CASE REPORT

Ringed telangiectasias: an unusual presentation of telangiectasia macularis eruptiva perstans

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SUMMARY

Telangiectasia macularis eruptiva perstans (TMEP) is a rare form of cutaneous mastocytosis. While most cutaneous mastocytoses occur in children and are asymptomatic, TMEP occurs predominantly in adults and is associated with systemic manifestations, requiring medical management. TMEP is typically characterised by scattered red-brown macules on the trunk and extremities, but must be differentiated from other telangiectatic conditions such as scleroderma, hereditary haemorrhagic telangiectasia and telangiectasias secondary to cirrhosis. Practitioners must be aware that variants to the classic presentation of TMEP exist, such as the ringed telangiectasias we describe. Diagnostic workup including tissue biopsy must be considered in such patients after a thorough history and physical have been performed and other telangiectatic processes have been ruled out. The treatment of cutaneous mastocytosis aims at controlling symptoms and preventing mast cell degranulation. Cosmetic treatment includes the use of Psoralen and ultraviolet A (PUVA) therapy, total skin electron beam radiation and flashlamp pulsed-dye laser treatment.

BACKGROUND

Telangiectasia macularis eruptiva perstans (TMEP) is a rare form of cutaneous mastocytosis. Unlike other forms of cutaneous mastocytosis, TMEP primarily affects adults. It typically presents with multiple ill-defined tan to red-brown macules distributed symmetrically on the trunk and extremities. Ring-shaped telangiectasias without the typical red-brown background constitute a rare presentation of the disease. Appropriate identification and diagnosis of this variant are important in differentiating TMEP from other systemic telangiectatic conditions and ensuring adequate treatment.

CASE PRESENTATION

A 70-year-old woman presented due to an increasing number of telangiectasias on her skin. Over the preceding 10 years, she had experienced itching and had developed many ring-shaped telangiectasias on the neck and torso. The pruritus was in non-telangiectatic skin. She complained of nocturnal hot flashes, episodic abdominal cramping and diarrhoea. She denied dysphagia, Raynaud's-type cold intolerance, wheezing, shortness of breath and stiffness or swelling in her fingers. Physical examination revealed many 1–2 cm oval clusters of telangiectasias on the neck, chest, back and proximal arms

(figures 1 and 2). The telangiectasias blanched to the touch and exhibited central clearing resulting in the appearance of ring-shaped telangiectatic macules. No periungual vascular changes, sclerodactyly or cutaneous calcinosis was appreciated.

INVESTIGATIONS

Initial investigation

Liver enzymes were within normal limits.

DIFFERENTIAL DIAGNOSIS

TMEP is a form of cutaneous mastocytosis that primarily affects adults. It typically presents with multiple tan to red-brown macules distributed symmetrically on the trunk and extremities. Skin biopsy helps differentiate TMEP from other conditions that cause telangiectasias.

Generalised essential telangiectasia is most common in women in their 40s and 50s and presents initially as dilated veins and capillaries on the lower legs and later spreads to involve much of the body. It is not associated with other skin lesions or systemic manifestations.¹

Dermatomyositis presents with telangiectasias and other vascular changes on the periungual skin and is associated with muscle weakness, a pink-violet (heliotrope) rash on the eyelids and a pruritic scaly rash on the shoulders (shawl sign).¹

CREST syndrome, a limited variant of systemic scleroderma, may present with telangiectasias. The telangiectasias are usually on the face, palms and mucous membranes, not the trunk as is the case in TMEP. The acronym, CREST, is used to summarise the association of calcinosis (C), Raynaud's phenomenon (R), esophageal dysmotility (E), sclerodactyly (S) and telangiectasias (T).

Telangiectasias seen in patients with cirrhosis may be accompanied by palmar erythema, scleral icterus, jaundice, pruritus and ascites, most of which are not present in our patient and is further less likely due to the normal liver enzymes findings.

Further investigation

Histopathological examination following biopsy revealed many telangiectatic dermal blood vessels. A Giemsa stain confirmed the presence of an increased number of mast cells (figure 3). These features are characteristic of TMEP.

Screening for systemic mastocytosis included a complete blood count, serum tryptase and lactate dehydrogenase, which were within normal limits.

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Figure 1 Multiple blanchable telangiectasias on the torso, neck and proximal arms.

TREATMENT

The patient was started on an antihistaminergic agent and was counselled to avoid mast-cell degranulators. She also underwent cosmetic treatment with pulsed-dye-laser on her back with excellent results (figure 4).

OUTCOME AND FOLLOW-UP

At her latest follow-up, the patient had no new telangiectasias and her existing lesions were less erythematous and her pruritus was significantly improved.

