# Spondyloepimetaphyseal dysplasias

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**Abstract**
*Spondyloepimetaphyseal dysplasia (SEMD) is a descriptive term of major radiological abnormalities of the spine, epiphyses and metaphyses of a disparate group of disorders with differing phenotypes, modes of inheritance and detailed radiographic abnormalities. Several clearly delineated types are recognised and these are described below, but the term SEMD is often used as a general purely descriptive or generic term when a precise diagnosis is not known and does not constitute a definite diagnostic label. In this latter situation it is not possible to predict the evolution of the changes, the mode of inheritance or the complications, except that in general patients with a predominant epiphyseal component as part of a skeletal dysplasia will develop premature osteoarthritis especially of the weight bearing large joints.*

**Key-words**
skeletal disorder, combined involvement of the epiphyses and metaphyses, defective growth and modelling of the spine and long bones.

## Spondyloepimetaphyseal dysplasia, Strudwick type

***Disease name and synonyms***
Spondyloepimetaphyseal dysplasia (SEMD) Strudwick type,
Spondyloepiphyseal dysplasia (SED) congenita with dappled metaphyses,
Strudwick syndrome.

***Brief description and clinical findings***
SEMD type Strudwick was identified as a specific variant form of SED congenita in 1982 by several authors, (Anderson *et al.,* 1982; Bartsocas *et al.,* 1982) although Spranger and Maroteaux (1982, 1983) questioned whether it should be considered a separate entity. Both conditions certainly represent type II collagenopathies and are inherited in an autosomal dominant manner. The gene is linked to chromosome 12q13. Mutations in the type II collagen gene *COL2A1* involving a mutation in the amino-terminal end of the protein have been demonstrated (Tiller *et al.,* 1995; Kaitilla *et al.,* 1996; Vikkula *et al.,* 1993; Murray *et al.,* 1989; Tysoe *et al.,* 2003).
At birth the patients are noted to have short limbs and a short trunk, with a cleft palate, small chest and protuberant abdomen. Respiratory distress may be present. Later myopia develops and this may progress to retinal detachment. There is mild dysmorphism with a flat face and hypertelorism. Stature is significantly reduced and a waddling gait, genu valgum (or varum) and pronounced lumbar lordosis develop. The

hands and feet are relatively normal. Intelligence and life expectancy are normal, although lung function may be compromised.

***Radiological findings***

In infancy these are identical to SED congenita. There is generalised platyspondyly and anisospondyly with L1 being larger than L5. At this age the vertebral bodies are oval but in childhood they become more pear-shaped with mild posterior constriction and rounded anterior borders. The ribs are short and the thoracic cage small. The long bones are all short with very short or even absent femoral necks. There is absent ossification of the epiphyses at the knee at birth and absent ossification of the pubic rami. The acetabular roofs are horizontal but the shape of the iliac bones is otherwise normal. There is marked delay in ossification of the capital femoral epiphyses and in early childhood a severe coxa vara deformity develops, with high-riding greater trochanters. Typically in infancy there is no metaphyseal irregularity in either SED congenita or in SEMD type Strudwick. Occasionally minor metaphyseal spurring may presage the development of the Strudwick type.

From about the age of four years the typical metaphyseal changes of SEMD type Strudwick develop. These consist of a striking flocculated or dappled fragmentation of the long-bone metaphyses, which are expanded with islands of relative sclerosis. These changes are initially apparent in the proximal femora. Typically the ulna is more severely affected than the radius and the fibula than the tibia. Rarely, deforming pseudarthroses may develop in some of these fragmented metaphyses, giving rise to secondary deformities such as humerus varus or tibia recurvatum. The severe bilateral coxa vara may progress to posterior dislocations. Typically the hands and feet are spared although there is some delay in maturation of the carpal centres on the radial side of the hand. Kyphoscoliosis may develop. Cervical kyphosis and cervical instability may occur as a result of a hypoplastic vertebral body (usually C3) and/or a hypoplastic odontoid peg.

***Histopathology findings***

The findings are the same as those seen in SED congenita. There is disorganisation of the growth plate with clustering of chondrocytes, which contain inclusion bodies. There is fine granular material seen in the rough endoplasmic reticulum on electron microscopy.

***Management***

*Early cleft palate repair.*

Regular orthopaedic review should be undertaken for coxa vara, hip dislocation, lumbar lordosis, kyphoscoliosis, cervical spine instability, genu valgum and premature osteoarthritis. Limb lengthening procedures are not usually recommended in this condition.

