Tuberous sclerosis

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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

