

## *Condrodisplasia Punctata and Foramen Magnum Stenosis in an Infant*

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### **Introduction**

Condrodisplasia punctata is a peroxisomal disease characterized with punctate calcifications in epiphyseal cartilage, symmetrical shrinking in long bones (in rhizomelic type), typical dysmorphic face, limitation of movement in joints, bilateral cataract, seizures, severe respiratory problems, egzema, physicomotor retardation and severe failure to thrive. These cases are uncommon and can be diagnosed with clinical and laboratory findings [1, 2]. Patients are presented with many neurological complications such as facial paralysis, flaccid paraparesis and spastic quadriplegia [2]. However, only one case is reported so far in literature with foramen magnum stenosis.

### **Case Report**

A two months old male infant was admitted to our hospital with a complaint of cyanosis and wheezing. The baby was known to have polyhydramnios in pregnancy during prenatal follow up. He was born with cesarean section at term, weighing 3240 gram and had no neonatal problem. His mother was 29 and father was 30 years old and there was no blood relation between them. He had a 6 years old healthy sister. There was no one with similar findings and diagnosis of chondrodysplasia among family members.

He had history of cough and cyanosis started ten days ago and these complaints were not related with feeding. The baby didn't have any problem until 50th day of birth. The patient was referred to our hospital with preliminary diagnosis of aspiration pneumonia and hypotony. At physical examination the patient's front and rear fontanelle was closed, there was prominent frontal and facial dysmorphism and low-set ears was present. Eye

examination of the patient showed existing bilateral cataract. On physical examination respiratory distress, tachipnea, severe hypotonicity and fasciculation of the tongue were detected and deep tendon reflexes could not be taken bilaterally at inferior extremities. Cardiac examination was normal. Laboratory findings, hemogram, biochemical markers, thyroid function tests, and TORCH (Toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus) serology were normal. Metabolic diagnostics revealed normal levels of ammonia, lactic acid, pyruvic acid and Very Long-Chain Fatty Acids (VLCFA). SMA gene analysis and mitochondrial tests were also normal. Patient underwent cardiac evaluation for an existing cardiac abnormality and echocardiography was performed. Cardiac findings were normal. Cytogenetic study revealed normal karyotype (46 XY). Dispersed located punctuate density increases were observed in the patient's direct graphies in cervical, thoracal, dorsal and sacral vertebrae at both transverse spinosis processes. By clinic condition, calcifications in the lateral servical spine at direct graphies (Figure 1), physical examination and laboratory test results, we diagnosed the patient as condrodisplasia punctata.

**Figure 1:** Radiograph demonstrates stippled calcifications in the lateral cervical spine



When he takes back his head, respiratory arrests were observed repeatedly so we performed servical MRI and detected that; the medulla spinalis made kink partially to the anterior from the inferior part and caused high stenosis in C1 level under foramen magnum level (Figure 2) , this severe narrowing situation caused secondary pressure on medulla spinalis, in C1 and C2 level serious spinal stenosis, probably secondary to edematous change distal to the stenosis led to relative increasing of medulla spinalis size and therefore respiratory arrests are observed every time when he takes his head backwards.

Despite the partial decrease of hypotonisity and improvement of lung infection at patient's follow-up, frequent need for intubation, severe breathing difficulties and recurring respiratory arrests required treatment with tracheostomy. The patient was discharged with tracheostomy and followed by home ventilation.

**Figure 2:** Foramen magnum stenosis and secondary cord compression in C1 nominal sagittal servical MRI profile



## Discussion

Condro Displasia Punctata (CDP) is a rare peroxizomal disease which has multiple epiphyseal displasia form [1, 2]. Chondrodysplasia punctata is a rarely occurring skeletal dysplasia characterized by stippled, punctuate calcifications around joints and within cartilages. Calcifications are most often located in the epiphyses of long bones and soft tissues around joints and vertebral column [3]. There are three different CDP syndrome such as genetical heterogene [4]; autosomal recessive (rhizomelic chondroplasia punctata), x linked recessive

(chondrodysplasia X1) and x linked dominant (chondrodysplasia 2 or conradi hunerman type). Except for these, disease also presents brachitephalangic, tibial metacarpal, humeral metacarpal and other many non-typical forms [5, 6]. Rizomelic chondrodysplasia punctata is related to cataract, hypertelorisma, optic atrophia, shortness of proximal extremities, physicomotor retardation, growth retardation and spasticity. Other characteristics in all variants are abnormalities in face; like cataract, seadle nose and frontal bossing [3]. While the patients are dying because of respiratory distress in infant or newborn stage in otozomal recessive rhisomelic form, the other types live longer and mental retardation does not occur [4]. Many cases like warfarin and fenitoin embriopaties may imitate CDP in epiphises and may cause punctate calcifications [7]. Normal caryotypes found in case presented in this publication allowed for exclusion of trisomy 18 and 21 as well as Turner syndrome as a cause of CDP. Based on medical history, we also excluded autoimmuneological diseases (systemic lupus erythematosus) in children's mothers, vitamin K deficiency and warfarin use. Zellweger syndrome was excluded based on normal VLCFA levels and magnetic resonance brain imaging. Molecular analysis of patient DNA cannot be performed.

Many neurological complications are described such as facial paralysis, flaxcid paraparesy, spastic paraparesy and spastic quadriplegy that occur due to sketelon displasia [8]. In these patients radiological symptoms might be spinal channel stenosis secondary to kyphosis, atlanto-axiel sublucsatation or fatal atlantoaxiel dislocation [9]. We detected cord pressure caused by foramen magnum stenosis in cervical MRI of the case because of radiography findings. In literature Yalin et al. demonstrated a case that has rizomelic CDP with foramen magnum stenosis in a term newborn [10].

Foramen magnum stenosis may lead to bilateral tetraplegia and this situation can be thought as hypotonia just like our patient and may cause death by respiratory arrests. In our patient, symptoms has got non-typical presentation such as hypotonia and aspiration pneumonia. We suggest that the diagnosis of foramen magnum stenosis and atlantoaxial these kinds of patients because it is uncommon and may cause death by respiratory arrest.

## References

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