Cognitive and behavioral features in Joubert Syndrome

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Introduction

- Joubert syndrome (JS) is a recessive **neurodevelopmental** disorder characterized by a distinctive cerebellar and brainstem malformation recognizable on brain imaging **"molar tooth sign".**

- The full spectrum of cognitive and behavioral phenotypes typical of JS is still far from being elucidated.

Objectif :

- To define the clinical phenotype and neurobehavioral features of patients with a neuroradiologically confirmed diagnosis of JS.

Méthodes :

- We conducted a descriptive retrospective study between 2004 and 2019 including patients followed up for JS in our department of Child and Adolescent Neurology in National Institute Mongi Ben Hmida of Neurology of Tunis. Epidemiological and radiological data, motor, cognitive and neuropsychiatric phenotypes were analyzed.

RESULTATS (1/2)

- 22 patients, 15 unrelated families (12 males and 10 females).
- Mean age : 10.8 years (entre 4 et 27 ans)
- Consanguinity :7 cases.
- Personal history of newborn respiratory distress: 4 patients.
- Initial clinical manifestation: psychomotor delay in all cases.
- Clinical examination :fig. 1.
- Brain MRI:
  - **Molar tooth sign** : all cases (fig. 2).
  - Associated supratentorial abnormalities: 5 cases.

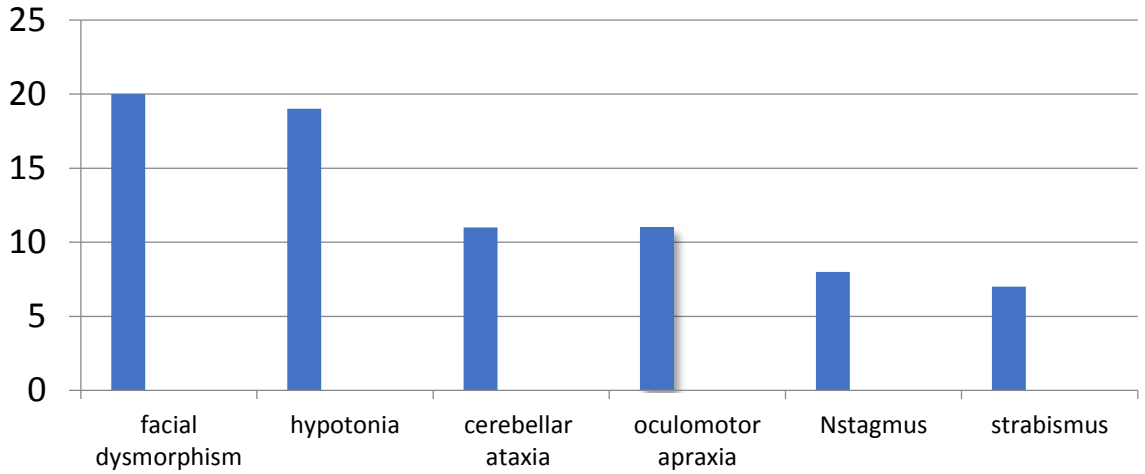

Figure 1: Data of clinical examination

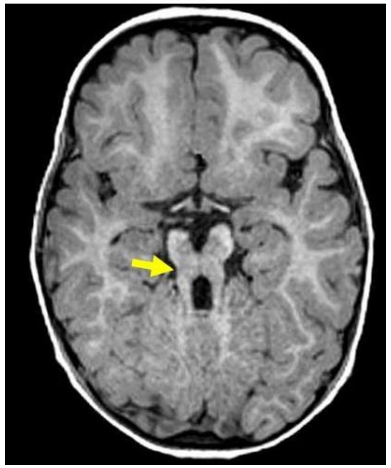

Figure 2: Brain MRI showing the "molar tooth sign" (yellow arrow)

RESULTATS (2/2)

- Cognitive and neuropsychiatric impairment: 12 cases (fig. 3).

