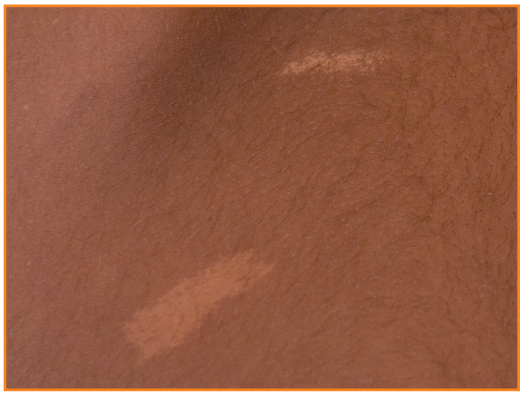
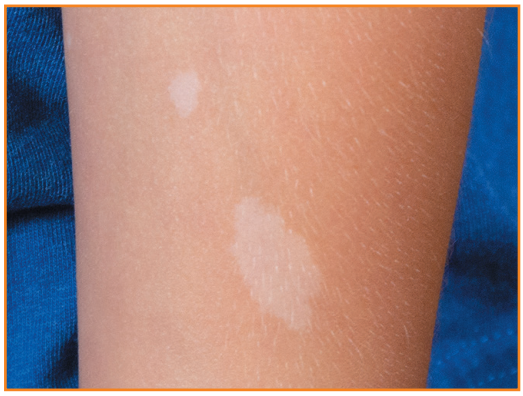
Racial differences in the dermatological manifestations of TSC

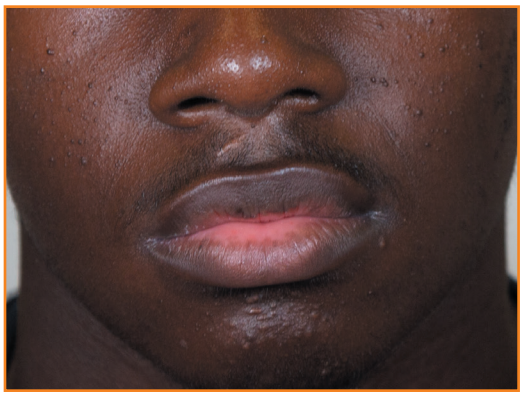
Tuberous sclerosis complex (TSC) is an autosomal dominant neurocutaneous disorder of non-malignant tumor growths throughout major organ systems and neurological, neuropsychiatric, renal, and pulmonary co-morbidities. Clinical diagnosis of TSC is based on the presence of a specific combination of major and minor features, of which four major and one minor are dermatological manifestations. Medical photographs of such manifestations are commonly shown as examples from White individuals, creating a potential barrier to accurately identifying these features in skin of color. Cutaneous manifestations are often the only visual manifestation of TSC for individuals with mild to moderate phenotypes of TSC. The aim of this infographic is to raise awareness of dermatological manifestations associated with TSC and compare the appearance of the four major skin manifestations between Black and White individuals with TSC.

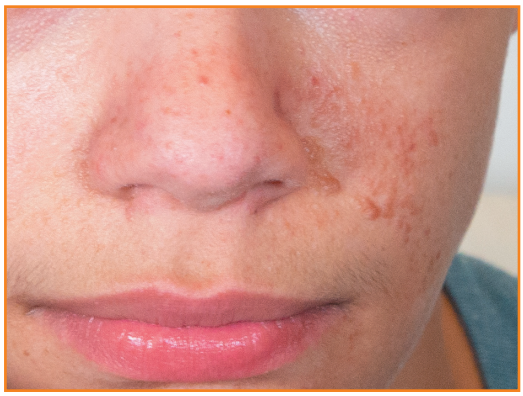
Hypomelanotic macules: Also called ash-leaf spots, these are the most common dermatologic feature of TSC in patients under the age of 1 year. They are often present on the limbs, trunk, and buttocks, but can be identified throughout the body. Hypomelanotic macules are typically present at birth and remain throughout life. A complete skin examination utilizing a Wood's lamp is recommended to visualize these findings across all races as mild hypopigmentation may be difficult to detect in darker skin.





