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# A rare case report of Joubert syndrome related disorder: Overlap of OFD type Ii & Type Vi

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**Abstract**
Joubert syndrome and related disorders (JSRD) are a clinically and genetically heterogenous group of disorders characterized by hypoplasia of the cerebellar vermis and developmental delay. The hallmark for diagnosis is molar tooth sign on MRI. Here we present a rare case of Joubert syndrome with Orofacial digital defects, an overlap of type II and type VI.

**Keywords:** Joubert, syndrome, overlap, orofacial

## 1. Introduction
Joubert syndrome is an autosomal recessive disorder characterized by agenesis or hypoplasia of the cerebellar vermis. Signs and symptoms include Hypotonia, Nystagmus, ataxia, distinctive facial features and intellectual disability [1, 2, 3]. Various other abnormalities may also be present. Affected children may have episodes of Hyperpnea which tend to occur shortly after birth. This may intensify with emotional stress but progressively improves with age and usually disappears around 6 months of age [3, 4]. The prevalence is approximately 1 / 1, 00, 000. Only about 200 cases have been reported worldwide [5].

## 2. Case report
A 14-year-old girl born of a second-degree consanguineous marriage, first in birth order with insignificant birth and family history with history of developmental delay presented with generalised tonic clonic seizures.
On examination, child had global developmental delay with DQ = 40 %, broad forehead, high arched eyebrows, broad nasal bridge, right eye rotatory Nystagmus with dissociated vertical deviation and right Esophoria, nodules over the tongue, Mesoaxial Polydactyly in both hands and feet. There was also Hypotonia, Nystagmus, ataxia, past pointing and intentional tremors. Child had an Ejection systolic murmur. Respiratory & gastrointestinal systems were normal.
MRI brain showed cerebellar vermis hypoplasia, MOLAR TOOTH SIGN and BAT WING Appearance of the fourth ventricle. 2D ECHO done showed mild thickening of aortic Valve. Ultrasound abdomen showed solitary renal cyst. X-ray of hands and feet showed Y shaped metacarpals and Mesoaxial Polydactyly. Haemogram and renal function tests were normal.

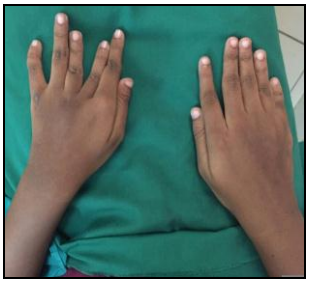

**Fig 1:** Showing Polydactyly

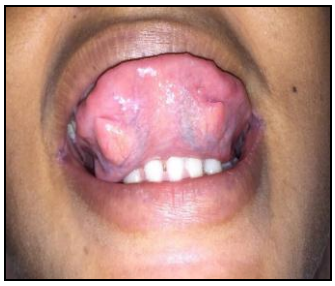

**Fig 2:** Tongue showing hamartomatous nodules

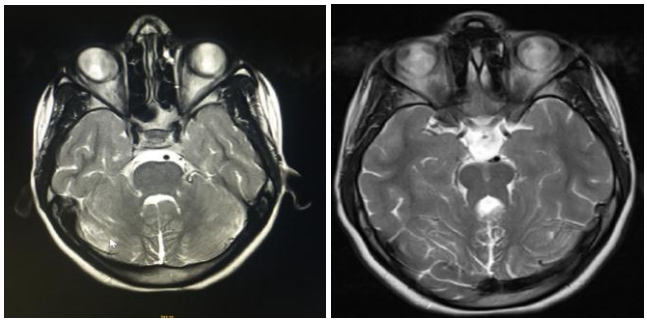

**Fig 3:** MRI brain showing Molar tooth sign, cerebellar vermis hypoplasia

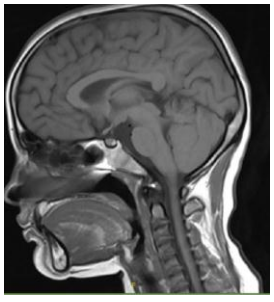

**Fig 4:** MRI brain showing cerebellar vermis hypoplasia

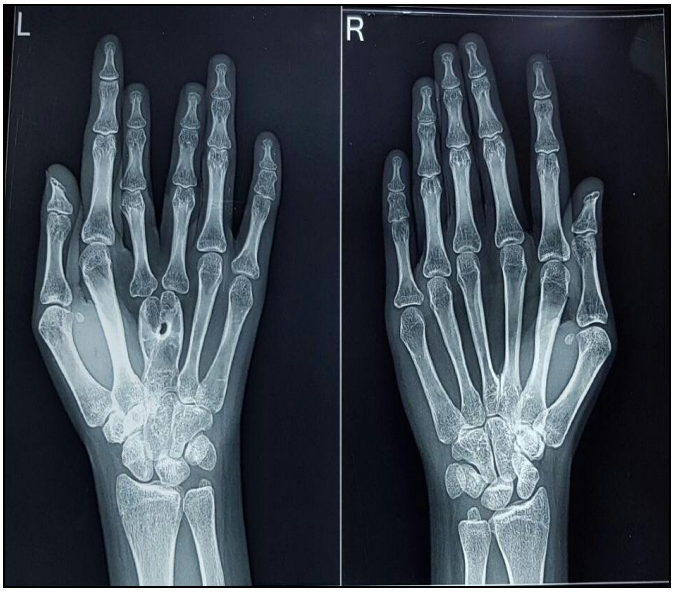


**Fig 5:** X-ray hand showing Y shaped metacarpals and mesoaxial poly Dactyly

## 3. Discussion

Classic or pure Joubert is a rare variant and diagnosis is based on three Criteria.

- MOLAR TOOTH sign on MRI
- Hypotonia in infancy and later development of ataxia
- Developmental delay

It is associated with other abnormalities like eye, skeletal, kidney, liver and endocrine problems. When the characteristic features of Joubert syndrome occur with one or more additional features, it is called Joubert syndrome and related disorders (JSRD) [3] or as a subtype of Joubert syndrome [6].

Joubert syndrome with Orofacio digital type has features of Joubert syndrome along with OFD syndrome features which are divided into six subtypes. OFD type II is associated with facial, hand, feet deformities, tongue Hamartomas and cardiac defects. OFD type VI is associated with mesoaxial Polydactyly with Y shaped metacarpals, renal disease and lobulated tongue with hamartomas.

Our case has features of Joubert syndrome with OFD features type II &VI.

## 4. References

1. NINDS Joubert Syndrome Information Page. NINDS. 2016. http://www.ninds.nih.gov/disorders/joubert/joubert.htm.
2. Overview. Joubert Syndrome & Related Disorders Foundation, 2016. http://jsrdf.org/what-is-js/.
3. Joubert syndrome. Genetics Home Reference, 2011, http://ghr.nlm.nih.gov/condition/joubert-syndrome.
4. Cassandra Kniffin L. Joubert Syndrome 10; Jbts10. OMIM, 2015. http://www.omim.org/entry/300804.
5. Choh SA, Choh NA, Bhat SA, Jehangir M. MRI findings in Joubert syndrome. Indian J Pediatr. 2009; 76:231-5.
6. Brancati, Francesco *et al*. Joubert Syndrome and related disorders. Orphanet Journal of Rare Diseases, 2010. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2913941/pdf/1750-1172-5-20.pdf.