

Cleidocranial dysplasia: a case report

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This article discusses the case of a 55-year-old man suffering from mild neck pain and chronic sinusitis. In addition the patient presented with clinical and radiological findings consistent with a congenital disorder of cleidocranial dysplasia. Diagnostic imaging and clinical manifestations are discussed.
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KEY WORDS: cleidocranial dysplasia, dysostosis, sinusitis.

Le présent article examine le cas d'un homme de 55 ans qui souffre d'une légère douleur cervicale et d'une sinusite chronique. De plus, le patient s'est présenté avec des résultats cliniques et radiologiques qui correspondent à une maladie congénitale, la dysostose cléïdo-crânienne. L'auteur traite de la visualisation diagnostique et des signes cliniques.
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MOTS CLÉS : dysostose cléïdo-crânienne, dysplasie, sinusite.

Case report

A 55-year-old male presented with a chief complaint of neck pain and stiffness. The pain extended from the inferior aspect of the left side of the occiput to the mid-scapular level. The pain was described as dull, tender to touch, and present for 3 weeks; it was insidious in onset with no history of recent trauma. No paraesthesia or weakness was reported.

Symptoms were aggravated by various sleeping postures, but were most severe in the right lateral decubitus position; relief was obtained with topical application of tiger balm. Secondary complaints included pain at the bridge of the nose, radiating across the forehead, also exacerbated by certain sleeping positions and neck flexion. The patient reports often awakening with a "stuffed" nose. He reports no previous treatment for his symptoms.

The patient's lifestyle included a history of smoking 2 packs/day and heavy alcohol consumption for 25 years,

but at present does neither. A sedentary lifestyle was reported with little exercise.

Previous injuries include a motor vehicle accident 10 years previous with resulting neck pain for 3 days with no other residual sequelae.

Previous illnesses include 30 throat infections over the last 5 years, for which he has received a regimen of Penicillin 4-5 times per year. The patient reports numerous dental infections.

The patient has known cleidocranial dysplasia, but is unaware of any other family members with this disorder. Bilateral hip surgery was performed at age 11 to correct congenital hip malformations. He also has 60% deafness in his right ear.

Examination revealed a 5'6" tall, 61.5 Kg healthy appearing male with unremarkable vital signs. The patient presents with a small face and enlarged frontal and parietal regions of the cranium. Hypertelorism is present with

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a depressed nasal bridge. The patient was able to approximate his shoulders anteriorly due to the absence of clavicles. Mild soft-tissue swelling was noticed in the left submandibular region.

Active cervical range of motion was reduced 40% in flexion and 25% in lateral flexion bilaterally. Cervical compression tests including bilateral cervical extension-rotation reproduced the neck pain. Lateral Maigne's challenge and facet rub tests were all provocative in the middle cervical spine. Motion palpation revealed joint restrictions in the upper cervical spine. Muscle examination revealed hypertonic suboccipitals, scalenes, trapezii, and rhomboids. Resisted muscle testing on extension increased the patient's posterior neck pain.

Neurological examination confirmed conduction-hearing loss of the right ear. The patient was otherwise neurologically intact.

Radiological examination of the cervical spine revealed moderate degenerative changes with abnormal kinematics in flexion of the mid cervical spine. The cranium showed multiple wormian bones, basilar invagination, delayed



Figure 1A Skull radiograph demonstrates multiple wormian bones in the occipital bone, delayed eruption of adult teeth and basilar invagination.

eruption of adult teeth, and spina bifida occulta of T3. Furthermore, clavicles were absent bilaterally (Figure 1A and 1B).

The patient was diagnosed with facet irritation of the mid-cervical spine with associated myofascial strain of the posterior cervical paraspinal musculature secondary to posture. In addition the clinical and radiological findings are consistent with a congenital disorder of cleidocranial dysplasia. His reported nasal complaint is most likely secondary to chronic sinusitis, a complication of cleidocranial dysplasia.

The patient was started on a course of gentle cervical and upper thoracic mobilization and cervical spinal manipulative therapy. Home stretching program was given to the patient. The patient reported symptomatic relief with treatment.

