Perioral Pigment Spots in a 5-year-old Girl: A Quiz

Rikke Wallentin[1], Ida Vogel[2] and Anette Bygum[1]
[1]Department of Dermatology and Allergy Centre, Odense University Hospital, DK-5000 Odense C, and [2]Department of Clinical Genetics, Aarhus University Hospital, DK-8000 Aarhus C, Denmark

A 5-year-old girl was referred to the Department of Dermatology with hyperpigmented macules around her mouth. The skin lesions had developed gradually over the previous 2 years. Clinical examination revealed scattered small brownish macules on her buccal mucosa (Fig. 1A) while gums were unaffected. On her lips and the adjacent parts of the skin she had multiple punctate and somewhat larger ink-coloured hyperpigmentations (Fig. 1B). Single lesions were identified at the root of her nose and below her eyes. No abnormal pigmentation was seen on her hands, feet or perianal skin. Besides a mild atopic dermatitis and a surgically corrected congenital valvular pulmonary stenosis, the girl was otherwise healthy. No hereditary diseases were known in the family.

What is your diagnosis? See next page for answer.

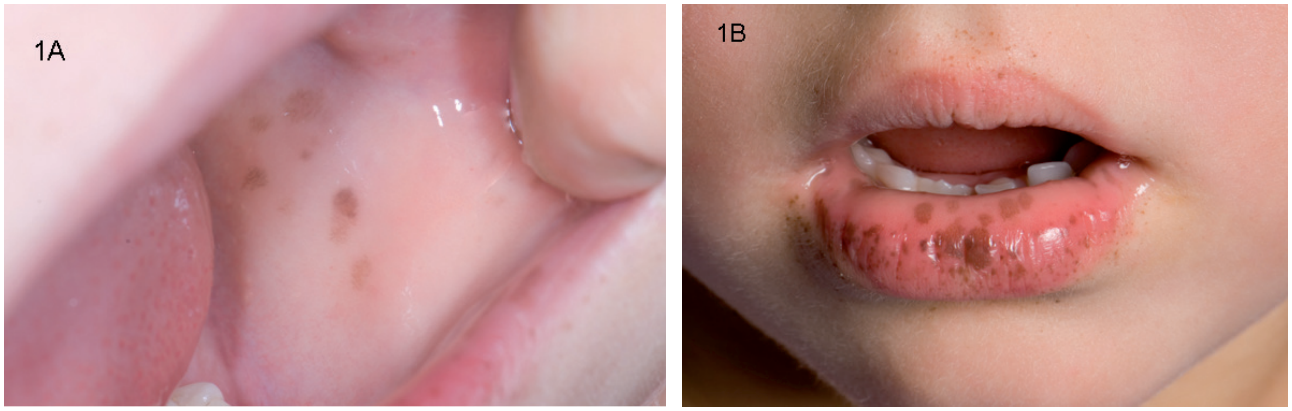

Fig. 1. A 5-year-old girl with small brownish macules on her buccal mucosa and multiple brownish and ink-coloured hyperpigmentations on the lips.

Perioral Pigment Spots in a 5-year-old girl: Comment

Diagnosis: Peutz-Jeghers syndrome

When the patient was seen at the Department of Dermatology the possibility of Peutz-Jeghers syndrome (PJS) was raised because of the mucocutaneous lentigines on the vermillion and buccal mucosa. She was referred to the Department of Paediatrics, where a capsule endoscopy was performed and a single polyp in the small intestine was found. A gastroscopy was also performed showing numerous polyps up to 1 cm in diameter in the stomach and duodenum. Histology revealed hamartomatous polyps. The family was referred for genetic investigation, and a *de novo* deletion of exon 1 and 2 of the *STK11* gene was detected in the girl.

A control regimen was planned with annual outpatient visits and haemoglobin analysis. Gastro- and colonoscopy were planned in addition to capsule endoscopy every third year.

At the age of 9 she presented with an acute abdomen and rectal bleeding caused by invagination of jejunum in relation to a pedunculated polyp measuring 2 × 2.4 cm. She needed acute surgery with resection of 6 cm of the small intestine.

PJS is a rare autosomal dominant condition. The incidence of PJS is estimated to be between 1:50,000 to 1:200,000 live births (1). It is characterized by distinctive mucocutaneous lentigines (around the mouth, nostrils, perianal area, hands, fingers, feet and toes) and intestinal hamartomatous polyposis. The polyps are generally benign hamartomas, but malignant transformation can occur.

The diagnosis is made on the basis of histologically confirmed hamartomatous polyps in combination with either a positive family history of PJS or mucocutaneous pigmentation. Eighty to 94% of the patients fulfilling the diagnostic criteria of PJS are found to have germline STK11 mutations or deletions (2, 3). As patients with PJS have an increased risk for different cancers (4) (breast, colon, pancreas, stomach, ovary, lung, small bowl, cervix, uterus), a suggestion for a surveillance regimen has recently been published – also to avoid complications with polyps, such as intussusception. Nevertheless, our patient had had an endoscopy performed only one month before the acute incident. While the treatment options for the gastrointestinal manifestations have been well established, the treatment of mucocutaneous melanosis is more empirical. Lentigines can be found in more than 90% of the patients and for some patients be a cosmetical nuisance (3). Reports on laser treatments and IPL can be found in the literature (5, 6). However, the hyperpigmented skin lesions will typically fade spontaneously when patients grow up.

We present this case to make clinicians aware that being observant to the cutaneous stigmata of PJS can lead to a diagnosis of this rare disorder, partly avoiding possible gastrointestinal complications which might appear already in childhood as well as associated cancer later in life. Also, even with a known diagnosis and regular examinations, the complications of PJS cannot totally be avoided.

References

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