

# A distinct form of spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)-leptodactylic type: radiological characteristics in seven new patients

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## Abstract

**Objective** This study presents seven cases of a rare but distinctive form of spondyloepimetaphyseal dysplasia with joint laxity-leptodactylic or Hall type to emphasize the characteristic clinical and radiological findings.

**Materials and methods** A multiinstitutional retrospective review was performed on seven patients. The patient population consisted of one family with an affected mother and two siblings and four unrelated patients; there were one adult, aged 40 years, and six children, ranging in age from 3 to 12 years. The gender ratio of females to males was 5 to 2. We reviewed the clinical data and skeletal surveys and focused on radiographs of the pelvis, knees, hands, and spine.

**Results** The outstanding clinical features were short stature, midface hypoplasia, and multiple dislocations and/or ligamentous laxity of the large joints, particularly at the knees with a genu valgum or varum deformity. Of seven patients, six patients showed normal intellect but one patient had mild mental retardation. The main radiological features included small, irregular epiphyses, metaphyseal irregularity with vertical striations that was a constant finding at the knees, constricted femoral necks, delayed ossification of the carpal bones, and slender metacarpals. Progressive thoracolumbar scoliosis was evident with aging; however, the vertebral bodies appeared normal in height or mild platyspondyly was noted.

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**Conclusion** In view of the orthopedic management of multiple joint dislocations and ligamentous laxity of the large joints, awareness of this disease entity and diagnostic precision solely based on radiological findings is of importance, particularly as the disorder is currently more common than initially reported.

**Keywords** Skeletal dysplasia · Joint laxity · Multiple dislocations · Short stature · Spondyloepimetaphyseal dysplasia · Sponastrime dysplasia

## Introduction

The spondyloepimetaphyseal dysplasias (SEMDs) are a heterogeneous group of disorders characterized by multifocal involvement of the epiphyses and metaphyses of the long tubular bones with mild to severe changes of vertebral bodies that are distinguished based on clinical and radiological features. Of the SEMDs group, SEMD with joint laxity (SEMDJL)-leptodactylic or Hall type is an uncommon disorder that was proposed as a distinct entity in the revised Nosology and Classification of Genetic Disorders of Bones [1]. The purpose of this report is to present a rare type of skeletal dysplasia in a newly discovered group of seven patients and to increase awareness of the radiological findings to avoid under/misdiagnosis when joint laxity and multiple dislocations are encountered in SEMDs patients.

## Materials and methods

We reviewed the data on six Korean patients and one Japanese patient from multiple institutions, where a panel of skeletal dysplasia experts confirmed the working diagnosis. The study was approved by the institutional review board of each institution. The patient group was composed of one family with affected daughter, mother, and son (patients 1, 2, and 3) and four isolated children (patients 4, 5, 6, and 7) from unrelated normal parents.

Patient 1 is a 7-year-old girl, the daughter of patient 2. This patient was first seen at the age of 3 years due to stunted growth and unstable walking. The patient was short and had a genu varum deformity. Under the impression of pseudoachondroplasia, the patient underwent distal femoral and proximal tibial osteotomies. The patient revisited the hospital because of recurrent knee joint instability and a genu varum deformity. When seen at the age of 7 years, the patient was short (99.5 cm, less than first centile). The patient had similar facial features as compared to her mother with midface hypoplasia, a saddle nose, and a prominent forehead. The patient had lax joints, particularly

at the knees with an asymmetric right valgus and left varus deformities.

Patient 2 is the mother of patient 1, the only adult in this study group. This patient first came for medical attention around the age of 10 years due to a short stature and instability of the knees while walking. The patient had been erroneously diagnosed with achondroplasia, and thereafter, no follow-up medical examination was performed. The patient was currently reexamined together with her daughter. When seen at age of 40 years, the height was 114 cm, and the patient had similar but more severe facial dysmorphology with marked depression of the nasal bridge, a prominent forehead, and a short neck. The range of movements of the hip and knee joints was increased; particularly, profound instability of both knees with over 40° of varus and valgus laxity was noted. The patient also had a right hip dislocation that was not noticed by the patient herself.

