



801 Roeder Road, Suite 750
Silver Spring, MD 20910

Toll free: (800) 225-6872

Phone: (301) 562-9890

Fax: (301) 562-9870

www.tsalliance.org

E-mail: info@tsalliance.org

October 27, 2008

To Whom It May Concern:

I am writing to provide information about facial angiofibroma, a very common manifestation of Tuberous Sclerosis Complex (TSC). I understand that you are seeking information about the medical necessity to remove these lesions. On some occasions this surgery has been incorrectly considered a cosmetic procedure. I would like to describe what these facial lesions are, why it is important that they be removed, and why insurance companies agree to cover the procedure after acquiring this information.

Facial angiofibromas were first described in 1885 by Balzer and Menetrier. At this time, they were incorrectly termed adenoma sebaceum. We now know that the sebaceous glands are only passively involved, if at all. Facial angiofibromas are actually hamartomas (defined as benign, tumorous nodules of superfluous tissue). They have nothing to do with acne, and do not respond to treatments for acne. There is nothing the individual can do to prevent them. The typical facial angiofibromas are red to pink papules/nodules when they first appear, with a smooth, glistening surface. They are usually, but not always, bilaterally symmetrical, distributed over the centropalpebral areas, particularly in the nasolabial folds, onto the cheeks in a butterfly fashion, and on the chin. They are sometimes seen on the forehead, scalp region, or even laterally on the face. The upper lip is usually spared, but lesions may be seen on the eyelids.

The tumors found on the forehead and scalp are large fibromas. These flesh-colored plaques are soft or compressible or doughy-to-hard lesions. Single large or multiple lesions may be seen. The angiomatous (highly vascular with a lot of blood vessels and appearing red) appearance is usually absent from these lesions; however, some forehead plaques can be bright red. The major findings in histopathology of the facial angiofibromas are dermal fibrosis and vasodilation. Elastic tissue is absent from these lesions. As the angiofibromas mature, the collagen becomes sclerotic and layered. Very often the facial angiofibromas will go from being small, soft, red nodules to become large, hard dark brown nodules with age, especially in individuals with dark skin complexion.

Facial angiofibromas may appear as early as the first year of life, but very often do not begin to appear until the child is four to six years of age, or older. As the individual enters puberty, the lesions may become even more numerous and prominent. However, once these tumors appear on the face, they persist for life, and usually increase in size, if they are not removed.

The Tuberous Sclerosis Alliance developed a consensus document for appropriate treatment and removal of facial angiofibromas in individuals with TSC. In the past, dermabrasion and surgical removal were the only options for removal of facial angiofibromas. The use of CO₂ and pulse dye lasers to treat facial angiofibromas are now widespread and are proving to be more effective, less painful, and less likely to leave scars than older treatments. Laser treatment is also more easily tolerated during the healing period. General anesthesia may

A national non-profit organization dedicated to research, education and support.

Mission statement: Tuberous Sclerosis Alliance is dedicated to finding a cure for tuberous sclerosis while improving the lives of those affected.



801 Roeder Road, Suite 750
Silver Spring, MD 20910

Toll free: (800) 225-6872

Phone: (301) 562-9890

Fax: (301) 562-9870

www.tsalliance.org

E-mail: info@tsalliance.org

be necessary dependent on the individual's symptoms and ability to cope with the procedure, especially for those individuals with TSC who have severe learning disabilities.

Two types of laser are commonly used on each patient, including the CO₂ laser for the larger, fibrous tumors, and the tunable dye, pulse-type laser for removing very small lesions and the redness of lesions. Sometimes these procedures are done in a two-step procedure, utilizing the CO₂ laser first and then the pulse laser 3 to 6 months later to normalize the color. Sometimes both of these lasers are used together. The expertise and skill of the physician, as well as his/her familiarity with TSC and these procedures, reduces the risk of scarring and optimizes outcome.

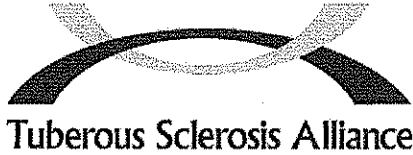
There is clinical evidence better results are obtained when facial angiofibromas are treated early after their appearance while they are still soft, red and not fibrotic. There is slower regrowth and less scarring if the angiofibromas are removed early rather than waiting for them to become large and fibrotic. Once the facial angiofibromas are removed utilizing laser treatments, repeated laser treatments may be required every 1-3 years depending on the rate of tumor regrowth. The growth rate and the interval between treatments will vary from one individual to the next, and should be tailored to the individual's medical needs.

