Acromesomelic dysplasia (Maroteaux type) associated with craniovertebral junction anomaly: A report of a rare case and review of literature

Jayesh C. Sardhara, Kumar Ashish, Anant Mehotra, Arun K. Srivastava, Kuntal K. Das
Department of Neurosurgery, Sanjay Gandhi Post Graduate Institute of Medical Science, Lucknow, India

ABSTRACT

Acromesomelic dysplasia Maroteaux type (AMDM) is a rare autosomal recessive osteochondrodysplasia. The responsible gene, AMDM gene, in human beings, has been mapped on 9p13-q12 chromosome by homozygous mapping and pathogenic mutation was later identified in natriuretic receptor B (NPR-B) which has been implicated in the regulation of skeletal growth. Till now, around 40 to 50 cases of AMDM have been described in the world literature. Association of the congenital craniovertebral (CV) junction anomaly has not been reported. Here we are presenting a case of AMDM, with CV junction anomaly. A 10-year boy presented with short stature (122 cm) with short distal limbs, symptomatic for thoracic kyphoscoliosis with back pain. On examination there were no neurological deficits. On radiological investigation, he was found to have short and broad phalanges and toes, thoracic kyphoscoliosis, abnormal pelvic ring, mild ventriculomegaly, cervical syringomyelia and tonsillar descent below foramen magnum, hydrocephalus, os odontoideum with Klippel-Feil anomaly. This was diagnosed as AMDM with congenital os odontoideum, Klippel-Feil anomaly with Arnold-Chiari malformation (ACM) type-1. The patient underwent posterior fossa decompression by removal of foramen magnum ring along with C1 arch for ACM type-1. Kyphosis was left for conservative treatment till further observation and required orthopedic correction in his further age. To the best of our knowledge this is a very rare entity of AMDM with congenital CV junction anomaly.

Key words: Acromesomyelia, craniovertebral junction, marotaeux variant, surgery

INTRODUCTION

Acromesomelic dysplasia Maroteaux (AMDM) type is a rare autosomal recessive osteochondrodysplasia belonging to the group of acromesomelic dysplasias (AMDs). The other varieties are Hunter-Thompson and Grebe type. AMDM was first described in 1971 by P. Maroteaux. It is characterized clinically by severe dwarfism with shortening of middle and distal segment of limbs and radiologically by short broad fingers, shortening of long bones with bowed radius, and vertebral abnormalities. The facial appearance and intelligence are normal. Recently, an AMDM gene has been mapped to human chromosome 9p13-q12. The prevalence of AMDM remains unknown. AMDM is listed as a “rare disease” by the Office of Rare Diseases (ORD) of the National Institutes of Health (NIH) [affects less than 200,000 people in the US population]. It is the most common form of the autosomal recessive AMDs. Only 40 to 50 cases have been reported till date.

We are reporting a case of acromesomyelic dwarfism, kyphoscoliosis, Arnold-Chiari malformation (ACM) type-1, and Klippel-feil syndrome (KFS). To the best of authors knowledge this is the first reported case of AMDM associated with syringomyelia, hydrocephalus, and tonsillar descent with ACM type-1 with KFS.

CASE REPORT

A 10-year boy, weighing 30 kg, height 122 cm, presented with short stature and stooping posture with back pain on prolonged sitting or walking without any weakness noticed for the past 3 year. Pain was localized on the upper thoracic region of spine and was radiating to the suboccipital region. There was history of trivial trauma due...
to fall down from height 1-year back. He was the second
cild, born out of a non-consanguineous marriage, by
full-term normal vaginal delivery and the antenatal period
was uneventful. The birth weight was 2.8 kg. Parents
were normal and though his father was of short stature,
family history of dwarfism was absent. Since birth, weight
gain was normal and his mental function and intellectual
development appeared normal. He was doing well in
studies as compared to other children of the same age.

Examination revealed normal higher mental function.
Sensory and motor examination revealed no sensory or
motor deficits. He had broad forehead, with small flat
nose, short neck with low hair line, and normal head
circumference. He had a stooped posture [Figure 1a],
short and broad fingers and toes (toes were slight
curved) with no polydactyly [Figure 1b and c]. There
was no disproportionate shortening of thigh to legs and
arms to forearms. Passive movements of shoulder were
normal, however mild restriction of 20 degree on flexion
at elbow on both sides, and 20 degree restriction on
flexion and extension was noted at wrist joint on both
the sides. There was multiple café–au-lait spot on both
legs approximately 5 × 2 cm on left leg medial aspect and
small two spots on left lower legs [Figure 1d].

On radiograph X-ray, skull and chest x-rays appear normal
[Figure 2d]. X-rays of the upper limbs revealed short and
broad phalanges and metacarpals, arms and forearm
bones normal [Figure 2b]. In lower limbs, the proximal
bones were normal but in foot, metatarsal and toes were
short and broad, great toe metatarsal bone appeared
larger than other corresponding metatarsals [Figure 2c].
Lower thoracic kypho-scoliosis, wedge-shaped thoracic
and lumbar vertebrae (T11, T12, L1) [Figure 2a] and
congenital odontoid process anomaly without atlanto axial
dislocation (Atlanto dental interval on CT scan: 2 mm)
were observed on radiology [Figure 3a]. X-ray pelvis
revealed hypoplastic lilac ring. MRI of the brain revealed
mild hydrocephalus with ventriculomegaly [Figure 3b],
small posterior fossa with crowding of structures, tonsillar
descent below foramen magnum with canal size at
foramen magnum (19 mm)[Figure 3c]. In addition, 3 mm
size dilatation of the central canal of upper cervical cord
noted in MRI [syringomyelia] at the level of C2 and T6
to T8 level [Figure 3d].

