Neurofibromatosis type 1 is defined by well-established clinical criteria.[5] The abnormality is located on chromosome 17 and exhibits autosomal dominant inheritance.

Most cases of Sturge-Weber syndrome are sporadic, but a familial distribution has been reported. As a result, the syndrome is believed to follow paradominant inheritance, such that the individual is heterozygotic for this inherited characteristic and phenotypically normal, and will only suffer from the disease should further mutation occur during the early phases of embryonic development.[6]

In neurofibromatosis type 1 the underlying alteration encodes a protein, neurofibromin, that is responsible for the pathogenesis of the condition. Sturge-Weber syndrome is explained by increased expression of fibronectin, which regulates angiogenesis and constitutes the cerebral response to chronic ischemic damage.

Therefore, there does not appear to be a relationship between the 2 neurocutaneous syndromes in either pathogenesis, transmission, or the underlying genetic defect. We raise the question of whether this case could be the result of the simultaneous occurrence of both process—an extraordinarily rare occurrence. Another explanation could lie in the pathogenesis of neurofibromatosis itself, since numerous articles link this condition with angioma and other vascular abnormalities.[7] Thirdly, it is possible that this could correspond to the most common neurocutaneous syndrome of all, Pascual Castroviejo type II syndrome, which encompasses neurologic abnormalities and various vascular abnormalities, including angioma. Reports of Pascual Castroviejo II syndrome include a description of a patient with neurofibromatosis type 1 and cutaneous and hepatic hemangioma.[8]

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Essential Progressive Telangiectasia

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To the Editor:

Essential progressive telangiectasia is a rare disorder that affects mainly middle-aged women. It is characterized by the insidious but progressive development of dilated blood vessels and telangiectases. These commonly begin on the lower limbs and gradually extend upwards, potentially extending to cover most of the body surface including the mucous membranes.

We report a 61-year-old woman with no relevant history who presented with the progressive appearance of erythematous lesions on both legs with onset 10 years previously. The lesions were occasionally pruritic and began to appear on both ankles before extending upwards. She reported that her sister and mother suffered from a milder form of a similar disorder. There was no personal or family history of repeated diarrhea, melena, mucosal bleeding, or neurologic abnormalities. Skin examination revealed a large number of enlarged blood vessels that blanched when pressed under glass, against a background of multiple violaceous erythematous punctate lesions from the ankle to the knee and in the distal portion of the thighs on both legs (Figure 1). There were no signs of atrophy, ulceration, tumors, or mucosal abnormalities, and the only outstanding feature was the presence of slight erythema on the face, with some associated papulopustular lesions. Laboratory tests including complete blood count, biochemistry for thyroid

Figure 1. Abundant telangiectases ascending the rear surface of both legs.

hormones and hepatic markers, protein analysis, immunoglobulins, antinuclear antibodies, anti-DNA, and cryoglobulins all gave normal or negative results. Histology showed the presence of a normal epidermis with a papillary dermis peppered with thin-walled dilated vessels and no signs of vasculitis. Treatment was prescribed with doxycycline 100 mg twice a day for 12 weeks, with no significant improvement. The patient rejected other treatment options like laser therapy, as she was suffering no systemic repercussions and the symptoms were currently stable.

Essential progressive telangiectasia is a rare clinical condition characterized by progressive dilation of the blood vessels, with no underlying cause, and with no association to any other visible sign. The progressive ascending and symmetrical development of this condition means that lesions can extend to the trunk and upper limbs.[1] Cases of conjunctival and oral mucosal involvement have been described.[2,3]

Systemic symptoms are unusual, although there have been isolated cases of associated gastrointestinal bleeding[3] and 1 case associated with autoimmune disease.[4] The condition has also occasionally been associated with bronchogenic carcinoma[5] or chronic sinusitis.[6]

From a histopathologic point of view, the presence of dilated veins in the upper dermis is a characteristic finding for this condition, sometimes accompanied by a slight superficial perivascular inflammatory infiltrate. Ultrastructural studies suggest the vessels affected by telangiectasis are principally dilated capillaries.[7]

The pathogenesis of the disease is unknown, although some authors have suggested it to involve a vascular reaction to the formation of fibrin microaggregates in capillaries located in areas of relative venous stasis following bacterial or fungal infection.[7] However, the lack of consistent association with bacterial or fungal infection and, above all, the very rare satisfactory response to treatment with antibiotics or fungicides, throw doubt on this hypothesis.

The differential diagnosis for essential progressive telangiectasia also includes both secondary telangiectasia associated with other conditions and other causes of primary telangiectasia. Differential diagnosis between primary telangiectasia and hemorrhagic telangiectasia, ataxia-telangiectasia, or serpiginous angioma is especially important. When considering secondary telangiectasia, we would also stress the importance of differential diagnosis with symptoms present in skin diseases due to venous stasis, connective tissue diseases, or telangiectasia macularis eruptiva perstans.

The condition has occasionally been reported to respond to treatment with tetracyclines,[8] funcigides,[7] and even acyclovir,[4] although resistance to these is also common. Laser treatment appears to be the best course of action currently available. Good results have been reported for 585 nm pulsed dye laser[9] and Nd:YAG (neodymium-doped yttrium aluminium garnet) laser.[10]

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