There are several medical reasons why the facial angiofibromas should be removed, including the following:

1. These tumors are highly vascular and bleed very easily and profusely. It is often very difficult to prevent bleeding and to stop bleeding once it has started. This is particularly true for young males as they start to shave, and for any active individual who may run the risk of hitting or rubbing the lesions, resulting in bleeding.
2. Because the facial angiofibromas are so vascular, they are subject to infection if they are damaged and bleeding ensues. This is particularly dangerous around the nose and eye area because infection in this area can easily be transferred into the intracranial blood vessels and carried to the brain.
3. Facial angiofibromas found on the eyelids may become so large as to obstruct vision and may bleed into the eye when the individual rubs their eyes and damages the tumor. These lesions should definitely be removed so as not to promote either visual obstruction or possible infection.
4. Facial angiofibromas found on the nose may become so large as to impair the use of glasses. Again, these lesions should definitely be removed so as not to prevent an individual from wearing their glasses and/or causing bleeding when the glasses are worn.
5. The psychological impact of having facial angiofibromas can be devastating to a young individual with TSC. These tumors may become very large and grotesque in appearance if they are not removed. Individuals with TSC have a difficult time coping with all of the various manifestations of the disease, and it is exceptionally hard to fend off taunts and teasing by peers who notice and make fun of the growths on the face. These tumors will only get worse with age – large, darker and more prominent. Therefore, it is imperative that these tumors are removed to increase the individual's quality of life.

A national non-profit organization dedicated to research, education and support.

Mission statement: Tuberous Sclerosis Alliance is dedicated to finding a cure for tuberous sclerosis while improving the lives of those affected.



801 Roeder Road, Suite 750
Silver Spring, MD 20910

Toll free: (800) 225-6872
Phone: (301) 562-9890
Fax: (301) 562-9870
www.tsalliance.org
E-mail: info@tsalliance.org

6. Since these lesions are hamartomatous growths, these tumors are likely to grow back with time. The rate at which they return is different from one individual to another. For some, the tumors will not grow back several years, whereas other individuals will see their return within months. It is likely these tumors will have to be removed several times over the course of an individual's life. For a fortunate few, one procedure is sufficient and they do not return. Recent clinical experience shows that if the facial angiofibromas are removed early in life, they grow back more slowly, do not become as fibrous, hard and sclerotic, and they are easier and less expensive to remove.

TSC researchers are studying the function(s) of the TSC genes and their protein products, tuberin and hamartin, in skin cells to determine how the growth of facial angiofibromas may one day be halted and/or prevented, but currently the treatment of choice for facial angiofibromas is laser treatment.

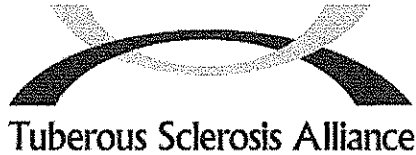
I would be happy to provide you with contact information for dermatologists and/or plastic surgeons who serve on the Tuberous Sclerosis Alliance Professional Advisory Board and who would be happy to answer your questions about treatment approaches for facial angiofibromas.

Thank you for your consideration of this information. Hopefully, this information has clarified the importance of treatment for all persons coping with these potentially disfiguring tumorous growths, facial angiofibromas. Please do not hesitate to contact the Tuberous Sclerosis Alliance if you would like additional information.

Sincerely,

Vicky Whittemore

Vicky Holets Whittemore, PhD
Vice President & Chief Scientific Officer
Tuberous Sclerosis Alliance
801 Roeder Road, Suite 750
Silver Spring, MD 20910
Phone: 301-562-9890
FAX: 301-562-9870
E-mail: vwhittemore@tsalliance.org



801 Roeder Road, Suite 750
Silver Spring, MD 20910

Toll free: (800) 225-6872
Phone: (301) 562-9890
Fax: (301) 562-9870
www.tsalliance.org
E-mail: info@tsalliance.org

References:

- Arndt KA (1994) Angiofibroma in tuberous sclerosis--argon laser. *Int J Dermatol* 33(7):522-3
- Darling T, Mausner M, Moss J (2009) Dermatological manifestations of tuberous sclerosis complex. In, *Tuberous Sclerosis Complex: From Genes to Therapeutics*, DJ Kwiatkowski, E Thiele, VH Whittemore (Eds), Wiley-VCH, Weinheim, Germany, In Press
- Bittencourt RC, Huilgol SC, Seed PT, Calonje E, Markey AC, Barlow RJ (2001) Treatment of angiofibromas with a scanning carbon dioxide laser: a clinicopathologic study with long-term follow-up. *J Am Acad Dermatol* 45(5):731-5
- Pantelis A, Bootz F, Kuhnel T (2007) [Laser skin resurfacing and fibrin sealing as successful treatment for facial angiofibromas in tuberous sclerosis.] *HNO* 2007 Oct 3 [Epub ahead of print] [Article published in German]
- Ratnam KV (1994) Cutaneous angiofibromas: treatment with the carbon dioxide laser. *Ann Acad Med Singapore* 23(1):67-8
- Rogers RS, O'Connor WJ (1999) Dermatologic Manifestations. In, *Tuberous Sclerosis Complex*, MR Gomez, JR Sampson, VH Whittemore (Eds), New York: Oxford University Press, pp. 160-180
- Sharma VK, Khandpur S, Khanna N (2004) An interesting case of unilateral angiofibromas successfully treated with pulsed dye laser. *J Eur Acad Dermatol Venereol* 18(5):641-2
- Song MG, Park KB, Lee ES (1999) Resurfacing of facial angiofibromas in tuberous sclerosis patients using CO2 laser with flashscanner. *Dermatol Surg* 25(12):970-3
- Tope WD, Kageyama N (2001) "Hot" KTP-laser treatment of facial angiofibromata. *Lasers Surg Med* 29(1):78-81
- Verheyden CN (1996) Treatment of the facial angiofibromas of tuberous sclerosis. *Plast Reconstr Surg* 98(5):777-83