- Our patients were classified into:
  - **Pure JS** :6 cases
  - **JS with ocular defect** :6 cases
  - **JS with oculorenal defect** :4 cases
  - **JS with hepatic defect** :2 cases
  - **JS with orofaciodigital defect** :4 cases

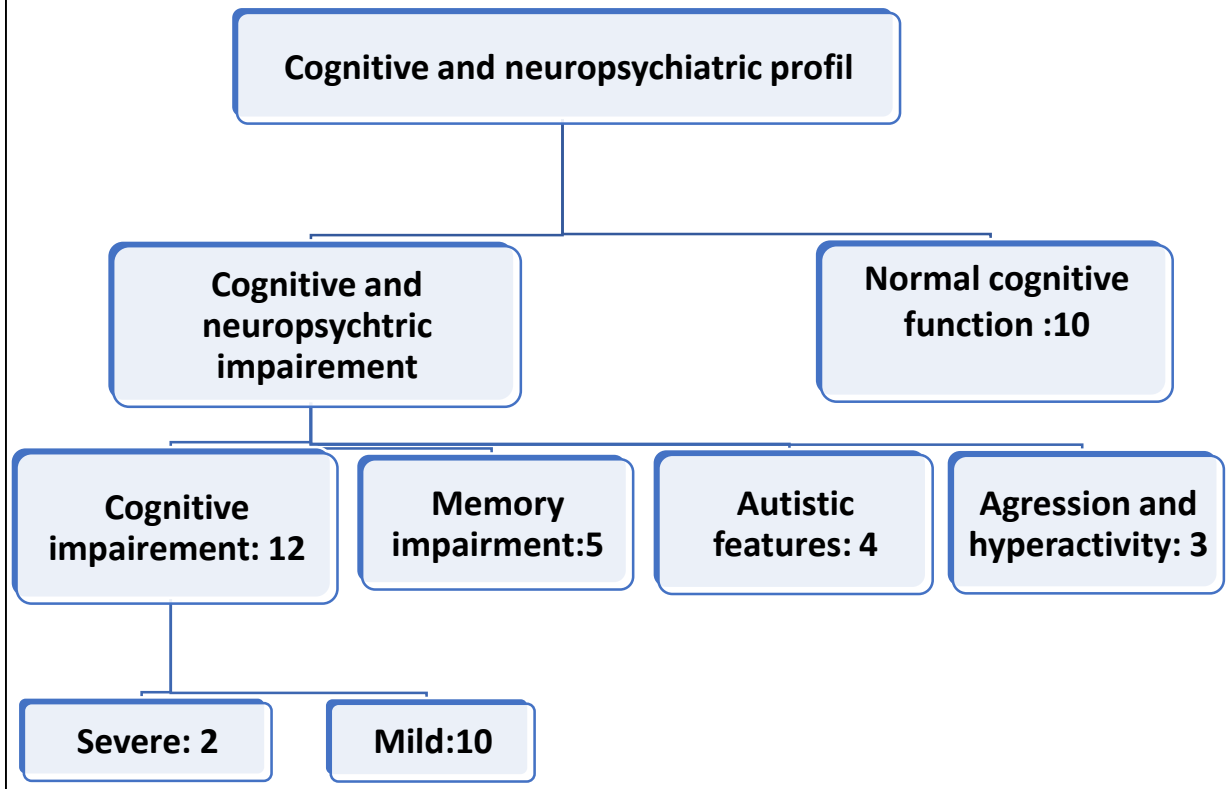

Figure 3: Cognitive and neuropsychiatric profil.

DISCUSSION ET CONCLUSION

- Our study reflects the **clinical variability** of JS and the high frequency of **cognitive and neuropsychiatric impairement.** It helps further the understanding of the multiple manifestations of atypical cerebrocerebellar development.

- These findings underscore the need for patients with JS to receive **neuropsychological and neuropsychiatric evaluations** in addition to routine medical and neurological tests.

- Detecting impairments in cognition and emotion in patients with JS **facilitates rehabilitation** measures to improve their functional skills and **quality of life**.

Chelsea L et al, 2018
Angela C et al, 2017