Common lesions in a rare entity – Gardner's syndrome

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Gardner's syndrome (GS) is a rare genetic disorder which is a variant of familial adenomatous polyposis (FAP). It is characterized by a coexistence of multiple intestinal polyps with bone and soft-tissue tumors, frequently impacted teeth or cutaneous and subcutaneous cystic lesions. The syndrome is caused by a mutation of the adenomatous polyposis coli gene (APC) located at chromosome 5q21. The majority of patients have a family history of this disorder [1, 2]. We demonstrate a female patient with this genetic syndrome, which had not been recognized for a long time

A 38-year-old female patient was admitted to the Dermatology Clinic at the Silesian School of Medicine in Katowice due to multiple skin lesions on the trunk. Clinically, these cutaneous findings were oval forms, 2–3 × 1–2 cm in size. All of them were palpable, soft and well circumscribed from the surrounding skin. An initial, clinical diagnosis of epidermoid cysts was further confirmed by histopathology of all five excised lesions (Figures 1, 2).

The first skin lesions appeared 12 years earlier and probably resembled cutaneous fibromas due to a previous clinical suspicion of Recklinghausen disease. Since the beginning, the lesions have been localized predominately on the skin of the abdomen and gradually they started to spread over the trunk and into proximal surfaces of the limbs. Presumptively, similar cutaneous findings were present in a father of our female patient. The man died 7 years earlier at the age of 54 due to a lung cancer and the skin lesions could not be investigated. The family history also highlighted a stomach cancer in a grandfather (died 12 years earlier), as well as a bone tumor of a maxillary sinus in a female cousin of our patient.

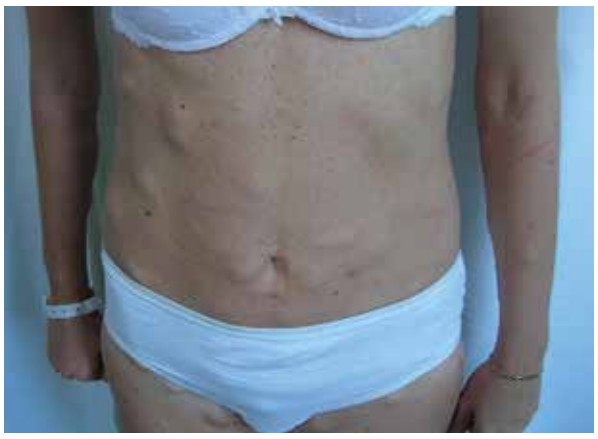

Figure 1. Subcutaneous tumors within the abdominal wall and around the inguinal areas

