#1930 - A case report of Joubert syndrome	
Authors and Affiliations	Hadi Sorkhi (MD) 1*, Mohammad Reza Salehi omran1, Neda Joghtaei (MD) 1, 1. Non-Communicable Pediatric Diseases Research Center, Health Research Institute, Babol University of Medical Sciences, Babol, IR Iran
Corresponding Author and email	Hadi Sorkhi, Department of pediatric Nephrology Address: Non-Communicable Pediatric Diseases Research Center, No 19, Amirkola Children's Hospital, Amirkola, Babol, Mazandaran Province, 47317-41151, IR Iran. Tel-Fax: +98-11-32346963 E-mail: hadisorkhi@yahoo.com
Body	**Introduction:** Joubert syndrome (JS) is a rare genetic disorder and autosomal recessive. Joubert syndrome and related disorders (JSRD) are a group of congenital anomaly syndromes in which the diagnostic hallmark is the molar tooth sign (MTS). MTS is a complex midbrain malformation obvious on brain imaging. The JS causes mental and motor developmental retardation. Kidney involvement in JS is a rare disorder and occurs in 2-20% of cases. We reported a patient of JS with renal involvement. **Case report:** A 15 years old boy that was referred with JS. His laboratory data were showed rising of urea and creatinine. Kidney ultrasonogragy report was decrease the size of kidney with increase echogenicity. He was treated with regular hemodialysis with diagnosis of end stage renal failure. **Conclusion:** The JS is a rare genetic disorder, and kidney involvement is rare organ disease in this syndrome .So, renal failure must be considered any patient with JS.
References	**References:** 1. Parisi MA, Doherty D, Chance PF, Glass IA. Joubert syndrome (and related disorders)(OMIM 213300). European Journal of Human Genetics. 2007;15(5):511-21.

2. Joubert M, Eisenring J, Andermann F. Familial dysgenesis of the vermis: a syndrome of hyperventilation, abnormal eye movements and retardation. Neurology. 1968;18(3):302.

3. Maria BL, Hoang KB, Tusa RJ, Mancuso AA, Hamed LM, Quisling RG, et al. " Joubert syndrome" revisited: key ocular motor signs with magnetic resonance imaging correlation. Journal of child neurology. 1997;12(7):423-30.

4. Romani M, Micalizzi A, Valente EM. Joubert syndrome: congenital cerebellar ataxia with the molar tooth. The Lancet Neurology. 2013;12(9):894-905.

5. Parisi M, Glass I. Joubert syndrome and related disorders. 2013.

6. Atalar MH, Salk I, Egilmez H. Classical Signs and Appearances in Pediatric Neuroradiology: A Pictorial Review. Polish journal of radiology. 2014;79:479.

7. Usta M, Urganci N, Ozcelik G, Cetincelik U, Kafadar I, Ozguven B. Joubert syndrome and related disorders: a rare cause of intrahepatic portal hypertension in childhood. Eur Rev Med Pharmacol Sci. 2015;19(12):2297-300.

8. Sampathkumar K, Sooraj YS, Karunakaran N, Ganesh R, Mahaldar AR. Joubert syndrome. Kidney international. 2008;74(9):1222.

9. Gleeson JG, Keeler LC, Parisi MA, Marsh SE, Chance PF, Glass IA, et al. Molar tooth sign of the midbrain–hindbrain junction: occurrence in multiple distinct syndromes. American journal of medical genetics Part A. 2004;125(2):125-34.

10. Poretti A, Huisman T, Scheer I, Boltshauser E. Joubert syndrome and related disorders: spectrum of neuroimaging findings in 75 patients. American Journal of Neuroradiology. 2011;32(8):1459-63.

11. Sönmez F, Güzünler-Şen M, Yılmaz D, Cömertpay G, Heise M, Çırak S, et al. Development of end-stage renal disease at a young age in two cases with Joubert syndrome.

The Turkish journal of pediatrics. 2014;56:458-61.

12. Saraiva JM, Baraitser M. Joubert syndrome: a review. American Journal of Medical Genetics Part A. 1992;43(4):726-31.

13. Alorainy IA, Sabir S, Seidahmed MZ, Farooqu HA, Salih MA. Brain stem and cerebellar findings in Joubert syndrome. Journal of computer assisted tomography. 2006;30(1):116-21.

14. Elhassanien AF, Alghaiaty HA-A. Joubert syndrome: Clinical and radiological characteristics of nine patients. Annals of Indian Academy of Neurology. 2013;16(2):239.