Patient 3, the son of patient 2, is a 5-year-old and came for medical attention due to a positive family history. The patient was short (height 72 cm, less than first centile). The facial features were the same as with his mother and sister. The patient showed a mild genu varum deformity and had difficulty in running.

Patient 4 is a 12-year-old girl who underwent follow-up for a corrective genu valgum deformity under the impression of multiple epiphyseal dysplasia. The developmental milestones of the patient in early childhood were normal. The patient was noted to have genu valgum and a waddling gait at the age of 3 years that had progressed, and subsequently, the patient underwent corrective orthopedic procedures to her lower limbs at the age of 6 years. On a current medical examination at the age of 12 years, her height was 129 cm (less than second centile). The patient showed an asymmetrical genu valgum and had a flat foot deformity. Facial features showed mild midface hypoplasia, but otherwise were not remarkable. The patient was described as having multiple lax joints in the large joints as well as in the fingers and wrists. The patient has developed thoracolumbar scoliosis.

Patient 5 is a 7-year-old girl that has received follow-up for the impression of multiple epiphyseal dysplasia. Development was normal, except for an atrial septal defect that was corrected at the age of 2 years. At approximately the age of 3 years, the patient developed a genu valgum deformity that required surgical correction. At the age of 7 years, the patient underwent additional bilateral femoral osteotomy, and subsequently, radial head dislocation was detected.

Patient 6 is a 10-year-old girl. The patient had a delay in developmental milestones. She was able to walk at 2 years and had generalized hypotonia. The patient was first seen at the age of 2 years and was diagnosed with unknown

dwarfism, probably “a variant of achondroplasia”. Subsequently, her knees became unstable, and the patient developed progressive genu varum that required corrective osteotomy at the age of 7 years. When currently seen at the age of 10 years, the patient was noted as short (height 117 cm, less than first centile). She had striking joint laxity at both knees. This time the diagnosis of a form of spondyloepiphyseal dysplasia was considered.

Patient 7 is a 4-year-old Japanese boy. His perinatal history was not remarkable except for intermittent stridor. At age of 4 months, he had developed sudden dyspneic spell. On fiberoptic bronchoscopy, tracheomalacia was observed, and tracheostomy was performed for 4 months. His developmental milestones were delayed. At age of 2 years, he visited rehabilitation center due to gait disturbance, particularly unstable walking. That time the diagnosis was unknown ligamentous laxity involving the multiple joints. At age of 4 years, he transferred under the impression of “atypical achondroplasia”. The patient was short (height

87.5 cm, less than second centile). He showed mild mental retardation. The patient had an asymmetrical genu valgum and striking laxity at the elbows and hip joints.

All radiographs of the patients were taken from early to late childhood (age range, 3–12 years) and one adult at the age 40 years. For comparative purpose of each patient, the radiographic review focused on the pelvis, knee, hands/feet, and the spine.

## Results

Following a review of clinical and radiographic material, we have identified seven patients with an identical form of SEMD where consistent manifestations permit recognition and a diagnosis of the SEMDJL-leptodactylic type. The clinical and radiographic findings are summarized in Table 1.

Three patients consisted of an affected mother and two siblings (patients 1, 2, and 3) with evidence of autosomal

**Table 1** Summary of clinical and radiological findings of seven patients

	Patients						
	1	2	3	4	5	6	7
<b>Clinical findings</b>							
Gender	F	F	M	F	F	F	M
Age of radiographic examination (years)	3	40	5	12	7	7	4
Family history	+	+	+	–	–	–	–
Short stature	++	++	++	+	+	++	+
Midface hypoplasia	++	++	++	±	±	+	+
Genu valgum (vg)/varum (vr)	vr	vg	vg	vg	vg	vr	vg
Intelligence	N	N	N	N	N	N	R
Associated internal abnormalities	–	–	–	–	ASD	–	TM
<b>Radiographic findings</b>							
Femoral necks/short or long	long	short	long	long	long	long	long
Constricted or slender femoral necks	+	+	+	+	+	+	+
Small irregular epiphyses	+	+	+	+	+	+	+
Metaphyseal irregularity	+	+	+	+	+	+	+
Metaphyseal striations	+	+	+	+	+	+	+
Slender metacarpals/metatarsals	+	+	+	+	+	+	+
Small carpal bones	+	+	NO	+	+	+	NO
Delayed carpal bone ages	+	+	++	+	+	+	++
Scoliosis	–	++	NA	+	–	–	–
Vertebral end plate irregularity	–	+	NA	+	–	±	–
Lumbar posterior scalloping	–	–	NA	–	+	–	±
Narrow lumbar interpedicular distance	–	–	NA	–	–	–	–
Disc space narrowing	–	+	NA	+	–	–	–
Platyspondyly	–	–	NA	–	±	±	–
Radial head dislocation	–	–	–	+	+	–	+
Distal ulnar hypoplasia	–	–	–	±	+	–	–
Ivory phalangeal epiphyses	+	–	–	+	+	+	–