Discussion

Cleidocranial dysplasia (CCD) is an autosomal dominant disorder with complete penetrance and variable expression.¹ However, an autosomal recessive inheritance has



Figure 1B Note the absence of clavicles and spina bifida occulta of T3.

been reported.^{2,3} It is a relatively uncommon disorder with the prevalence being 0.5 per 100,000 live births.⁴ One third of cases are considered to be due to sporadic mutations.²

Marie and Sainton coined the term cleidocranial dysostosis in 1898, although descriptions of the disorder can be traced back to the 1760s.^{2,4} The term dysostosis means defective ossification or defect in the normal ossification of fetal cartilages (Gr. *osteon* – bone).³ In dysostoses the distribution follows a defect in ectodermal or mesenchymal tissues. Rarely are all bones involved. As derived from the Greek, dysplasia refers to abnormality of development or “ill formed” (Gr. *plassein* – to form); in pathology it means alteration in size, shape, and organization of adult cells. Bone dysplasias are those in which the predominant error is an intrinsic one; it affects all growing bones similarly and is generalized in distribution.³ At present, this congenital condition is known to be a generalized disorder of bone, and as such was renamed in 1978 cleidocranial dysplasia.⁵ Cleidocranial dysplasia although congenital may show a wide variation in somatic presentation. The main defect lies in faulty ossification of selected endochondral and intramembranous bones, and although any bone may be affected, a classical triad of cranial, clavicular, and pelvic anomalies comprise the most striking changes. This is an early developmental disorder of mesenchyme or connective tissue, producing retarded ossification of the membranous and cartilaginous precursors of bone, especially at the junction of various bones. This may lead to delayed or even failure of ossification of portions of the skeletal structure. The syndesmoses between cranial bones and symphyses of other bones are basically connective tissue junctions. The medial and lateral centers of ossification of the clavicle are also separated by a fibrocellular structure.³ The distribution follows a pattern in which syndesmoses or suture lines of connective tissue and bones in which cartilage nuclei grow by direct apposition of a fibrocellular layer into proliferative cartilage.³ The classical aspects of the disorder consist of generalized midline defects such as patent anterior fontanelle, persistent metopic suture, nasal bone absence, non-union of the mandibular symphysis, bipartite hyoid bone, spina bifida occulta and delayed closure of the symphysis pubis.⁵

Clinically, the diagnosis is often made at birth due to the soft cranium, but may not occur until later, when per-

sistence of the widely open anterior fontanelle and sutures (representing incomplete ossification of cranial bones) or short stature incite parental concern.⁶ Individuals with this disorder present with some or all of very characteristic features. These include a large brachycephalic head, small and angular face, prominent frontal and parietal bones and drooping shoulders with excessive mobility.^{4,6,7} Depending on the amount of clavicular involvement, the patient may be able to approximate the two acromial regions below the chin.⁴ Height is reduced in both sexes, but dwarfism is not a common finding. In a longitudinal study by Jenson (1990), height discrepancy was found to be more significantly decreased in females.⁵ The chest may be narrowed or funnel-shaped leading to potential respiratory distress in childhood.^{2,4,7} Because of delayed mineralization, there may be abnormal dentition with late eruption and impaction of the deciduous and permanent teeth. The teeth may appear small, irregularly spaced, and crowded. Furthermore, the teeth often display aplasia, malformed roots, enamel hypoplasia, enhanced caries, and supernumerary teeth.^{4,7,8} However, extraction of the deciduous teeth does not stimulate eruption of the permanent teeth.⁶ Eruption often continues into the second decade.⁶ A 26-year-old female was reported to have 63 unerupted teeth.⁶ Other facial features include a high forehead, broad base of the nose, depressed nasal bridge, narrow high-arched palate, absent paranasal sinuses and hypertelorism.² Hearing loss is due to structural abnormalities of the ossicles leading to a middle ear conduction problem. This is often associated with dense sclerosis of the petrous and mastoid portions of the temporal bone.^{4,6,9} Limb abnormalities manifest as genu valgum and short, stubby fingers with rounded deep-set nails.^{2,6} Gait abnormalities are due to hip and femoral deformities, which may require surgically intervention later in life. The patient's cognitive abilities and laboratory values are normal.

