Chapter 20
Tuberous Sclerosis Complex

Patricia Treadwell

20.1 Introduction

Tuberous sclerosis complex (TSC) is a neurocutaneous disorder characterized by mutations in the TSC1 gene (encoding of hamartin) or TSC2 gene (encoding of tuberin). The mutations result in dysregulation of cellular hyperplasia.

20.2 Epidemiology

TSC occurs at a rate of 1:6000 to 1:10,000 live births [1]. TSC1 is located on chromosome 9q34 and encodes hamartin. TSC2 is located on chromosome 16p13 and encodes tuberin. Inheritance of TSC is considered to be autosomal dominant; however, the rate of spontaneous mutations is approximately 70%.

20.3 Clinical Findings

Cutaneous findings include hypomelanotic macules (ash leaf spots), facial angiofibromas (Fig. 20.1), periungual fibromas (Fig. 20.2), and connective tissue nevi – fibrous cephalic plaque (on the head and neck) (Fig. 20.3), or shagreen patch (often located on the lower back.

Other associated findings are epilepsy, learning difficulties, autism, and attention deficit disorders.

P. Treadwell (✉)
Department of Dermatology, Indiana University School of Medicine, Indianapolis, IN, USA
e-mail: ptreadwe@iupui.edu

P. Treadwell et al. (eds.), *Atlas of Adolescent Dermatology*,
https://doi.org/10.1007/978-3-030-58634-8_20

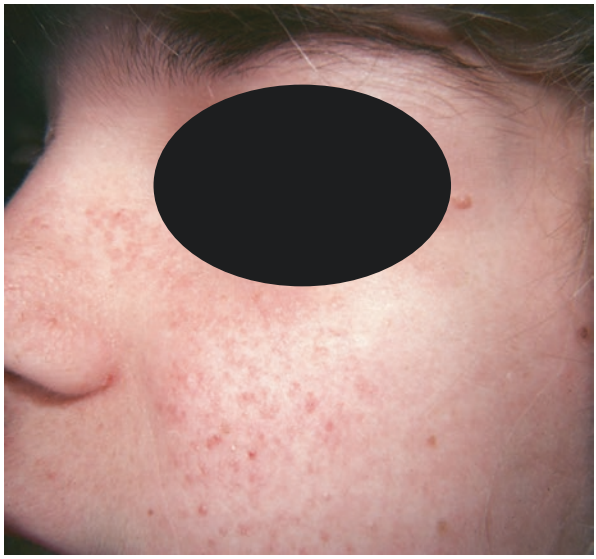

Fig. 20.1 Facial angiofibromas

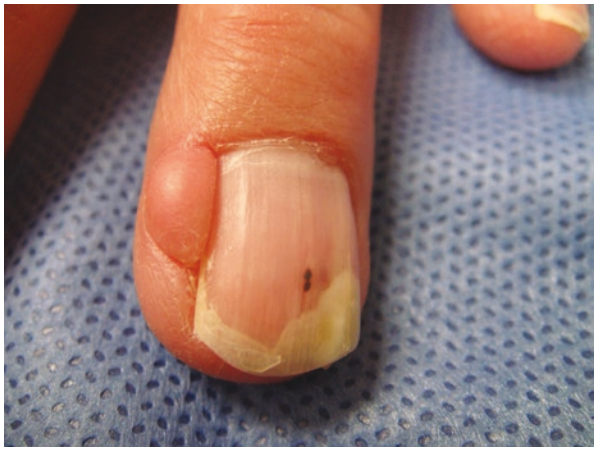

Fig. 20.2 Periungual fibroma

Hamartomas which may develop are retinal hamartomas, subependymal giant cell tumors (SGCTs), cardiac rhabdomyomas, renal and non-renal angiomyolipomas (AMLs), and pulmonary lymphangiomyomatosis (LAM).

Dental enamel pits and intraoral fibromas may be noted.

20.4 Laboratory

Imaging may show "cortical tubers" (occurring in about 80% of TSC patients), which are composed of abnormal neurons and glia.

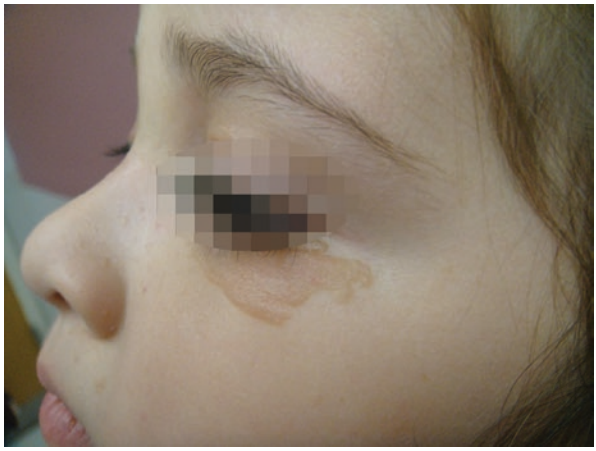

Fig. 20.3 Fibrous cephalic plaque

20.5 Treatment

Specific guidelines for surveillance assist in management of the hamartomas and associated findings. Consultation with neurology can be helpful to treat epilepsy. Topical or systemic mTOR inhibitors have been useful for specific complications.

20.6 Prognosis

Although the hamartomas are typically benign, they may cause a mass effect.

Reference

1. Randle SC. Tuberous sclerosis complex: a review. Pediatr Ann. 2017;46:e166–71.