ASD atrial septal defect, F female, M male, N normal, NA not available, NO no carpal ossification, R mild mental retardation, TM tracheomalacia

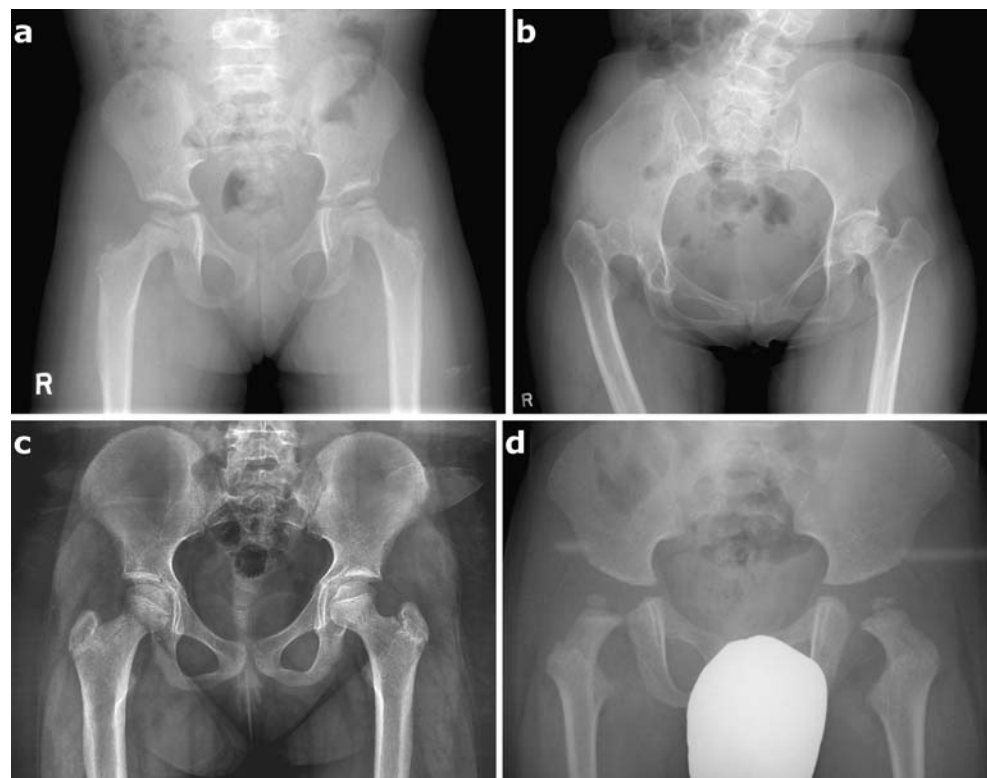
dominant (AD) inheritance. The other four patients (patients 4, 5, 6, and 7) were isolated cases from families with unrelated normal parents. All patients presented with a short stature, gait disturbance, and/or delayed walking. By the age of 3 to 5 years, the patients developed joint laxity, initially at the knees, that led to a genu varum or valgum deformity and subsequently involved the hips and ankles. Radial head dislocation was also present in patients 4, 5, and 7. The joints at the hand and wrist were lax in patient 4. Developmental milestones, particularly standing and walking, were delayed in patients 6 and 7, and also generalized hypotonia was noted in patient 6. No cleft palate was found in any patients. Patient 7 showed mild mental retardation but all remaining patients were of normal intelligence. There were no other clinical abnormalities, including neurological, ophthalmological, and auditory abnormalities. Most of the patients did not have significant internal organ abnormalities except for two patients: Patient 5 had congenital heart disease and had received surgery for an atrial septal defect, and patient 7 had undergone a tracheostomy for tracheomalacia. The results of blood and other laboratory examinations were all normal.