The radiologic features of this disorder are very characteristic. With respect to the skull, it is the membranous portion and not the base that is affected. As such, delayed ossification leads to delayed closure of sutures (sagittal and coronal) and fontanelle (metopic) (Figure 2A).⁷ With time, numerous Wormian bones form within the sutures (Figure 2B). Wormian bones are named after Oluff Worm (1588–1654), professor of anatomy at the University of Copenhagen.¹⁰ Marked brachycephaly, platybasia and increase in the basilar angle may be present.^{4,7} Although

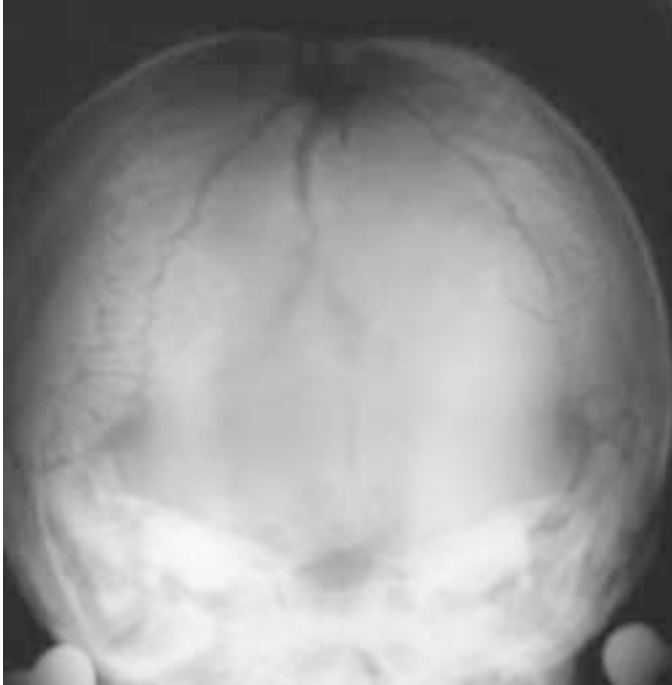


Figure 2A Sutural Widening. The anterior fontanel and the metopic suture remain wide and open. (Film courtesy of American College of Radiology, Reston, Virginia.)



Figure 2B Wormian Bones. Note the multiple intrasutural bones throughout the occipital region. These are common in cleidocranial dysplasia but not pathognomonic. (Film courtesy of American College of Radiology, Reston, Virginia.)



Figure 2C Teeth. Note the late eruption and impaction of permanent teeth. (Film courtesy of American College of Radiology, Reston, Virginia.)

parietal bone ossification may be absent at birth calvarial thickening frequently occurs in adults in the supra-orbital region, temporal bone, and occipital bone producing prominent ridges.^{11,12} Dental anomalies as mentioned are common. It is interesting to note that most of the permanent teeth fail to erupt or do so one at a time throughout adulthood (Figure 2C).¹¹ There is hypoplasia of the maxilla and zygomas which leads clinically to a small face.² In contrast, the mandible is large leading to prognathia and a late closing mandibular suture.⁴ Often the paranasal sinuses are hypoplastic or absent as well as failure of nasal bones to ossify accounting for the depression in the bridge of the nose.^{4,8} Rarely, the hyoid bone may be cleft.

With respect to the thorax a common finding is some form of abnormality to the clavicle. As the clavicle forms from three separate ossification centers, one or more of

these centers may be affected (Figure 3A, B). Complete absence is found in only 10% of cases. More commonly, small fragments are present medially, or sternal and acromial ends are present with the middle aspect being absent.⁷ When the middle portion is absent a pseudoarthrosis may develop with the sternal end positioned more anterior and superior to the acromial portion at the pseudoarthrosis (Figure 3C).¹¹ Although involvement of the clavicles is a

typical feature, there are reports of patients with normal clavicles.² The scapulae may be small, winged, or elevated.⁴ The chest is narrow and cone-shaped which appears to result from hypoplasia or to delayed development of a segment of the posterior part of the vertebral body, the posterior wedging remaining in many adults as residual deformity.¹¹ The ribs are often normal, but on occasion may be short and obliquely oriented.⁸ Rarely, the sternum



Figure 3A Clavicle. Agenesis of the lateral portions of the clavicles. (Film courtesy of American College of Radiology, Reston, Virginia.)



Figure 3B Clavicle. Agenesis of the medial and middle portions of the clavicles with hypoplastic lateral portions. (Film courtesy of American College of Radiology, Reston, Virginia.)