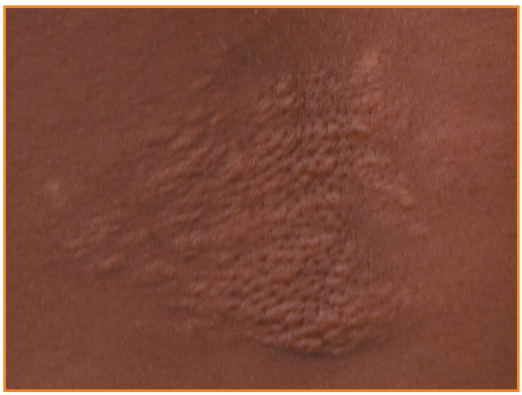
Facial Angiofibroma: Later in childhood, facial angiofibromas, also referred to as adenoma sebaceum, are a hallmark feature of TSC. Facial angiofibromas occur in up to 75% of patients and become more prominent with age. Facial angiofibromas often appear erythematous, raised, in a butterfly-shaped appearance across the nasolabial folds, and are more likely to appear hyperpigmented in darker skinned individuals. In Black individuals, facial angiofibromas may be confused for seborrheic keratoses or other benign epidermal growths or even other neurocutaneous disorders.

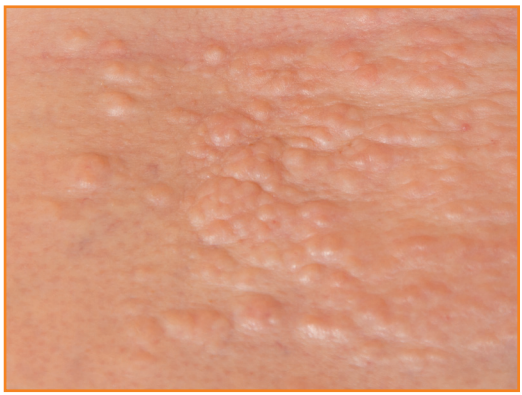




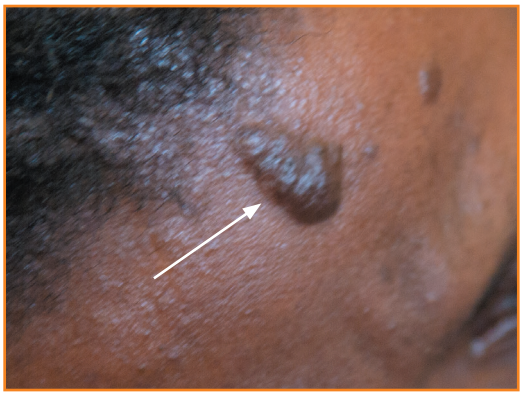
Shagreen Patches: Shagreen patches are a type of connective tissue nevus that typically appear as large, irregular, firm skin and colored to hypopigmented plaques across the lower back; however, they can appear throughout the upper and middle back, buttocks, and thighs. These lesions usually appear during early childhood. In the examples below, both patches are located on the lower back.

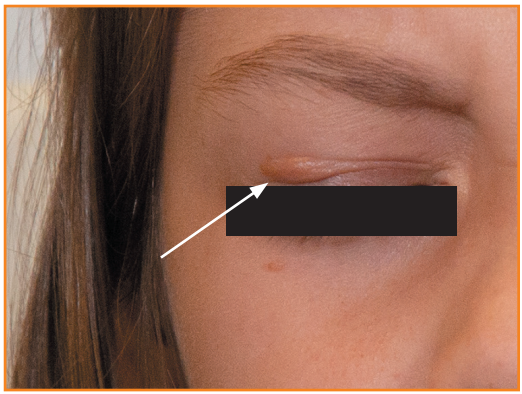
Scan the QR code to see common locations of shagreen patches.





Fibrous Cephalic Plaque: Fibrous cephalic plaques (FCPs) can be present from birth but are more noticeable during early childhood or adolescence. FCPs can be located on the face, scalp, or forehead and may appear rubbery to firm, smooth to bumpy, skin-colored, pink, red, or brown. Individuals with darker skin tones tend to have much darker lesions relative to surrounding skin than those with lighter skin tones. FCPs on the scalp were associated with decreased hair density in the location of the lesion. The size and distribution of FCPs is still not well understood; however, between 1-5 cm is commonly noted throughout the literature.





Photographs also shown in Pounders, Rushing et. al., Therapeutic Advances in Rare Disease. Infographic prepared by Ashley Pounders, FNP-C, Director, Medical Affairs, and Gabrielle Rushing, PhD, Director, Research, at the TSC Alliance. The authors kindly thank Dr. Tom Darling and Dr. Oyetewa Oyerinde for their thoughtful review and feedback and the individuals with TSC who allowed their photographs to be used.

Scan the QR code to read the *Therapeutic Advances in Rare Disease* article.

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