Radiographic abnormalities were present throughout the skeleton. The pelvis, knees, hands/feet, and spine were specific regions necessary to establish a firm diagnosis, and the characteristic radiological manifestations are described below.

**Pelvis** The capital femoral epiphyses were small, irregular, and flattened but well located within the acetabula (Fig. 1). Patient 2, the only adult in this series, showed hip dislocation on the right side and significant capital femoral epiphyseal changes consisting of flattened, dysplastic appearance, and hip joint space narrowing due to degenerative osteoarthritis on the left side (Fig. 1b). In patients 1 and 5, one side of the capital femoral epiphysis showed fragmentation and most probably represented superimposed aseptic necrosis (Fig. 1a, d). The femoral necks were consistently constricted and elongated in all children; however, the adult patient showed short femoral neck although that was still constricted in appearance. All children showed rather small iliac bones, but the ischium and pubis were of a normal appearance.

**Knees** All patients had a genu valgum or varum deformity of different severities (Fig. 2). The initial presentation of genu varum was noted in patients 1 and 6 (Fig. 2a, f), whereas a valgus deformity was noted in the remaining children. The epiphyses at the knee joints were small, flat, irregular and/or delta shape, and impinged upon the flattened distal femoral condyles in early childhood (Fig. 2a, c, e, g). In the adult patient, the femoral condyles and the tibial plateau were eroded and flat. The knee joint subluxated laterally and bilateral patellar dislocation was noted (Fig. 2b). The most conspicuous and consistent

**Fig. 1** Radiographs of the pelvis. **a** Patient 1 at age of 3 years. The femoral epiphyses are small and irregular, with aseptic necrosis of the left capital femoral epiphysis. The femoral neck is rather long and constricted. **b** Patient 2 at 40 years of age. The femoral epiphyses are severely flattened, almost not seen at the right side. The femoral necks are short but slender in appearance. Degenerative joint disease in both hip joints results in narrowing of the joint spaces and dysplastic derangement of the right acetabulum. The right iliac bone is tilted because of the hip dislocation. **c** Patient 4 at age 12 years. The femoral epiphyses are small and flattened. The femoral necks are slender. **d** Patient 7 at age of 4 years. The femoral epiphyses are small and irregular. The femoral necks are constricted and elongated, with mild coxa valga





