

## INTRODUCTION

Cleidocranial dysplasia is rare autosomal dominant skeletal disorder. This disorder is characterized by a multiple skeletal and dental anomalies and deformities.<sup>1</sup> This disorder is caused by a haploinsufficiency of the runt related transcription 2 gene (RUNX2). This gene is used to control osteoblast and chondrocyte formation and maturation.<sup>2</sup>

## MANIFESTATIONS

Some of most common clinical manifestations include shortened or absent clavicles (See image 1), presence of extra teeth, wormian bones, and frontal bossing of the forehead. These symptoms occur in approximately 80-99% of cases.<sup>3</sup>

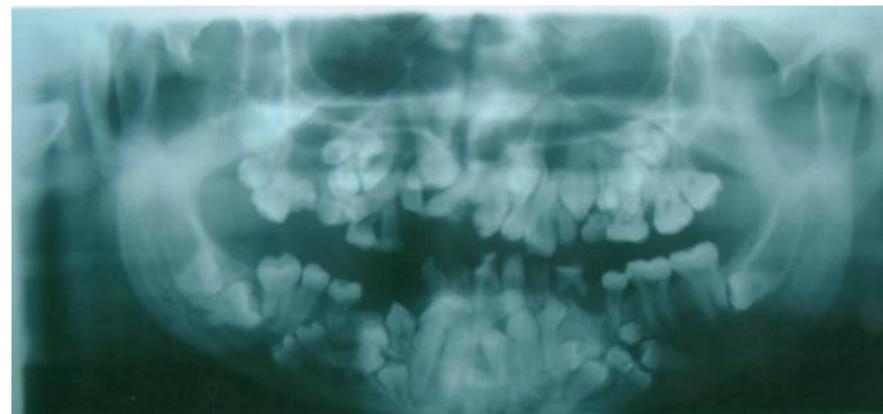
Hyperdontia, or excessive number of teeth is also a common manifestation in patients with cleidocranial dysplasia. (See image 2) The excess teeth can cause overcrowding and may be misshapen due to abnormal dental enamel.<sup>3</sup>

Many patients diagnosed with cleidocranial dysplasia have a shorter stature. On average, male patients are approximately six inches shorter than their unaffected counterparts and females are approximately three inches shorter.

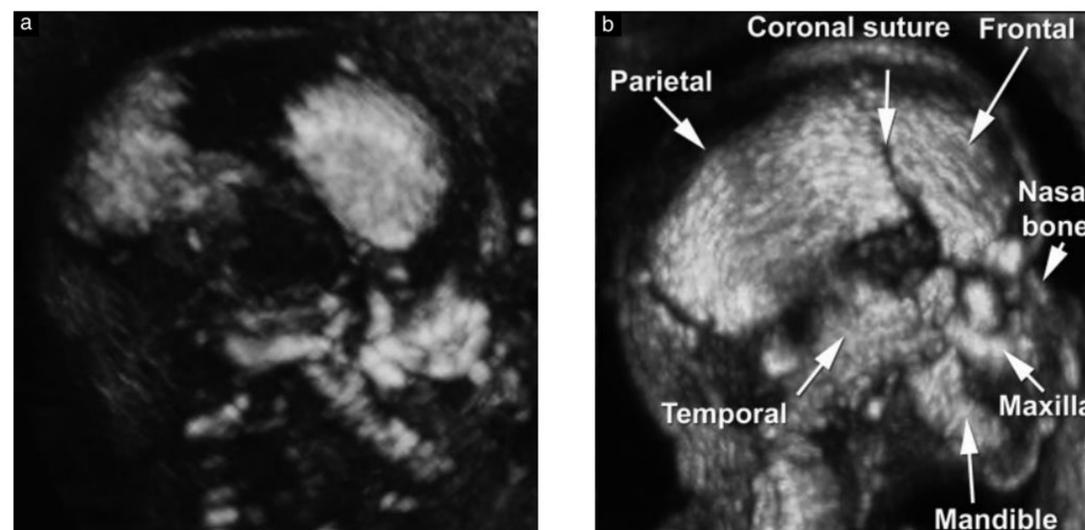
Other less common manifestations can include but are not limited to, hearing impairments, osteoporosis, bell-shaped thorax, pes planus (flat feet), genu valgum (knock knee deformity), and scoliosis.<sup>4</sup>



**Image #1:** Absent right clavicle and hypoplastic left clavicle.<sup>5</sup>



**Image #2:** Presence of impacted and supernumerary teeth.<sup>6</sup>



**Image #3:** a) widened sutures in fetus with CCD at 18+3 weeks gestation. b) normal view of a fetal sutures at 18+3 weeks gestation.<sup>7</sup>

## DIAGNOSIS

There are many ways to diagnose cleidocranial dysplasia. Radiographs are used to diagnose by using a full skeletal study. The chest radiographs will show the shape of the thorax as well as the length of the clavicles. Projections of the skull will show any supernumerary teeth as well as any underdeveloped sinuses.<sup>4</sup>

Prenatal diagnosis is helpful in confirming a diagnosis in cases with a family history. Ultrasonography is used to show ossification of the cranial bones, wide sutures (See image 3), length of the long bones, and brachycephaly of the developing fetus.<sup>7</sup>

## TREATMENT

While there is no cure for cleidocranial dysplasia, patients have a normal life expectancy and most treatments are for functional or esthetic reasons.

Some patients will choose to have cosmetic surgery to correct frontal bossing. Orthodontic surgeries and prosthetic implants may be used to improve cosmetic look and functionality of teeth and to remove the supernumerary and impacted teeth.<sup>4</sup>

## REFERENCES

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