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A clinical case of eruptive congenital xanthomatosis

Xanthomas are localised infiltrates of lipid-containing foamy histiocytes, typically found in the dermis or tendons. Xanthomas predominantly arise in the presence of lipoprotein metabolism disorders. Due to the skin manifestations, a dermatologist is often the first to diagnose this condition. The term «xanthomatosis» is used when multiple lesions are present. A distinction is made between primary xanthomatosis, associated with familial cases of hypercholesterolemia, and secondary xanthomatosis, which arises as a result of an acquired disturbance in lipid metabolism. The onset of eruptive xanthomatosis, which manifests as multiple raised rashes with signs of inflammation that often cause discomfort, may be a warning sign of acute pancreatitis and type 2 diabetes, and is usually caused by high blood triglyceride levels.

Case presentation. *Patient M.*, aged 1 month, was admitted for treatment with a raised body temperature and a widespread rash. On examination, multiple flat and papular lesions of a yellowish colour, ranging from 0.1 to 0.5 cm in diameter were observed on the skin of the face, trunk and limbs. Physical and laboratory examinations were carried out. Elevated lipid metabolism markers were detected. A family history of type V hyperlipidemia was confirmed. A diagnosis of eruptive congenital hereditary xanthomatosis was made.

Conclusions. Primary xanthomatosis is a rare familial disorder that manifests in early childhood. It presents as skin rashes against the background of abnormal lipid metabolism and may lead to the development of severe systemic complications. Timely diagnosis allows for the prevention of further disease progression and improves patients' quality of life and life expectancy.

Keywords

Xanthomatosis, lipid metabolism, skin diseases, diagnosis, skin imaging.

Xanthomas are localised infiltrates of lipid-containing foamy histiocytes, typically found in the dermis or tendons. Xanthomas predominantly arise in the presence of lipoprotein metabolism disorders. Due to the skin manifestations, a dermatologist is often the first to diagnose this condition. In fact, as specialists, we have a unique opportunity to identify early changes in lipid metabolism and prevent serious associated conditions — atherosclerotic cardiovascular diseases. Lipids that accumulate in xanthomas are predominantly free esterified cholesterol, but sometimes, to a significant extent, they may consist of other sterols and triglycerides [4].

The term «xanthomatosis» is used when multiple lesions are present. They can first appear at various ages — in childhood, adolescence or adulthood — and multiple rashes are rarely encountered in clinical practice. A distinction is made between primary xanthomatosis, associated with familial cases of hypercholesterolemia, and secondary xanthomatosis, which arises as a result of an acquired disturbance in lipid metabolism [2, 3]. In clinical practice, there are also sporadic cases of xanthomatosis in patients with normal blood lipid levels [1, 6]. The onset of eruptive xanthomatosis, which manifests as multiple raised rashes with signs of inflammation that often cause discomfort, may be a warning sign of acute pancreatitis and type 2 diabetes, and is usually caused by high blood triglyceride levels.

Primary xanthomatosis is a rare hereditary condition that occurs in newborns or manifests in early life against the background of hyperlipidemia, high blood levels of triglycerides and chylomicrons. The efflorescences are flat, nodular, round or elongated, 2—3 cm in diameter, with relatively distinct borders, and yellow or brown in colour. They are distributed across the entire skin surface, most commonly on the buttocks, knees and elbows. As blood parameters normalise, the number of lesions decreases, and some

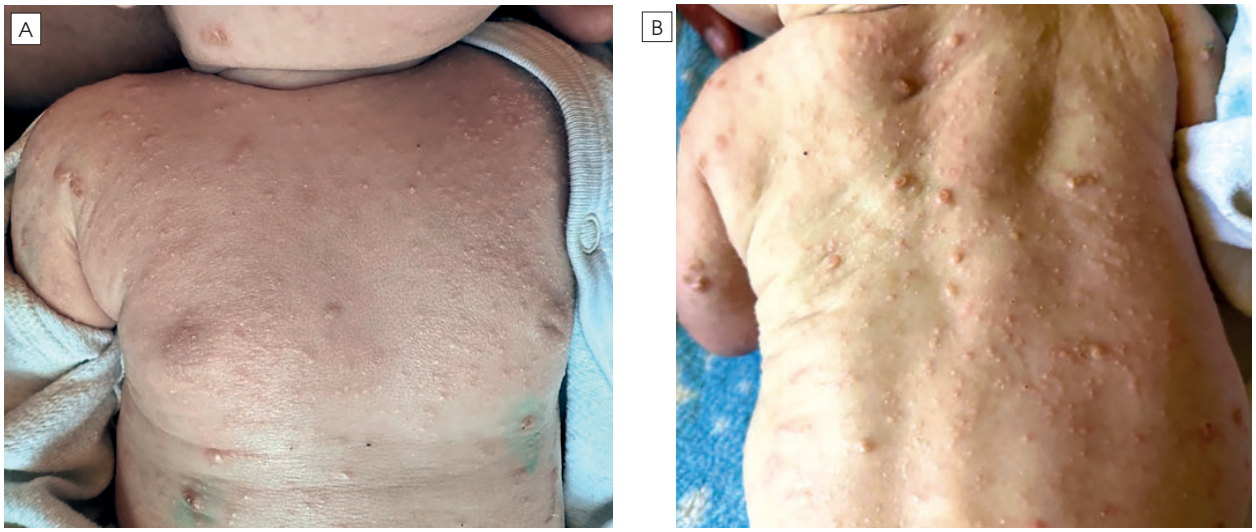

Fig. 1. ***Patient M.*** **with multiple flat and nodular xanthomas**
A — skin of the trunk; B — skin of the back.

of them disappear. We present a case of primary xanthomatosis arising against the background of familial hypercholesterolaemia.