Ophthalmological review will evaluate myopia and prevent retinal detachment.

***Genetic advice***

Inheritance is autosomal dominant. Reports of affected siblings appear to represent parental mosaicism rather than an autosomal recessive inheritance (Anderson *et al.,* 1982; Kousseff and Nichols, 1984)

***Differential diagnosis***

Until early childhood the clinical and radiological findings are the same as SED congenita and there remains doubt as to the validity of splitting type Strudwick from SED congenita. Other forms of SEMD should be considered as should some types of spondylometaphyseal dysplasia such as type Jansen and the corner fracture type.

***Prenatal diagnosis***

Prenatal diagnosis can be made if one parent is affected. Otherwise short limbs can be identified on prenatal ultrasound. This is apparent before 20 weeks gestation.

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## Spondyloepimetaphyseal dysplasia with joint laxity

***Disease name and synonyms***
Spondyloepimetaphyseal dysplasia with joint laxity ( SEMD-JL)

***Brief description and clinical findings***
Torrington (1991) identified two Afrikaans speaking women as the progenitors of this condition in South Africa in the seventeenth century. Although the vast majority of patients have been identified in South Africa, other cases have been described in North and South America and Europe.
At birth there is short stature and joint and ligamentous laxity with hip dislocation in about one quarter of patients and dislocation of the radial heads. Kyphoscoliosis is present and is rapidly progressive, in severe cases leading to paraplegia or early death in mid childhood from cor pulmonale. A mobile talipes equino-varus deformity is present. The face is oval with a long philtrum and prominent eyes with variably blue sclerae and hyperelastic, soft skin. Almost half the patients have a cleft or high arched palate. Congenital cardiac anomalies, predominantly septal defects, may be present. Other reported findings include mental retardation, myopia, lens dislocation and Hirschprung disease.

***Radiological findings***
There is a severe and progressive kyphoscoliosis and platyspondyly with biconvex vertebral bodies with irregular endplates. The iliac wings are flared and the sacro-sciatic notches short. In the long-bones epiphyseal ossification is delayed, the metaphyses are wide and irregular and the trabecular pattern is coarse. There is coxa valga with hip dislocation and dislocation of the radial heads. The distal radius and ulna are expanded. Traction exostoses may be present. The tubular bones of the hands and feet are short.

***Genetic advice***
Inheritance is autosomal recessive.

***Differential diagnosis***
Differentiation is required from other SEMDs, diastrophic dysplasia, Larsen syndrome, and the mucopolysaccharidoses.

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# Spondyloepimetaphyseal dysplasia with multiple dislocations (type Hall)

### *Disease name and synonyms*
Spodyloepimetaphyseal dysplasia with múltiple dislocations (MD). Spondyloepimetaphyseal dysplasia with múltiple dislocations type Hall. Spondyloepimetaphyseal dysplasia with múltiple dislocations (leptodactylic type). SEMD-MD

### *Brief description and clinical findings*
Langer *et al.* (1997) initially identified this as a distinct entity and illustrated one case included in a paper on sponastrime dysplasia. He identified a further case in a paper on sponastrime dysplasia by Camera *et al.* (1994). Hall *et al.*, 1998 described three further unrelated cases and used the term SEMD with multiple dislocations to differentiate it from the group known as SEMD with joint laxity. There is an equal gender distribution. Presentation is at birth with marked hypotonia, short stature and some facial dysmorphism with midface hypoplasia and a depressed nasal bridge. None of the patients has had a cleft palate. About one third of affected patients have had significant laryngeal stenosis or tracheomalacia in early childhood, some requiring tracheostomies. Intelligence is normal. There is progressive joint laxity with hip dislocation and genu valgum and dislocations at the knees with weight bearing. A mild scoliosis develops during childhood.

### *Radiological findings*
There is a generalised delay in epiphyseal ossification and when present they are small, flattened and irregular. At the hips dislocation may develop and the femoral necks are narrow, curved and tapered. At the knees there is progressive subluxation through childhood. The epiphyses and patellae are small and irregular and the adjacent metaphyses irregular with some longitudinal sclerotic striations. In the spine there is only very mild platyspondyly with some minor irregularity of the vertebral endplates. The interpedicular distances fail to widen in the normal manner and there is spinal dysraphism of the sacrum. In the thoracic region the vertebral bodies are pear-shaped with a mild posterior constriction. In the adult, the vertebral bodies have a biconcave configuration. Diagnostic features are present in the hands with small, sclerotic, fragmented epiphyses and carpal bones, an overall reduction in the size of the carpus, especially affecting the proximal row, gracile metacarpals and squared distal ends of the middle phalanges.