Figure 3C Clavicle. Agenesis of the middle ossification centre of the clavicle, with a pseudoarthrosis. (Film courtesy of American College of Radiology, Reston, Virginia.)



Figure 4 PUBIC SYMPHYSIS. Incomplete ossification of the pubic bones has led to a midline diastasis of the pubic symphysis. (Film courtesy of American College of Radiology, Reston, Virginia.)

may be cleft.

Ossification of the pelvic bones are delayed. As a result hypoplasia of the iliac wings results in a loss of the normal ilial flaring, which produces an almost vertical lateral margin.¹¹ Furthermore, the pubic and anterior ischial rami are underdeveloped forming a small pelvic bowl.^{4,7} Commonly there is failure of the rami to approximate anteriorly leading to a diastasis (Figure 4). A distinctive lateral notching develops in the proximal femoral ossification center.¹¹ Although development of coxa valga is more common, unilateral or bilateral coxa varus can develop, possibly secondary to weight-bearing stresses on the poorly mineralized femoral neck leading to gait disturbances.^{4,12}

Spinal changes include spina bifida occulta in the cervical and upper thoracic regions (Figure 5).^{6,12} The vertebrae may show delayed mineralization and retain their infantile shape with convex upper and lower endplates into childhood.⁶ Faulty development of the ossification centers for the embryonic vertebral bodies and vertebral



Figure 5 SPINA BIFIDA OCCULTA. Note the multiple midline defects in the cervical and upper thoracic spine. (Film courtesy of American College of Radiology, Reston, Virginia.)

arches results in neural arch defects, hemivertebrae and posterior wedging of the thoracic vertebrae.¹¹ Furthermore, there is an increased association with spondylolysis and spondylolisthesis in the lumbar spine.^{4,11}

With regards to the extremities the most marked changes are located in the hands. Here multiple squared-off supernumerary pseudoepiphyses of the metacarpals (second through to the fifth) especially of the second metacarpal, and with growth from both ends, leads to an elongated digit (Figure 6). In addition, the distal phalanges are hypoplastic and pointed.^{4,7,11} The feet show similar changes although less common. The long tubular bones are affected less often but, may be thin and gracile.^{4,7} Furthermore, the radius may be found to be short with an abnormal wrist articulation.^{4,5} In one longitudinal

study of 17 patients with cleidocranial dysplasia most bones in the hand were reduced in size. Although the middle phalanges showed the greatest variation from normative values, an overall proximal to distal reduction in size was found, which is consistent with other studies.⁵

This disorder although benign has complications associated with it. These include dental caries and delayed eruption of permanent teeth which may lead to osteomyelitis of the mandible or maxilla, hearing loss, cesarean delivery in the pregnant female due to a narrow pelvis, dislocations of the shoulders and hips, scoliosis, respiratory distress in early infancy due to a narrowed thorax, and crowding of the brain stem and upper cervical cord due to platybasia and basilar invagination.^{4,8} However, even with these potential complications the life span of an individual with cleidocranial dysplasia is normal. Treatment is often directed at orthopedic and dental correction. These include correcting multiple dislocated shoulders, radial head, or hips, and correction of malocclusion or impacted teeth.⁷

Conclusion

Cleidocranial dysplasia is an uncommon disorder, however, its clinical and radiological manifestations are characteristic. Although this disorder is benign, it must be



Figure 6 HANDS. Note the accessory epiphysis for the base of the second metacarpal. In addition the distal phalanges are hypoplastic and flattened. (Film courtesy of American College of Radiology, Reston, Virginia.)

remembered that it carries with it several medicolegal implications in terms of complications. First, abnormal dentition may result in dental caries. These, if left untreated, may progress to osteomyelitis of the maxilla or mandible. Therefore, a patient complaining of facial pain, should have the condition of their teeth investigated. Second, although unusual, platybasia and basilar impression may lead to crowding of the brain stem and upper cervical spinal cord. This would be an absolute contraindication for cervical manipulation.

Many of the spinal changes related to this disorder lead to abnormal postural changes that may be addressed under chiropractic care. Medical treatment is mainly directed at orthopedic and dental correction. Management must be tailored specifically to the patients needs.

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