ECRONICON OPEN ACCESS

EC DENTAL SCIENCE
Case Report

# Dental Management of COACH Syndrome

**Huda Al Rakaf* and Randa Al Saud**

*Dental Department, Prince Sultan Military Medical City, Riyadh, Saudi Arabia*

***Corresponding Author**: Huda Al Rakaf, Dental Department, Prince Sultan Military Medical City, Riyadh, Saudi Arabia.

**Received:** August 14, 2016; **Published:** August 24, 2016

## Objective

This Case report will present the orodigital facial manifestation of JS and related disorders and present the experience in providing dental care.

## Introduction

### Coach syndrome

This article reported two cases of Saudi pediatric male and female patients with growth retardation. Parents are first cousins supporting an autosomal recessive mode of inheritance. Male and female are equally affected and occurs in 1 to 100,000 live births.

The literature review reveals COACH Syndrome:
**C**erebellar venis defect (Joubert Syndrome) by Dr. Joubert 1969
**O**ligophrenia- mental retardation
**A**taxia- lack of muscle control or hypotonia
**C**oloboma- malformation of the retina or ither parts of the eye
**H**epatic fibrosis

Characterized as a rare brain malformation by the absence or underdevelopment of the cerebellar vermis - an area of the brain that controls balance and coordination as well as a malformed brain stem in which the obligatory hallmark is the molar tooth sign (MTS) through magnetic imaging.

## Case 1

A 5-year-old male presented to the emergency clinic complaining of continuous biting on lip and tongue causing deep ulcer on them, difficulty in feeding and sleeping.

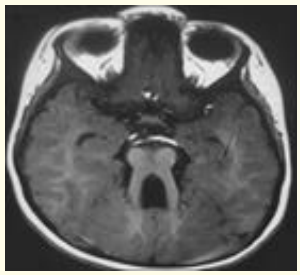
Past history revealed that the child was born by normal vaginal delivery in a hospital of full term pregnancy. Patient was diagnosed by antenatal ultra sound finding occipital frontal circumference larger than normal ventriculomegaly.

A child with the classic features of JS include ataxia, hyperpnea (abnormal breathing pattern), sleep apnea, abnormal eye and tongue movements, 4 arched eyebrows and eyelid ptosis with- spaced eyes.

On examination, patient's weight is 15 kg and .75 m height. All deciduous teeth are normal and free of caries. Deep ulcer on lip, tongue with bus exudate, continuous biting on tongue and lip. Patient was referred to neuro consultation and IV fluid replacement. Blood examination showed evidence of dehydration as shown in Table 1.

## Treatment

Decision was taken by group consultation for extraction of the anterior teeth under local anesthesia and supported by nitrous oxide. Two weeks later, tongue and lip healed with scar. Patient developed good sleeping habits, had stopped lip and tongue biting, and improved psychologically.



## Case 2

Female 13-years-old came with brother with no complain. Diagnosed with JS since age 6 month. With growth and mental retardation, ataxia, feeding, breathing patterns is less severe than her brother.

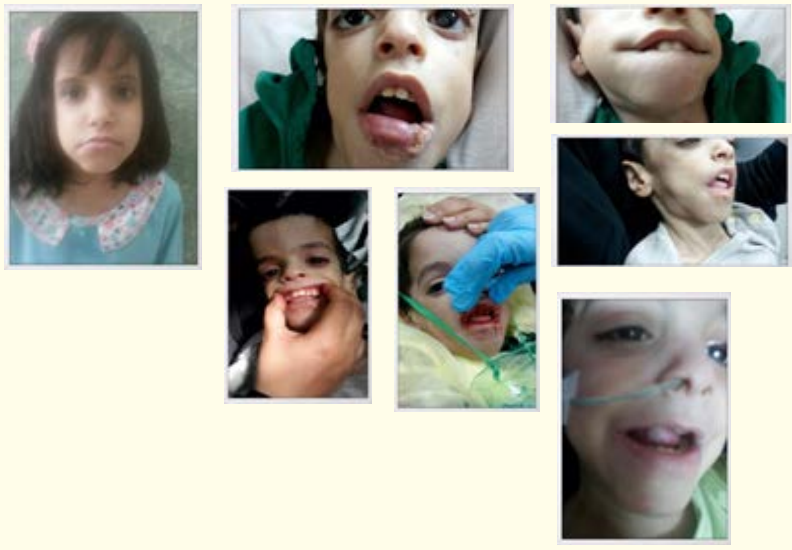
On examination, blood examination was normal, free of caries, with normal lip; tongue free of ulcer, no convulsion was noted.

