



GENETICS AND GENODERMATOSES

CURRENT AND EMERGING MEDICAL THERAPIES FOR CUTANEOUS MANIFESTATIONS OF TUBEROUS SCLEROSIS COMPLEX

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Tuberous sclerosis complex (TSC) is a genetic multisystem disease with variable manifestations that can prominently involve the skin. The cutaneous manifestations of this disease can have a tremendous impact on the patient's quality of life. Reducing the dermatological burden, emerging treatments (e.g., topical mTOR inhibitors) have been shown to be effective in the management of angiofibromas. Shagreen patches, however, are more difficult to treat and possibly more challenging to manage with mTOR inhibitors as these fibroblast populations respond differently to mTOR inhibitors. A comprehensive review of the PubMed database was performed, with several studies and reports investigating topical mTOR inhibitors with varying formulations (cream, ointment, and gel) and concentrations. Recent multicenter clinical trials have provided additional insights into the management of TSC cutaneous lesions. This review will explore the current and emerging treatments for the primary cutaneous manifestations of TSC, and provide a framework for dermatologic management.

