Constellation of Five Facial Features of Tuberous Sclerosis in a Child with a TSC2 1808A>G Mutation

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Tuberous sclerosis complex (TSC) is a rare neurocutaneous disorder that often causes renal and brain tumors [1–3]. Most patients with TSC have a small but variable number of cutaneous features that may give a clue to the diagnosis. We present the case of an 8-year-old boy with a TSC2 1801A>G mutation. On clinical examination, the patient was found to have five facial features of TSC, including a rash of facial angiofibromas, a shagreen patch, a forehead plaque, gingival fibromas, and dental pitting (Fig. 1).

The presence of so many features is unusual and interesting because the detailed imaging presented provides an excellent educational illustration of the facial features of TSC. There is no family history of TSC in this case. Diagnostic criteria for TSC usually include two major features or one major and one minor feature. The forehead plaque, angiofibromas and shagreen patch (connective tissue nevus) are major features, whereas dental pits and gingival fibromas are minor features. Although approximately 80% of cases of TSC generally have skin signs, most patients generally have one or at the most two features, so five features is very unusual. There is no clear correlation of the skin features with the mutation in most cases.

Oncologists should bear in mind the diagnosis of TSC for patients presenting with renal tumors. Often the face may give a clue to the diagnosis and indicate screening of other organs in the patient, including the brain for subependymal giant cell astrocytomas (SEGAs) [4].

Recently, everolimus has been licensed for the treatment of SEGAs. Evidence suggests that it may be effective in reducing the number and size of renal tumors and may reduce the intensity of facial features [5]. Development of a skin preparation, such as an ointment or emollient containing an mammalian target of rapamycin (mTOR) inhibitor, in the future may help reduce the appearance and severity of skin lesions for patients with TSC.

Figure 1. Facial features of tuberous sclerosis complex: connective tissue nevus/shagreen patch (1), angiofibromas (2), “forehead plaque” on cheek (3), gingival fibroma (4), and dental enamel pitting (5).

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