**Table 1. Blood examination results of the patients**

| BLOOD EXAMINATION | PATIENT 1 | RANGE |
|---|---|---|
| WBC | 14.0 | 5.0 – 14.5 |
| RBC | 3.15 L | 3.9 – 5.30 |
| HGB | 8.8 L | 11.5 – 15.5 |
| HCT | 0.285 L | 0.360 – 0.440 |
| MCV | 91 H | 73 – 89 |
| MCH | 27.9 | 24.0 – 30.0 |
| MCHC | 30.9 L | 31.0 – 35.0 |
| RDW | 16.4 H | 11.0 – 14.0 |
| PLT | 431 | 150 - 450 |
| MPV | 9.2 | 7.8 – 11.0 |
| LYMPH | 4.2 | 2.2 – 9.8 |
| MONO | 1.8 H | 0.1 – 1.0 |
| ALANINE TRANSAMINASE | 47 H | 2 – 40 |
| ALKALINE PHOSPHATASE | 80 | 0 – 269 |
| SODIUM | 141 | 135 – 145 |
| POTASSIUM | 4.1 | 3.2 – 5.0 |
| UREA | 17.6 H | 2.3 – 7.5 |
| CREATININE | 58 H | 23 – 37 |
| TOTAL BILIRUBIN | 4 | 2 – 22 |
| ALBUMIN | 44 | 38 – 51 |
| CALCIUM | 2.39 | 2.12 – 2.57 |
| CORRECTED CALCIUM | 2.29 | 2.12 – 2.57 |

## Prognosis

The prognosis for individuals with JS varies. Some patients have a mild form with minimal motor disability and good mental development, while other may have severe motor disability and moderate mental developmental delays.



## Conclusion

It was concluded that early dental treatment include symptomatic and supportive for patient diagnosed with JS. Education for parent is necessary to avoid possible future complication such as eating or feeding difficulties with dehydration.

Infant stimulation and physical occupational speech and hearing therapy may benefit for some patient.

## Bibliography

1. National Institute of Neurological Disorders and Stroke.
2. Kumandas S., *et al.* "Joubert Syndrome: Review and report of seven new cases". *European Journal of Neurology* 11.8 (2004): 505-510.
3. Joubert M., *et al.* "Familial agenesis of the cerebellar vermis. A syndrome of episodic hyperpnea, abnormal eye movements, ataxia, and retardation". *Neurology* 19.9 (1969): 813-825.
4. Pugash D., *et al.* "Sonographic 'molar tooth' sign in the diagnosis of Joubert syndrome". *Ultrasound in Obstetrics and Gynecology* 38 (2011): 598-602.
5. Brancati F., *et al.* "Joubert Syndrome and related disorders". *Orphanet Journal of Rare Diseases* 5 (2010): 20.
6. Joubert M., *et al.* "Familial dysgenesis of the vermis: A syndrome of hyperventilation, abnormal eye movements and retardation". *Neurology* 18.3 (1968): 302-303.
7. Maria BL., *et al.* "Clinical features and revised diagnostic criteria in Joubert syndrome". *Journal of Child Neurology* 14.9 (1999): 583-590.
8. American Brain Foundation.

**Volume 4 Issue 6 August 2016**