Genetics and Imaging Aspects of Joubert's Syndrome (JS)

Cherif Mohamadou Aidara*, Abdoulaye Dione Diop, Madeleine Diop, Delphine Diop, Abdoulaye Ndoye Diop and Sokhna Ba Diop

Radiology Office of Fann University Hospital, Switzerland

Submission: February 23, 2017; Published: March 27, 2017

*Corresponding author: Cherif Mohamadou Aidara, Radiology Office of Fann University Hospital, Cheikh Anta Diop Street, BP 5035 Dakar Senegal, Switzerland, Email: matouz@gmail.com

Commentary

JS was described for the first time by Dr. Marie Joubert four decades ago. It is an autosomal recessive neurodevelopmental genetic disorder. It is estimated to affect approximately 1 in 100,000 newborns without sex predominance [1].

JS involves the normal development of the cerebellum, the vermis and the brainstem. This explains significantly the clinical features during the course of the disease. In the neonatal period, axial hypotonia, abnormal ocular movements and respiratory disorders are more frequently observed. Axial hypotonia is important. Intensity of symptoms is variable but tend to decrease with age [1,2]. Developmental delay and mental retardation are also variable from one individual to another, which can be responsible for a difficulty in learning or a significant delay in schooling. Oculomotor disorders appear to be characteristic in this condition. It is an oculomotor apraxia associated with a nystagmus. Strabismus or ptosis may also be observed. Autistic abnormalities are also described. However, penetrance of this condition can vary from normal development for some children [2,3].

In addition to abnormalities of the vermis (dysplasia and hypoplasia) and the brain stem which are constant, Poretti et al described other abnormalities such as cephalocele, polymicrogyria, periventricular heterotopia, ventriculomegaly, hippocampal malrotation, hypoplasia or even agenesis of the corpus callosum and agenesis of the septum pellucidum. These findings show the usefulness of neuroimaging in this condition [2,3]. In our so called developing countries MRI is not often available and CT scan must be used efficiently.

Identification of responsible genes is important for genetic counseling especially in a level of restricted population or an identified group of individuals. At least 11 genes have been identified. Mutations are described on chromosomes 2, 6 or 12 among others. But their role in brain development is unclear [2,4].
Prognosis of this condition is favorable in the moderate form and depends initially on the intensity of respiratory disorders and other visceral abnormalities that are possibly associated; renal disorders (cystic dysplasia and/or nephronophthisis), liver disorders (hepatic fibrosis). Eye disorders may be associated with retinopathy. The mental retardation and developmental delay may require care in a specialized center depending on the specific demand of each child [1].

References
1. Joubert syndrome.