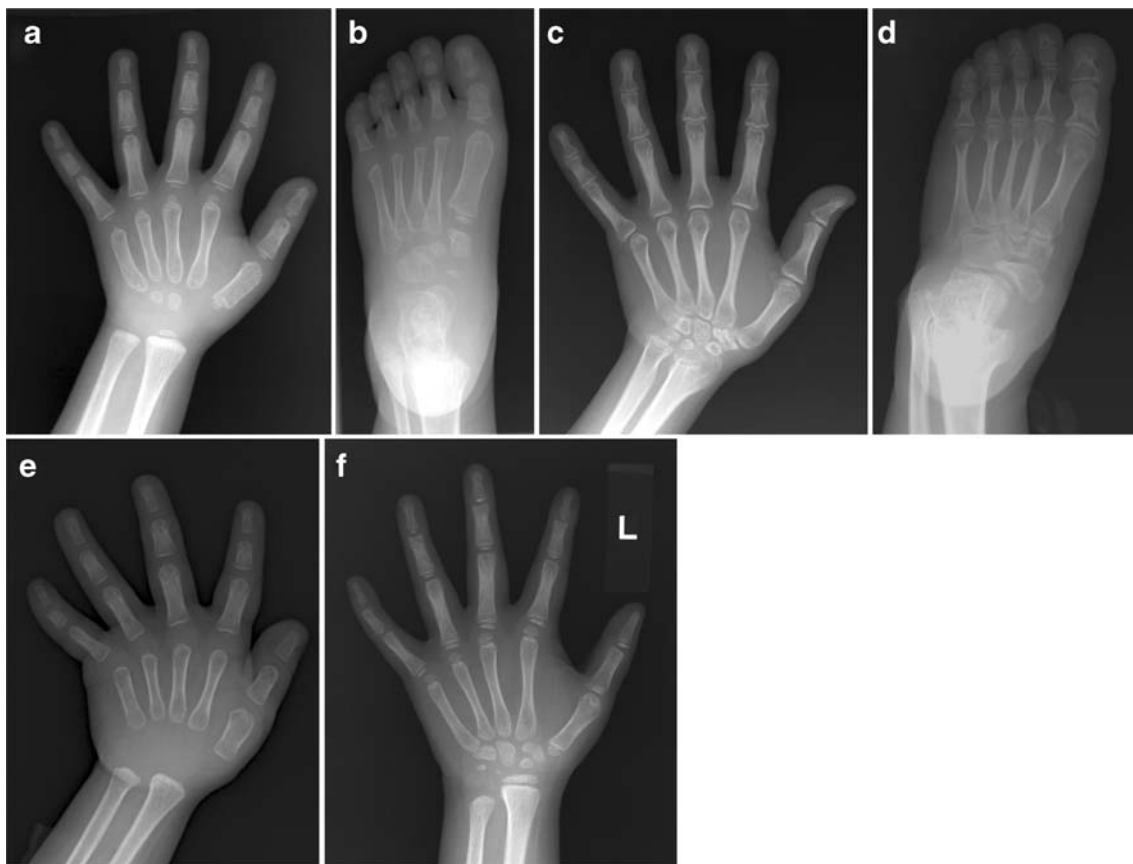
**Fig. 2** Synopsis of the knee radiographs in all patients. **a** Patient 1 at age of 7 years, **b** patient 2 at 40 years, **c** patient 3 at 5 years, **d** patient 4 at 12 years, **e** patient 5 at 7 years, **f** patient 6 at 7 years, **g** patient 7 at 4 years. Patients 1 and 6 show a genu varum deformity, whereas a symmetrical or an asymmetrical genu valgum is noted in the remaining patients. Patient 4 (**d**) had the right femoral osteotomy at the age of 7 years but had recurrent joint laxity that requires repeated corrective operation. The epiphyses are small, irregular and flattened.

The distal femoral epiphyses show delta shape and impinged upon the femoral condyles except for adult patient 2. The metaphyses are flared and irregular. There are thin or thick, irregular vertical striations in the distal femora and proximal tibial metaphyses in all patients. Similar sclerotic striations are also noted in the ankles. The adult patient (**b**) shows flat and eroded appearance of the distal femoral condyles and proximal tibial plateau. There is lateral subluxation of the right knee joints, with dislocation of the patella

findings at the knees were metaphyseal flaring and an irregularity with sclerotic metaphyseal striations. These metaphyseal vertical streaks were also found at the femoral necks and ankles but less profound and rarely at the wrists.

**Hands and feet** Radiographs of the hands were available in all patients and patients 1, 2, and 5 had additional feet radiographs. There were considerably consistent and identical abnormalities of the metacarpals in all patients. The characteristic changes were unusually slender metacarpals

and a particularly constricted appearance of diaphyses (Fig. 3). The metatarsals showed similar slenderness and constriction of middiaphyses with the absence of medullary spaces (Fig. 3b, d). In the hand, the phalangeal epiphyses were small and flat. In patients 1, 4, 5, and 6, multiple dense, ivory epiphyses were shown. The overall width of the phalanges was slender but not so striking as compared with the metacarpals. The size of the carpus space became narrow, and the individual carpal centers were small, flat, and irregular. The narrowing of the carpus was remarkable



**Fig. 3** Hands and feet radiographs. **a, b** Patient 1 at age of 3 years. The metacarpals and metatarsals, especially through two to four digits are slender. The carpal ossifications and phalangeal epiphyses are small for age and delayed. **c, d** Patient 2 at age of 40 years. The metacarpals and metatarsals are extremely slender, particularly constricted at the mid-diaphyses with almost obliteration of the medullary spaces. Individual carpal bones are small and irregular. The distal fibula is subluxated laterally, and the tarsal bones are small and flattened. **e** Patient 3 at age of 5 years. The hand reveals severe delay of bone age, totally absent of ossification of carpals and distal