### *Management*
Management is aimed at maintaining mobility and preventing dislocations. Knee braces have helped the severe joint laxity here.

### *Genetic advice*
Four affected parent/offspring families have been described and inheritance is autosomal dominant.

### *Differential diagnosis*
The major differential diagnosis is sponastrime dysplasia. This is inherited in an autosomal recessive manner. Radiologically there is severe platyspondyly in infancy and early childhood. The characteristic sclerotic metaphyseal striations do not become apparent until mid childhood. Apart from a delay of bone maturation, the modelling of the tubular bones in the hands is normal.

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## Spondyloepimetaphyseal dysplasia, Iraqi type

***Disease name and synonyms***
Spondyloepimetaphyseal dysplasia (SEMD) Iraqi type
SEMD type Sohat

***Brief description and clinical findings***
Sohat *et al.* (1993) reported three affected individuals in a large Iraqi Jewish family and Figuera *et al.* (1994) reported a further case from Mexico. Presentation is at birth. Clinically there is short stature because of limb shortening, a protuberant abdomen and hepatosplenomegaly, lumbar lordosis, a short neck, joint laxity and genu varum deformity. The face is described as being round with thin lips.

***Radiological findings***
The tubular bones are all short with irregular, flared metaphyses and delayed epiphyseal ossification. The short tubular bones in the hands show metaphyseal cupping. In infancy the fibula is disproportionately long and the femoral necks short. Later coxa vara and genu varum develop. In the spine there is platyspondyly with central notches of the superior and inferior vertebral end-plates, possibly representing previous coronal cleft vertebrae. There is some narrowing of the interpedicular distances. In the pelvis the iliac bones are short and wide and the acetabular roofs are horizontal. The thorax is short and mildly narrow. The ribs have pronounced cupping of their anterior ends. Multiple wormian bones in the skull may be an additional finding.

***Genetic advice***
Autosomal recessive inheritance

***Differential diagnosis***
Other forms of SEMD, SED congenita and achondroplasia in infancy. Also consider Dyggve Melchior Clausen disease and metaphyseal chondrodysplasia with pancreatic insufficiency and cyclical neutropenia.

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## Spondyloepimetaphyseal dysplasia, Irapa type

***Brief description and clinical findings***
This condition was first described by Arias *et al.* (1976) in the Irapa Indians of Venezuela and later in a Mexican family. Clinical presentation is about the age of five years with rhizomelic shortening, walking difficulty and joint pains. The joints are enlarged with a reduced range of movement and premature osteoarthritis develops. There is brachydactyly but the index fingers and second toes are relatively long.

***Radiological findings***
In the spine there is generalised platyspondyly with vertebral end-plate irregularity. The tubular bones are short with wide, irregular metaphyses especially of the proximal femora leading to coxa vara and distal humeri, and there is delayed epiphyseal ossification. The carpal bones are small and irregular and carpal fusions may be present. The 3rd-5th metacarpals and metatarsals are short and wide distally. In the pelvis the iliac bones are short, the acetabula dysplastic and the symphysis pubis irregular. The anterior ends of the ribs are expanded with irregular ossification. There are changes of premature osteoarthritis and generalised osteoporosis.

***Genetic advice***
Inheritance is autosomal recessive.

***Differential diagnosis***
Other forms of SEMD.

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## Spondyloepimetaphyseal dysplasia, short limb-abnormal calcification type

***Disease name and synonyms***
Spondyloepimetaphyseal dysplasia (SMED) short limb-abnormal calcification type
SMED short limb-hand type

***Brief description and clinical findings***
This disorder was first described in 1993 by Borochowitz *et al.* and eight further cases by Langer *et al.,* 1993. Presentation is at birth with severe limb shortening, short hands and feet and a relatively long trunk. There is some joint laxity. Kyphoscoliosis subsequently develops leading to a short trunk. The thorax is narrow. There is facial dysmorphism with a relatively large head and a prominent forehead with midface hypoplasia, a broad, depressed nasal bridge, short, upturned nose, hypertelorism and prominent eyes. The philtrum is long and there is micrognathia.
Complications include cervical cord compression from odontoid hypoplasia and ligamentous laxity, with subluxation of C1 and C2; cor pulmonale as a result of a small thorax and progressive kyphoscoliosis and optic atrophy.