There was complete congenital fusion of anterior
cervical vertebral body of 2nd and 3rd cervical vertebrae
along with partial fusion of posterior elements of same
vertebrae [Figure 4]. Genetic evaluation was not possible.

He underwent foramen magnum decompression and
excision of C1 posterior arch and C2 lamina for ACM
type-1. Intraoperative finding included tonsillar descent
till 1st cervical vertebral ring, fibrous adhesions between
dura, arachnoids, and tonsils with occlusion of foramina
of Luschka and Magendie. The tonsils were separated
easily. Post operative course was normal, pain was
reduced significantly with no neurological deficit on
3 month, 6 month, and 1-year follow up. For thoracic
kyphoscoliosis spinal extension brace was applied and
further surgery was planned for kyphoscoliosis after
16 year of age.

**DISCUSSION**

AMD is an extremely rare, \[1\] inherited, progressive
skeletal disorder that results in a particular form of short
stature. The disorder is characterized by acromelia and
mesomelia. \[2\] Over time, the apparent disproportion
becomes even more obvious, especially during the first

![Figure 1](image1.png)

*Figure 1: (a) Acromesomelic dwarfism with thoracic kyphoscoliosis: Bullet-shape thoracic and lumbar vertebrae with kyphosis. (b) Short and broad fingers (c) Short and broad toes (d) Café-au-lait spots over left leg*

![Figure 2](image2.png)

*Figure 2: (a) Thoracic kyphosis with Cobbs angle -35 degree (b) Short and broad phalanges and metacarpal bone (c) Short and broad toes, metacarpal of great toes is larger than other toes. (d) Normal lung field with thoracic scoliosis*
s few years of life. Affected individuals may have additional abnormalities resulting from abnormal cartilage and bone development, including limited extension of the elbows and arms and/or progressive abnormal curvature of the spine. Other characteristic abnormalities include a relatively enlarged head (macrocephaly), slightly flattened midface, and/or small, pug nose. AMD is inherited as an autosomal recessive genetic trait. Genetic counseling is of benefit for affected individuals and their families. As genetic evaluation was not possible in these patients, AMDM was diagnosed on clinical ground.

Treatment may require the coordinated efforts of a team of specialists. Pediatricians, specialists who assess and treat skeletal abnormalities (orthopedists), physical therapists, and/or other health care professionals may need to systematically and comprehensively plan an affected child’s treatment. Abnormal curvature of the spine (i.e. low thoracic kyphosis and/or lumbar hyperlordosis) may be treated with a combination of exercises and physical therapy, other supportive techniques, braces, casts, and/or, in severe cases, corrective surgery.

ACM is a heterogenous developmental disorder of impaired CSF circulation at foramen magnum due to tonsillar herniation which leads to blockage of pathway. Treatment, if indicated, is surgical decompression of posterior fossa. Although there are various treatment for the disease, removal of foramen magnum through a posterior approach with partial or complete cervical posterior arch excision \{laminectomy\} is considered standard in majority of patients. It is usually associated with syringomyelia (30-70%).

Incidence of KFS in this disease remains unknown due to its rarity and the fact that it is frequently asymptomatic.[13] Due to rarity of disease and paucity of literature available for such CV junction anomaly associated with AMDM, management strategy has not yet been established. ACM type-1 was probably correlated with congenital low-lying tentorium and crowding of posterior fossa structure or may be due to hydrocephalus. As pain in this patient may be related due to ACM, further CSF flow obstruction may progress the syringomyelia caudally and hydrocephalus proximally. Foramen magnum decompression was the best possible treatment option to arrest further neurological deficit. As odontoid process anomaly and Klippel-Feil anomaly was incidental finding and atlanto dental interval (ADI) was normal, it was treated conservatively.[13] Kyphoscoliosis treatment remains conservative in this patient as it was planned that it will be corrected surgically if the patient remains symptomatic for long. On follow-up after 2 months, the patient was having complete relief from back pain. Till date 40 to 50 cases have been reported; those all were about AMDM type. To the best of author knowledge till now there has been no case report available in which AMDM is associated with ACM type-1 and Klippel-Feil syndrome.

REFERENCES


How to cite this article: Sardhara JC, Ashish K, Mehotra A, Srivastava AK, Das KK. Acromesomelic dysplasia (Marotaeux type) associated with craniovertebral junction anomaly: A report of a rare case and review of literature. Indian J Neurosurg 2014;3:106‑9.

Source of Support: Nil, Conflict of Interest: None declared.

Author Help: Reference checking facility

The manuscript system (www.journalonweb.com) allows the authors to check and verify the accuracy and style of references. The tool checks the references with PubMed as per a predefined style. Authors are encouraged to use this facility, before submitting articles to the journal.

- The style as well as bibliographic elements should be 100% accurate, to help get the references verified from the system. Even a single spelling error or addition of issue number/month of publication will lead to an error when verifying the reference.
- Example of a correct style
- Only the references from journals indexed in PubMed will be checked.
- Enter each reference in new line, without a serial number.
- Add up to a maximum of 15 references at a time.
- If the reference is correct for its bibliographic elements and punctuations, it will be shown as CORRECT and a link to the correct article in PubMed will be given.
- If any of the bibliographic elements are missing, incorrect or extra (such as issue number), it will be shown as INCORRECT and link to possible articles in PubMed will be given.