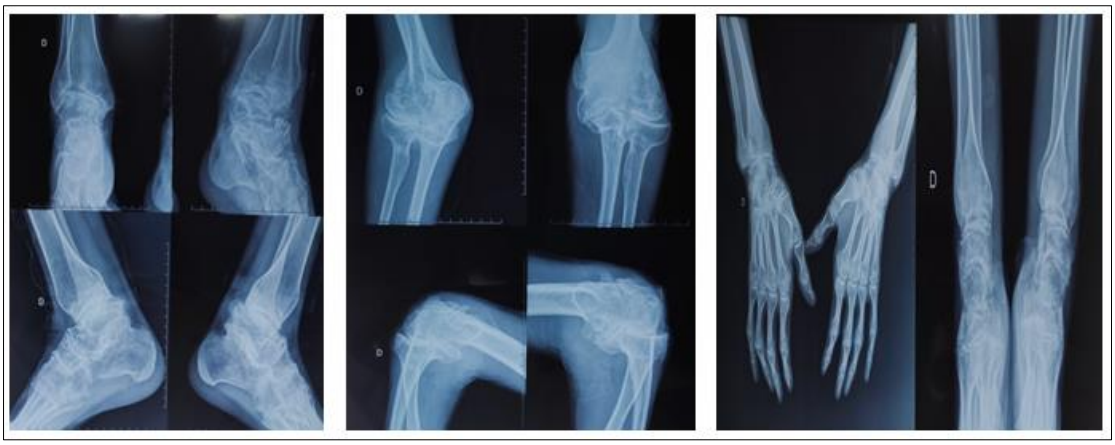

Figure 5: X-rays of ankles, elbows, wrists: Epiphyses enlarged in volume, clear cystic images (geodes), joint narrowing

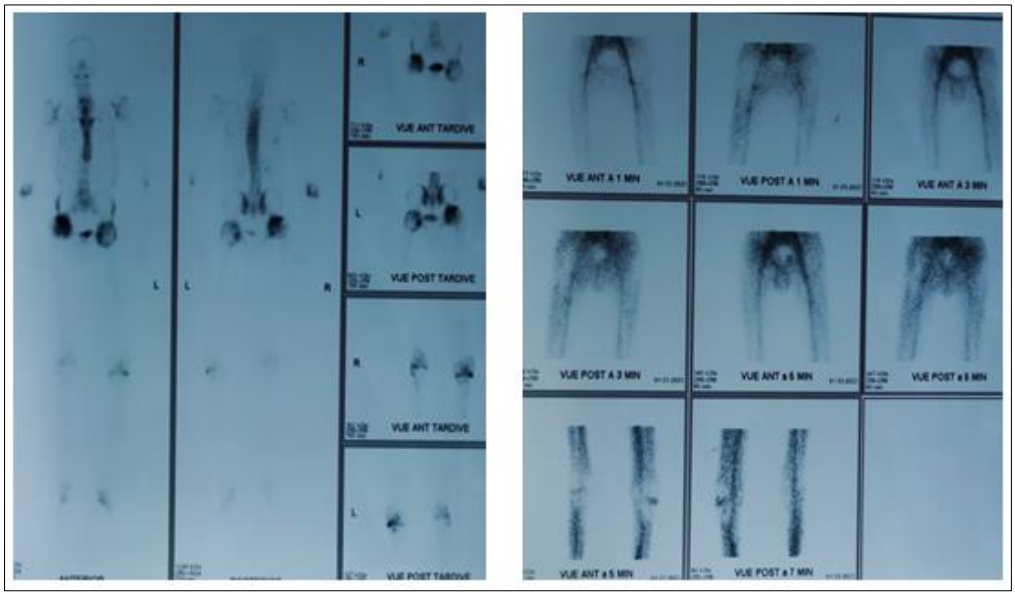

Figure 6: Bone scintigraphy: - Hips: Intense, heterogeneous hyperfixation, straddling the hypertrophied femoral heads and acetabulum, more pronounced on the right, suggestive of periarticular ossifications

- Knees: Moderate, heterogeneous, linear hyperfixation, involving the medial and lateral femorotibial compartments, more pronounced on the left, suggestive of active bone bridges.
- Ankles, elbows, shoulders (humeral heads): Heterogeneous hyperfixation with bone deformities.
- Spine: Spinal hypertrophy, slight thoracolumbar scoliosis.
- Remainder of the skeleton: Homogeneous and symmetrical uptake, without abnormalities.