DISCUSSION

Mastocytosis is a disorder characterised by mast cell proliferation and infiltration into various organs. The skin is the most common organ affected, but the gastrointestinal tract, liver, spleen, lymph nodes and bone marrow may also be involved.² Cutaneous mastocytosis usually presents in childhood with the majority of patients (85%–90%) experiencing a marked reduction in symptoms or complete resolution by adolescence.³ In adults, mastocytosis also typically presents initially on the

Figure 3 Histopathological image with Giemsa stain confirming the presence of increased number of mast cells within the dermis.

skin, but it is much more likely to be associated with systemic progressive disease. The disorder results from mutations in the proto-oncogene c-kit, which codes for a receptor tyrosine kinase that binds a mast cell growth factor. Adult onset mastocytosis has been associated with a somatic point mutation in codon 816 that results in a constitutively active receptor and thus mast cell hyperplasia.⁴

TMEP, a rare form of cutaneous mastocytosis, accounts for less than 1% of cases.⁵ It typically presents with multiple ill-defined tan, red-brown macules distributed symmetrically on the trunk and extremities. Despite the name, telangiectasias may not be present. Localised or systemic symptoms secondary to degranulation of the infiltrating mast cells may include: pruritus, blistering, flushing, dyspnoea, lightheadedness, heartburn and diarrhoea. Unlike other forms of cutaneous mastocytosis, TMEP primarily affects adults.⁶ We describe a unique case of TMEP in which large numbers of ring-shaped telangiectasias were present without other red-brown pigmentation. Therefore, it is important to consider TMEP in the differential diagnosis in individuals presenting with diffuse telangiectasias. Other entities

Figure 2 Closeup of biopsy site.

Figure 4 Closeup of biopsy site after pulsed dye laser treatment.

resulting in increased numbers of telangiectasias include: localised scleroderma or CREST syndrome, generalised essential telangiectasia, hereditary haemorrhagic telangiectasia and telangiectasias due to hepatic cirrhosis.

In cutaneous mastocytosis, serum tryptase levels are normal to mildly elevated. Tryptase values greater than 20 ng/mL strongly suggest systemic disease.⁷ While TMEP is generally thought to be isolated to the skin, systemic involvement has been reported.⁸ Therefore, patients presenting with TMEP should undergo screening using serum tryptase, and if over 20 ng/mL, bone marrow biopsy should be considered.

The treatment of cutaneous mastocytosis aims at controlling symptoms and preventing mast cell degranulation. Factors that trigger degranulation include: friction, exercise, certain foods (shellfish, cheese and fermented meats), alcohol, temperature changes and medications (NSAIDs, opioids, muscle relaxants, local anaesthetics). TMEP is often refractory to therapy, and there is not a gold standard treatment regimen. However, there are reports of decreased pruritus with antihistamines and leukotriene antagonists.⁹ The telangiectasias are amenable to PUVA therapy,⁵ total skin

electron beam radiation¹⁰ and a 585 nm flashlamp pulsed-dye laser treatment if desired for cosmetic reasons. The cosmetic therapy from the pulsed-dye laser is thought to be secondary to the reduction in vasculature without effecting mast cells and thus may only serve a temporary reduction in symptoms.¹¹

Learning points

- There are many causes of telangiectasias. A complete physical examination, review of systems and sometimes a skin biopsy is indicated to help determine the cause.
- As telangiectasia macularis eruptiva perstans (TMEP) is associated with systemic manifestations, patients diagnosed should have a serum tryptase checked for systemic disease. If greater than 20 mg/mL, the patient should be referred to Hematology-Oncology for a bone marrow biopsy.
- Patients with TMEP should avoid mast-cell degranulators such as alcohol, opioids, aspirin, quinines, scopolamine, polymyxin-B and d-tubocurarine.
- Topical creams are not typically helpful in the treatment of telangiectasias, but laser therapy, such as pulsed dye laser, can be very effective and lead to high patient satisfaction.

Patient's perspective

It was extremely frustrating to be dealing with these symptoms without knowing what was causing them or how to treat them. It started off with just 1 or 2 spots, but over the span of 10 years, I developed so many more. And at some point during this time, I started feeling itchy in different parts of my body. I was told these symptoms might be due to liver disease, but many doctor visits later, I was found to have a normal liver. I was scared. I did not know what was happening to my body. And I definitely did not like having these spots all over my skin. When I was finally diagnosed with TMEP by Dr Seiverling, I was thankful to finally know what was causing all of these symptoms, but I now had a new set of worries. Tests were being done to check if this disease involved my bone marrow or intestines. It was scary. And then I was given a list of foods and drugs that I should stay away from to prevent this from flaring up. It took a little time to get used to this new lifestyle, but it was worth it. One of the best things was the laser therapy I received. My skin started to look much nicer as the spots faded and the itching went away too.

So my overall perspective after living with this disease for so many years is that things are not always as they appear. I spent years being told I may have condition X or disease Y, all while my ability to function in the world was going down. I was so itchy and red that I could not stand to be out of the home at all. So when I was finally diagnosed with TMEP and treated, it was like a whole new life for me. The laser treatments really helped my itching and appearance of my skin. I no longer feel limited in my daily activities. And learning about how this disease can affect my other organs was scary, but I feel safe knowing that I am being monitored carefully every year for spread of the disease and can be treated immediately if a new symptom comes up or if something is detected from my blood work. I am very thankful to Dr Seiverling for everything she has done for my health.

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