Cumhuriyet Tıp Derg 2013; 35: 133-134
Cumhuriyet Med J 2013; 35: 133-134

Letter to the Editor-Editöre mektup
http://dx.doi.org/10.7197/1305-0028.1924

Tuberous sclerosis

Tuberoskleroz

Hakim Rahmoune*, Nada Boutrid, Belkacem Bioud

Department of Paediatrics (R.Hakim, MD, B. Nada, MD, Prof. B. Belkacem, MD), University Hospital of Setif, 19000 Setif, Algerıa

Geliş tarihi/Received: January 12, 2013; **Kabul tarihi/Accepted:** January 28, 2013

***Corresponding author:**

Hakim Rahmoune, MD, Department of Paediatrics, University Hospital of Setif, 19000 Setif, Algeria. E-mail: rahmounehakim@gmail.com

We read with interest the recent publication in Cumhuriyet Medical Journal from Aggarwal et al. [1] about a case report of a young patient with Tuberous Sclerosis (TS).

As explained in the article, TS is diagnosed by pooling different clinical and radiological signs; but those are not all present in the same period of life, some of them being prenatal or precocious, while other are more latent and are expected in late childhood or even in adulthood [2].

The authors also mentioned that TSC genes are tumor suppressor genes without referring to the mammalian Target of Rapamycin (mTOR) complex they inhibit [3].

In fact, this complex is the cornerstone of major (if not all) TS expressions; and that is why mTOR inhibitors like everolimus and sirolimus are more and more used/tested to treat a wide spectrum of TS clinical signs [4, 5].

These inhibitors also seem to be effective in both seizures and behavior disorders in affected patients [6, 7].

The Intellectual Quotient (IQ) of TS population is considered to be linked, as mentioned by authors, to the phenotype (mainly neurological with comitiality & tubers number) as well as to the TSC2 genotype [8]

This may open a window of opportunity to a genotype/phenotype tailored treatment of this phakomatosis.

References

1. Aggarwal EH, Jain D, Jain P, Kaverappa V, Kumar A, Yadav S. Tuberous sclerosis - A case report and review of literature. Cumhuriyet Med J 2012; 34: 79-85.
2. Yates JR, Maclean C, Higgins JN, Humphrey A, le Maréchal K, Clifford M, Carcani-Rathwell I, Sampson JR, Bolton PF; Tuberous Sclerosis 2000 Study Group. The Tuberous Sclerosis 2000 Study: presentation, initial assessments and implications for diagnosis and management. Arch Dis Child 2011; 96: 1020-5.
3. Dobashi Y, Watanabe Y, Miwa C, Suzuki S, Koyama S. Mammalian target of rapamycin: a central node of complex signaling cascades. Int J Clin Exp Pathol 2011; 4: 476-95.
4. Davies DM. Tuberous sclerosis: from gene to targeted therapy. Clinical Medicine 2012; 12: s7-s10.
5. Kohrman MH. Emerging treatments in the management of tuberous sclerosis complex. Pediatr Neurol 2012; 46: 267-75.

6. Galanopoulou AS, Gorter JA, Cepeda C. Finding a better drug for epilepsy: the mTOR pathway as an antiepileptogenic target. Epilepsia 2012; 53: 1119-30.
7. Russo E, Citraro R, Donato G, Camastra C, Iuliano R, Cuzzocrea S, Constanti A, De Sarro G. mTOR inhibition modulates epileptogenesis, seizures and depressive behavior in a genetic rat model of absence epilepsy. Neuropharmacology 2012.
8. Jentarra GM, Rice SG, Olfers S, Saffen D, Narayanan V. Evidence for population variation in TSC1 and TSC2 gene expression. BMC Med Genet 2011; 12: 29.