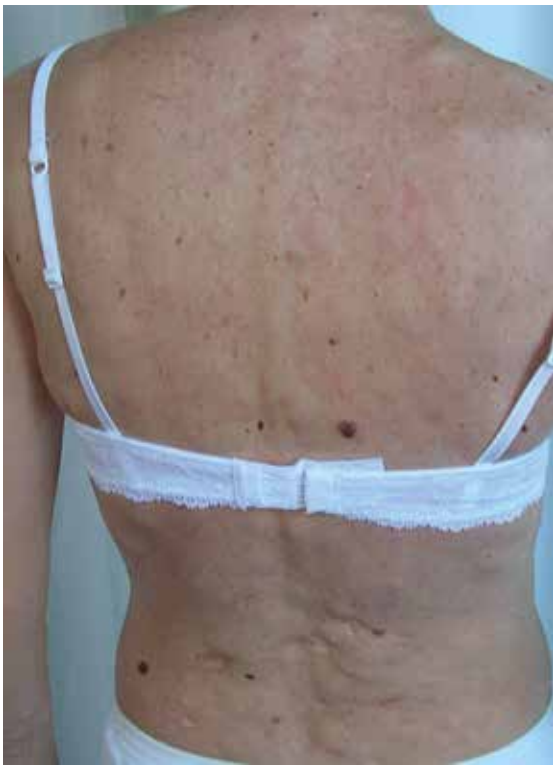

Figure 2. Multiple disfiguring skin findings on the back

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All these pathologies in the family motivated our female patient to examine herself for potential malignancies. Retrospectively, none abnormalities were detected in several diagnostic tests such as X-ray of the chest, ultrasound imaging (thyroid, abdominal and breast), as well as mammography (taken earlier than the usual screening test in Poland). Cervical smear tests were negative. The most of laboratory blood measurements (ESR, glucose, lipids, electrolytes, urinalysis, TSH, fT3, fT4) stayed within normal limits. Only the plasma iron level was significantly lowered (9 µg/dl; normal: 60–180 µg/dl) and there was a mild anemia: hemoglobin 10.4 g/dl (normal: 12–16 g/dl), red blood count 3.8 million/µl (normal: 3.5–5.2 million/µl) with mean cell volume 78 fl (normal: 82–92 fl). Panendoscopy did not reveal any pathologies, but 23 small polyps were detected at colonoscopy. Their histopathology provided a diagnosis of adenomatous polyps. Eventually, the third colonoscopy (2 years after the first one) together with numerous cutaneous lesions (later classified as cystic ones) provided a suspicion of the genetic basis of reported pathologies. Then, the patient was referred by a local surgeon to genetic tests towards mutations in the APC gene. All these cutaneous and colonic manifestations together with detected mutations in the APC gene provided a diagnosis of GS.

Due to the known extracolonic manifestations of GS, the patient was further referred to the ENT doctor, dentist and ophthalmologist. All of them, as well as dental X-ray or fundoscopy, did not reveal any abnormalities. Further colonoscopies (repeated every 6 months) have still detected another polyps, however the patient has not agreed to preventive proctocolectomy suggested by surgeons. Fortunately, panendoscopy as well as other imaging tests have still remained free from pathologies. Parameters of erythrocytes were normalized after an oral iron supply. The patient has stayed at constant follow-up of local surgeons and specialists mentioned above.

She was admitted to our Dermatology Clinic due to progressively appearing skin lesions to have them removed surgically, however for cosmetic reasons only. It should be mentioned that we had the first contact with this patient 3 years after she was diagnosed with GS and this case is largely based on retrospective analysis of her previous medical history.

An association of intestinal polyposis, "surface and bone tumors" and dominant inheritance was reported by Eldon Gardner in 1951 [1]. Gardner's syndrome was initially thought to be an entity separate from familial adenomatous polyposis (FAP), but currently it is known for one tail of FAP spectrum disorders [2, 3]. Apart from the multiple colonic polyps that are typical for FAP-related disorders, GS can also manifest some characteristic extracolonic findings: soft tissue tumors, benign osteomas and some dental abnormalities [4]. Frequently observed skin lesions (in 50–65% of GS patients) are epidermoid cysts. Their onset is usually prior to the puberty, often yet before intestinal polyps are considered. Cysts are asymptomatic, but occasionally they can be inflamed, pruritic with a tendency to rupture [2, 5]. Typically, no malignant complications are related to them. Although, cutaneous lesions can be a prominent disfigurement for GS patients, the main medical concern is associated with multiple adenomatous polyps of the colon. Polyposis can stay silent for a long time. Non-specific symptoms such as remittent colicky abdominal pain, diarrhea, blood-stained stools, rectal discharge of mucus as well as loss of weight or anemia may occur before a malignant transformation of the polyps [6–9]. As GS is a variant of FAP, it confers the same risk for colon cancer as classic FAP. It has been estimated that by the age of 30, even 50% of GS patients may experience a malignancy in the colon or the rectum. Interestingly, a risk of colon cancer is up to 100% in older individuals [10–12].

A knowledge of the genetic status has a special significance for children of GS patients. Indeed, there is no need for endoscopic screening tests in the offspring without inheritance of the faulty APC gene. In those families where a mutation cannot be identified, the only way to identify the affected individuals is by bowel screening. It is suggested to start with such screening colonoscopy from the age of 10–12 in all relatives with a 50% risk of inheritance of GS. Typically, polyps develop before adolescence, however if polyps are not detectable by the age of 40, endoscopic surveillance is not further necessary [13–15].

Conflict of interest

The authors declare no conflict of interest.

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