DISCUSSION

Autosomal dominant SED related to COL2A1 accounts for approximately 20% of non-lethal SED, with an estimated prevalence of 1/200,000 births, i.e. rarer than the X-linked form but more severe in terms of joint involvement [1]. The differential diagnosis includes X-linked late-onset spondyloepiphyseal dysplasia (absence of affected sister, height < 160 cm) and other collagen dysplasias (type II, type IX) [2]. In our case, a height of 1.67 m and the sister's involvement strongly point to autosomal dominant inheritance, with a probable mutation in the COL2A1 gene on chromosome 12q13 [3]. The age of onset at 16 years is late for congenital SED (which manifests at birth or early childhood), but it is perfectly compatible with attenuated forms of COL2A1, where symptoms may not appear until adolescence or adulthood [4]. The bilateral knee flexion contracture of 20° and hip stiffness with zero indifferent rotation are classic signs of advanced osteoarthritis secondary to dysplasia, justifying early intervention before complete ankylosis [5]. A walking distance of 500 metres and a BMI of 16 reflect severe functional and nutritional impact, with an increased risk of sarcopenia.

Although standard radiographs are sufficient for diagnosis, MRI and bone scintigraphy can provide additional information for functional management [6]. MRI allows precise assessment of articular cartilage status, detection of early osteonecrosis or osteophyte-synovial impingement, and investigation of

compressive myelopathy in cases of atlantoaxial instability [7]. Technetium-99m bone scintigraphy, less specific, remains useful in patients with polymorphic pain: it identifies sites of active metabolic hyperfixation, guiding the choice of the "target" joint responsible for the most disabling pain before infiltration or arthroplasty [8]. In our observation, MRI was not performed initially because the radiological and scintigraphic findings were already highly suggestive. However, MRI could be proposed in the event of worsening pain or suspicion of aseptic necrosis.

Current recommendations from the European Society of Paediatric Radiology (ESPR) and the working group on bone dysplasias advocate systematic screening for extra-articular complications: myopia or retinal detachment by annual ophthalmic examination, conductive hearing loss by audiometry every 5 years, and ascending aortic aneurysm by echocardiography every 2-3 years [9].

Total hip arthroplasty is the most beneficial intervention in young patients with SED, providing excellent functional results at 10 years, despite a higher risk of aseptic loosening (approximately 15% at 15 years) due to the small size of the implants and osteopenia [10]. In forms with severe coxa vara, the use of extra-thin (diameter ≤ 8 mm) or custom-made femoral stems improves primary fixation [11]. Preoperative (muscle strengthening) and postoperative (range of motion recovery) physiotherapy should be supervised by an experienced therapist to avoid dislocations [12]. Because of odontoid hypoplasia and the risk of atlantoaxial instability, any intubation or cervical manipulation (anaesthesia, spinal surgery) must be preceded by dynamic radiological assessment, and protection with a rigid cervical collar is indicated [13]. Genetic counselling is essential: the risk of transmission to offspring is 50% for each child, with possible phenotypic variability; prenatal diagnosis by fetal DNA analysis (chorionic villus sampling or amniocentesis) can be offered [14]. In the absence of curative treatment, multidisciplinary management (rheumatologist, orthopaedic surgeon, geneticist, physiotherapist, dietitian) helps optimise quality of life and prevent complications [15].

CONCLUSION

Autosomal dominant spondyloepiphyseal dysplasia (COL2A1 type) is a rare but severe disorder in young adults; the diagnosis should be suspected in the presence of early familial hip and knee osteoarthritis. Management combines symptomatic treatment, adapted physiotherapy and, most importantly, bilateral total hip arthroplasty to restore walking ability. Specialised follow-up for extra-articular complications (ophthalmic, auditory, aortic) and genetic counselling are mandatory. Our case illustrates the need for a personalised, coordinated approach across multiple specialties to improve long-term functional outcome.

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