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Joubert Syndrome: A Rare Genetic Disorder Reported in Kurdish Family

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Abstract : Joubert syndrome regards as a congenital cerebellar ataxia caused by autosomal recessive carried on X chromosome. The disease diagnosed by brain imaging—the so-called molar tooth sign. Neurological signs were present from the neonatal period and include hypotonia progressing to ataxia, global developmental delay, ocular motor apraxia, and breathing dysregulation. These signs are variably associated with multiorgan involvement, mainly of the retina, kidneys, skeleton, and liver. 30 causative genes have been identified so far, all of which encode for proteins of the primary cilium or its apparatus, The purpose of our project was to detect the mutant gene (INPP5E gene) which cause Joubert syndrome. There were many methods used for diagnosis such as MRI and CT- scan and molecular diagnosis by doing ARMS PCR for detection of mutant gene that we were used in this research project. In this research for individual family which reported, the two children with parents, the two children were affected and were carrier.

Keywords : Joubert syndrome, genetic disease, Kurdistan region, Sulaimani
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