

# Misdiagnosed AR Joubert syndrome 6 as congenital oculomotor apraxia: A Case Report

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## INTRODUCTION

Joubert syndrome is a rare autosomal recessive genetic disorder characterized by **hypoplasia of cerebellar vermis**. The typical clinical manifestations include hypotonia, ataxia, oculomotor apraxia and intellectual disability. However, there are also variety of presentations according to the subtypes of the syndrome and this variation makes it hard to get proper diagnosis. In this report, we describe **Joubert syndrome 6 which was misdiagnosed as congenital oculomotor apraxia**.

## CASE DESCRIPTION

### 27-years-old male

### Chief complaints

: **unwilled external rotation of left leg**

### Past history

- Hypertension (-), Diabetes mellitus (-)
- Hepatitis (-), Pulmonary tuberculosis (-)
- Trauma history (-)
- Operation history (+)
- Medial hamstring lengthening & derotational osteotomy due to left hamstring tightness and tibia deformity (2021.08.31.)

### Family history

- His father also had similar symptoms

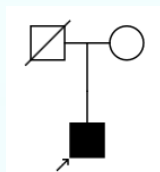
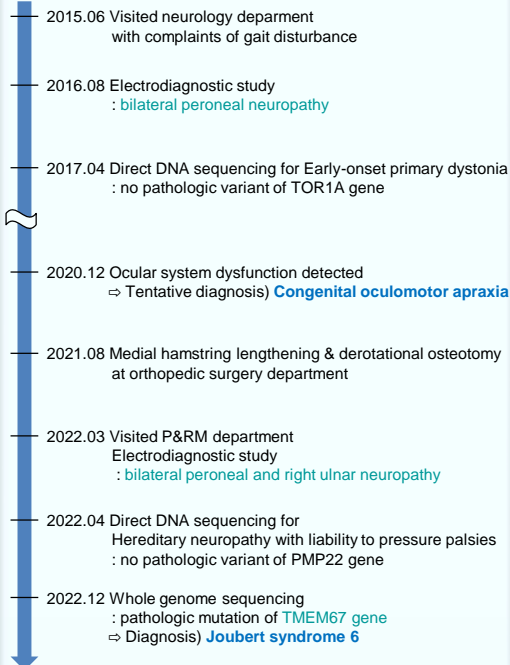


Fig. 1. Pedigree chart

### Physical examination

- Manual muscle test: decreased motor on all extremities (upper & lower limb: MRC grade 4)
- Range of motion limitation (-)
- Decreased one leg standing balance, limping gait pattern
- Berg balance scale: 50/56
- Modified Barthel index: 92/100
- Ophthalmic evaluation
  - decreased saccadic movement
  - convergence insufficiency
  - lateral gaze limitation

## Diagnostic flow



## Imaging study

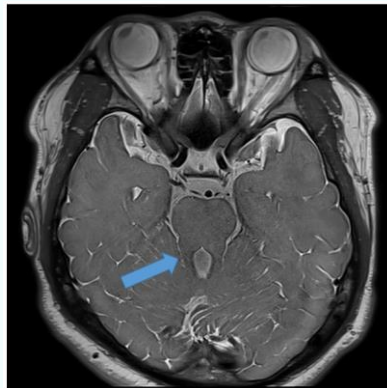


Fig. 2. Superior cerebellar peduncles on axial T2-weighted image

## CONCLUSION

In this report, we described a case of Joubert syndrome 6 with mild clinical presentation which was misdiagnosed as congenital oculomotor apraxia. The mean age of diagnosis of Joubert syndrome is 33 months, but this patient was diagnosed at the age of 27 with presentation of gait disturbance. It was difficult to make a proper diagnosis as the symptoms were not typical and relatively mild. **More detailed genetic testing like WGS could be useful to find accurate genetic diagnosis**. Also, further research will be needed about association of Joubert syndrome and **multiple mononeuropathies**.