epiphyses of the radius and ulna. Epiphyseal ossification of the proximal phalanges are vaguely seen at the second and third fingers. There is shortening and clinodactyly deformity of the middle phalanx of the fifth finger. **f** Patient 5 at age of 10 years. The carpal bones are small, and the bone age is markedly delayed, estimated between 5 and 6 years of age. The phalangeal epiphyses are small and ivory epiphyses are seen through the distal phalanges. Dense epiphysis of the third finger is fragmented. The epiphysis of the distal radius is flat. The distal ulna is hypoplastic and the epiphysis is not seen. The second to fourth metacarpals are slender

in the adult patient (Fig. 3c). All of the affected children showed retarded carpal bone age. In patients 3 and 7 at the age of 5 and 4 years, respectively, the distal radial epiphyses and carpal bones were totally not ossified (Fig. 3e). There was mild metaphyseal irregularity of the distal radius and ulna with some sclerotic streaks.

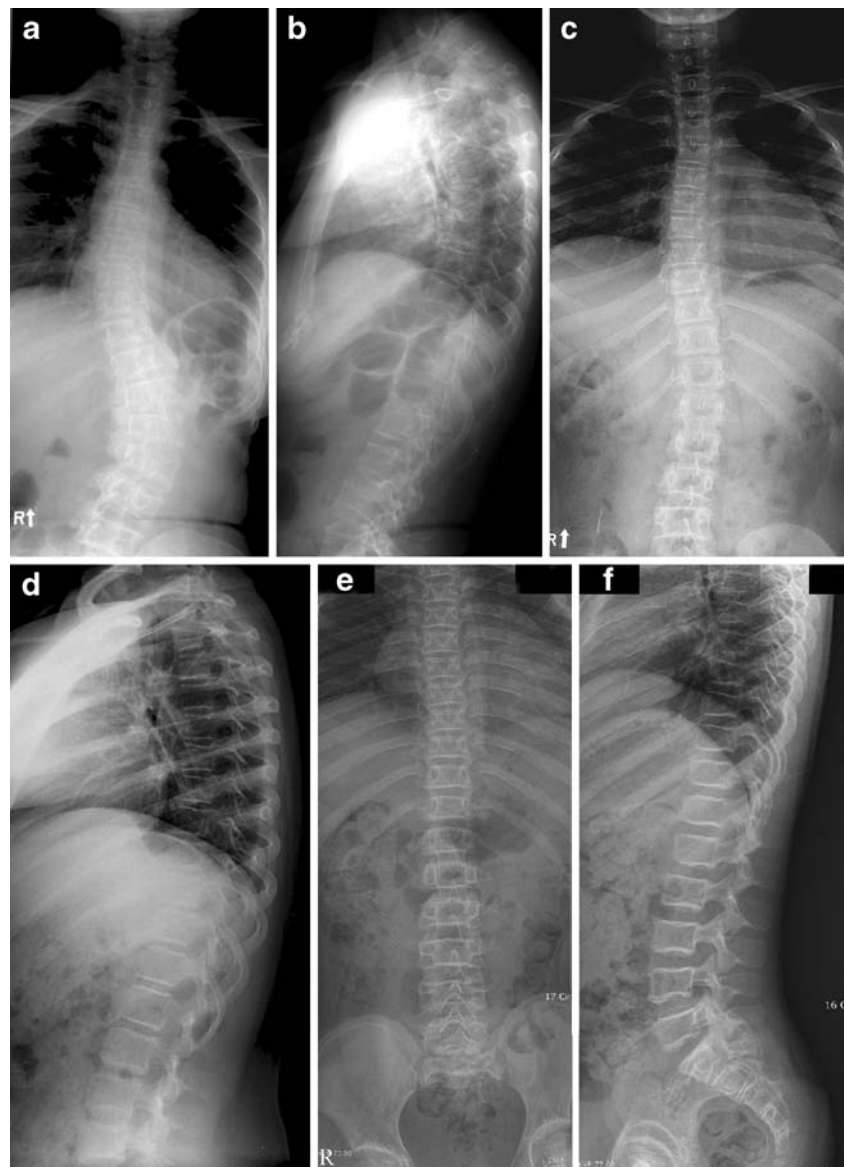
**Spine** Frontal and lateral spine films were not available for patient 3. The radiographic findings of the spine were normal to mild spondylar changes on the lateral view as compared with severe epimetaphyseal changes of the long bones (Fig. 4). There was subtle posterior scalloping of the vertebral bodies in patients 5 and 7. Interpedicular distance narrowing of the lumbar spine was inconspicuous in all patients, but it was difficult to evaluate in patient 2 due to scoliosis. Mild irregularity or concavity of the vertebral end

plates was noted in patients 2, 4, and 6. The presence of mild to severe scoliosis was detected in patients 2 and 4 that seemed to progress with age (Fig. 4a, c).