***Radiological findings***
In infancy there is premature stippled calcification in the regions of the epiphyses, laryngeal cartilages, tracheal and bronchial cartilage and costochondral junctions. The diagnosis at this stage is often of a form of chondrodysplasia punctata. Later there is advanced ossification of the carpal centres and of the iliac crest apophyses. The long bones are short with pronounced metaphyseal widening and flaring giving a dumbell appearance. The stippled areas and adjacent metaphyses progress to larger flocculated areas interspersed with lucent areas. The tubular bones of the hands and feet are short with triangular distal phalanges. The calcanea are small and stippled. In the thorax the ribs are short with cupped anterior and posterior ends and the clavicles are relatively long. In the spine there is mild, generalised platyspondyly with wide intervertebral spaces. There is poor ossification of the vertebral bodies in the cervical spine and atlanto-axial subluxation may occur. The vertebral bodies may be pear-shaped or rounded with deficient ossification posteriorly and mild anterior tonguing.

***Genetic advice***
Inheritance is autosomal recessive

***Prenatal diagnosis***
Ultrasound can identify the short limbs *in utero*, but the diagnosis can only be confirmed if there have been previously affected sibs or after radiographic evaluation.

***Differential diagnosis***
In infancy the premature stippling requires differentiation from chondrodysplasia punctata. The dumbell appearance of the long bones, narrow thorax and progressive kyphoscoliosis may resemble metatropic dysplasia. Other SEMDs require consideration, particularly SEMD metatropic type and SEMD type Strudwick both of which have an autosomal dominant inheritance. The expanded metaphyses with flocculated ossification and the changes in the spine may be confused with metaphyseal dysplasia type Jansen.

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## Spondylometaepiphyseal dysplasia, X-linked type

***Disease name and synonyms***
Spondylo-meta-epiphyseal dysplasia (SEMD) X-linked type
SEMD cone-shaped epiphyses type

***Brief description and clinical findings***
Camera *et al.* (1993) reported eight males in five generations of one family. Presentation is not until two years of age with short stature. There is radial deviation of the hands due to relatively long ulnae. There are no dysmorphic features.

***Radiological findings***
Radiologically there is progressive platyspondyly with irregular vertebral endplates and anterior tongues of the vertebral bodies. In the lumbar spine the interpedicular distances are narrow. The posterior ends of the ribs are cupped and

the clavicles short. The iliac wings are small. There is coxa valga. The metaphyses are broad and the upper humeral metaphyses broad. There is a generalised delay in epiphyseal ossification and cone-shaped epiphyses are present at the distal radii, knees and ankles. The tubular bones of the hands are short and there are cone-shaped epiphyses of the phalanges.

***Genetic advice***
Inheritance is probably X-linked dominant.

***Differential diagnosis***
Other SEMDs

***References***
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## Sponastrime dysplasia

***Brief description and clinical findings***
Fanconi *et al.* (1983) who described four sisters first described the condition. The term 'sponastrime' is derived from SPOndylar, NAsal anomalies and STRiation of Metaphyses. The dysmorphic features include a depressed nasal bridge, short nose, frontal bossing and a relatively large head. Camera *et al.* (1993) and Verloes *et al.* (1995) described a subgroup with microcephaly and mental retardation. Short stature becomes apparent from birth. A progressive kyphoscoliosis and lumbar lordosis develop. The skeletal changes may be more severe in affected males.

***Radiological findings***
Marked platyspondyly is present at birth with wide intervertebral spaces. There is some mild posterior constriction of the vertebral bodies with rounded or slightly tongued anterior borders. There is an increase in height of the vertebral bodies during early childhood developing a biconcave shape, with the endplate concavity towards the posterior parts of the vertebrae. The proximal femora have a characteristic 'spanner-like' appearance with prominent lesser trochanters and short curved tapered femoral necks. The hips may dislocate. In infancy and early childhood the metaphyses and epiphyses are mildly irregular and from about the age of four years irregular longitudinal metaphyseal sclerotic striations become apparent. These are most pronounced at the knees and wrists.

***Management***
Management is largely concerned with dislocations and joint laxity.

***Genetic advice***
Inheritance is autosomal recessive.

***Differential diagnosis***
Differentiation is required from other forms of SEMD and in particular from SEMD with multiple dislocations. The metaphyseal striations are similar to osteopathia striata but there are no changes in the spine in this condition.

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