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Tuberous sclerosis in a patient from Nigeria.

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Abstract

Tuberous sclerosis complex (TSC) is an autosomal dominant syndrome characterized by mostly benign tumors of the brain, skin, heart, kidney, and eye. Aberrations in the genes TSC1 and TSC2 which encode hamartin and tuberin, respectively, cause TSC. Because disease manifestations develop over time, early diagnosis and intervention are imperative for patients. TSC is not well described in patients from sub-Saharan Africa or of black African ancestry. Here, we report on a 4-year-old Nigerian boy with skin lesions and cardiac anomalies associated with TSC. Furthermore, we note that in areas with limited resources for genetic diagnoses, the common skin manifestations found in TSC may be especially useful clinical markers.

KEYWORDS: Nigeria; diverse populations; tuberous sclerosis

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