## Discussion

The International Nosology and Classification of Constitutional Disorder of Bone first described two disorders of SEMD with dislocation and joint laxity: SEMDJL and SEMD with multiple dislocations, the Hall or leptodactylic type [2]. Recently, the Nosology Group of the International Skeletal Dysplasia Society has released the 2006 revision of Nosology and Classification of Genetic Skeletal Disorders where they proposed the use of the united term “joint

**Fig. 4** Frontal and lateral radiographs of the spine. **a, b** The spine radiographs of patient 2 at age of 40 years. There is a thoracolumbar scoliosis, with biconcavities of the vertebral body endplates. Platypondyly is not evident. **c, d** Patient 4 at age of 12 years shows mild thoracolumbar scoliosis. The vertebral bodies are normal in height, with mild endplates irregularity at lower lumbar area. **e, f** Patient 6 at age of 10 years. There is mild platyspondyly with concavity at the lower end plates of the fourth and fifth and lumbar vertebral bodies



laxity” instead of the term “multiple dislocations” [1]. These included two conditions: SEMDJL-Beighton type (autosomal recessive) and SEMDJL-leptodactylic or Hall type (autosomal dominant inheritance). Of these, the leptodactylic type is the more descriptive term as the slender appearance of the metacarpals is so distinctive as to make a definitive diagnosis.

The clinical and radiological findings of the patients in this study are compatible with the typical findings of the SEMDJL-leptodactylic type as described primarily by Hall et al. [3]. This type of skeletal dysplasia usually presents with short stature. Other clinical characteristics include articular abnormalities associated with generalized joint laxity such as a genu varum or valgum deformity with or without patella subluxation, talipes equinovarus, and hip and elbow dislocations. Many of the typical

radiological findings of the disorder are distinguishable at a very early age and evolve over time with growth [4]. In our patients, an earlier radiographic examination was performed at the age of 3 years, and the diagnostic radiological findings were apparent.

SEMDJL-leptodactylic type has been reported to be inherited as AD trait or sporadic occurrence [1]. There have been two previous reports that have described AD inheritance of this disorder. In one report, the disorder was inherited from father to daughter [5], and in the other report, the disorder was inherited from mother to son [6]. Our report is the third family reported to date with evidence of dominant transmission of this disorder from a mother to a daughter and son continuously. Other published cases of the SEMDJL-leptodactylic type have all been sporadic [3–8]. In our series, the remaining four

patients were sporadic cases with normal unrelated parents.

The ratio of the affected gender has been shown to be equal [3]. However, female patients are more affected in our series (M/F=2:5) and in a report by Nishimura et al. (M/F=1:3) [4] although the clinical and radiological manifestations are not different between genders. Previously, three adult patients have been described in the literature [4, 5], and we describe the fourth adult to date with the SEMDJL-leptodactylic type. As compared with the previously described adult cases, our adult patient also presented with a severe short stature of less than 120 cm. There were no internal malformations in our adult patient and the two other reported adult patients, and thus, life expectancy seems to be normal.

Facial features seen have been variable and include mild midface hypoplasia to a severely depressed nasal bridge with a broad and upturned nose. In our patient group, the familial cases showed more severe features of facial dysmorphism. Short stature is usually the outstanding clinical abnormality that brings the patient to medical attention. The patients have a variable degree of short stature, all values largely less than third centile for age-matched normal height. By virtue of the striking short stature, misdiagnosis may occur. In our patient series, atypical achondroplasia, pseudoachondroplasia, multiple epiphyseal dysplasia, and spondyloepiphyseal dysplasia were the impressions before this review.

Other uncommon clinical presentations reported in the literature include laryngotracheomalacia, muscular weakness probably due to myopathy, and mild mental retardation [3, 4, 6]. In our series, one patient only had manifested tracheomalacia requiring tracheostomy as well as having mild mental retardation.

Joint problems for patients with the SEMDJL-leptodactylic type include subluxation of the large joints, particularly at the hip and knee joint with patella dislocations, and less frequently at the elbow and wrist joints. Due to the ligamentous instability of joints, hip, and knee, dislocations and feet deformities are often persistent after the corrective osteotomies for recurrent bony deformity. Of our patients, three children had recurrent instability of the knees after corrective osteotomy that required repeated surgery. In addition to instability of joints in the extremities, thoracolumbar scoliosis that is progressive with age was noted. In our patients, the adult patient and follow-up data on the pre-teenage patient 4 demonstrated a scoliosis as compared with the other patients of the younger ages. With advancing age, premature osteoarthritis of the hip joints is also a problem. The adult patient 2 showed degenerative hip arthropathy that resulted in dysplastic acetabulum and hip dislocation.

Hand changes are particularly characteristic. The metacarpals are uniformly gracile, and the phalanges also appear to be slender without shortening. There is also significant delay in the appearance of the phalangeal epiphyses and the ossification of carpal bones. The carpals are individually small and irregular, and the overall size of the carpus is reduced. Ivory epiphyses of the phalanges are also significantly consistent findings in children that seemed to disappear after epiphyseal fusion of the phalanges.

Previous reports by Hall et al. [3, 5] and Nishimura et al. [4] stressed the posterior scalloping and narrow interpedicular distances of the lumbar vertebral bodies. However, our patients showed scalloping of the posterior aspects of the lumbar vertebral bodies in only two of the six pediatric patients. The interpedicular distance narrowing of the lumbar spine was also not apparent in our series. The vertebral bodies showed a normal height to subtle platyspondyly in all patients.

The SEMDJL-leptodactylic type shows some overlapping features with sponastrime dysplasia [4, 5]. The clinical features, including short stature and midface hypoplasia of the two conditions, are virtually similar. The radiological findings also show some confusion with sponastrime dysplasia because of sclerotic metaphyseal striations at the knees in both conditions. However, severe epiphyseal involvement and diagnostic appearances in the hands in SEMDJL-leptodactylic type patients can aid in the distinction of the former from the latter disorder. Radiological finding of the spine is also different; the SEMDJL-leptodactylic type shows only mild platyspondyly in contrast to sponastrime dysplasia that shows more severe platyspondyly with biconcave vertebral end plates [9–11]. The inheritance pattern of SEMDJL-leptodactylic type occurs by autosomal dominant trait or sporadically in contrast to sponastrime dysplasia, which is inherited only as an autosomal recessive disorder.

Another SEMDJL that needs to be distinguished is the autosomal recessive SEMDJL-Beighton type [12–14]. This latter disorder also shows articular hypermobility with dislocation of the hips and elbows rather than the knees. Despite some similarities clinically with the SEMDJL-leptodactylic type and Beighton type, the radiological findings are clearly distinctive. For the Beighton type, there are no metaphyseal streaks. The metacarpals are short and wide rather than having a slender appearance as in the leptodactylic type. The early onset of spinal malalignment with severe kyphoscoliosis is also the major differential feature from the leptodactylic type.

The molecular defect in the SEMDJL-leptodactylic type has not been identified. In the future, accumulation of cases and the discovery of the causative gene will aid in an accurate diagnosis in addition to the distinct clinical and radiographic features.



In conclusion, the SEMDJL-leptodactylic type is a distinctive form of skeletal dysplasia characterized by a short stature, generalized articular hypermobility, and progressive scoliosis. The most important radiographic features of this disorder consist of severe epiphyseal changes with metaphyseal sclerotic streaks, small and irregular femoral capital epiphyses with constricted femoral necks, subluxation or dislocation of the knee joint, and the presence of slender metacarpals/metatarsals without brachydactyly of the phalanges. In view of the clinical and genetic implications, awareness of this rare disorder and distinct clinical and radiological features will help in diagnostic precision since the disorder probably is more common than has been initially reported.

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