Case presentation

Patient M., aged 1 month and 17 days, was admitted to the regional children's hospital following his mother's reports of a fever up to 39 °C, lethargy, and a rash on the face, trunk and limbs. The boy had been unwell for a month when isolated lesions began to appear; a widespread rash developed against the background of a fever. The child had previously been treated at the district children's hospital with a diagnosis of vesiculopustulosis and anemia, and had been prescribed antibiotic therapy. On the third day of his stay, his hemoglobin level rose from 110 g/l to 280 g/l, which led to his transfer to the next level of medical care.

His mother's first pregnancy was complicated by furunculosis. This was her first delivery; labour induction was performed at 41 weeks. Birth weight and height were within normal limits. The early neonatal period was uneventful. From the first days of life, multiple cases of neonatal acne and milia were noted on the face. A family history revealed a similar condition in the child's father's third cousin; type V familial hyperlipidemia was confirmed, which manifested at the age of four.

On examination, the child's general condition is serious. The response to examination is appropriate. Visual and auditory focus is present. Spontaneous motor activity is reduced. Muscle dystonia, hyperreflexia, body temperature 39.5 °C. Mucous membranes are pale, skin has a greyish tinge and is pasty. Multiple flat and nodular xanthomas are present on the skin of the face, trunk and limbs, ranging from 0.1 to 0.5 cm in diameter, yellowish in colour, some reddish-blue, and some covered with hemorrhagic crusts and scales (Fig. 1).

Dermoscopy of the rashes was performed. Microscopically — multiple elements of varying sizes, whitish-yellow in colour; focal, indistinct vascular inclusions are present; some of the lesions are covered with scaling (Fig. 2).

The abdomen is distended and tender on palpation. Urination is unimpeded and adequate. Stools are yellow in colour, homogeneous, and free of foreign matter. No abnormalities were detected on chest X-ray of the lungs or heart. ECG and echocardiogram are normal for the patient's age. Abdominal ultrasound: the liver is in its normal position, protruding 1.5 cm below the costal margin; the right lobe measures 6.6 mm, the left lobe 3—4 mm. The parenchyma shows slightly increased echogenicity; the bile ducts are not thickened; the gallbladder is contracted. Pancreas, kidneys and adrenal glands show no pathological changes. The spleen is slightly enlarged, with a homogeneous structure. Neurosonography is within normal limits.

Complete blood count: marked anemia (86 g/l), with a reduced red blood cell count ($2.6 \cdot 10^{12}$/l), eosinophilia (23 %), erythrocyte sedimentation rate (ESR) 26 mm/h. Biochemical analysis: elevated C-reactive protein (18.5 mg/l), elevated bilirubin, GGT 370 units per litre, LDH 454 U/l (hepatocellular damage, cellular breakdown). Lipid profile: triglycerides 6.47 mmol/l — markedly elevated, cholesterol 5.4 mmol/l, LDL-cholesterol 2.97 mmol/l — also elevated. Urinalysis and stool analysis showed no abnormalities.

The boy was examined by an ophthalmologist and an endocrinologist: no abnormalities were

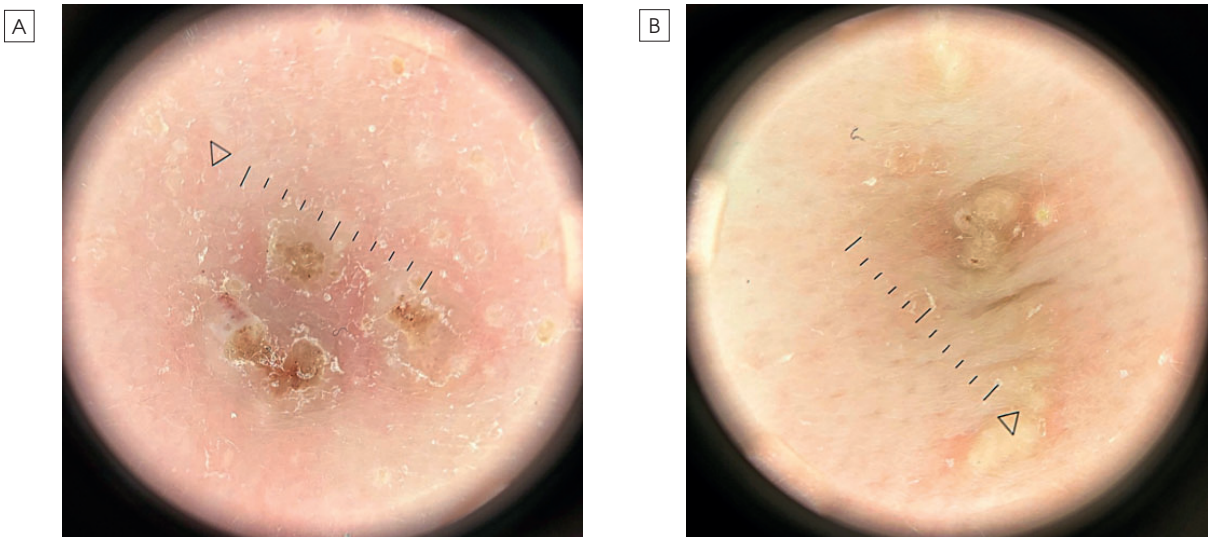

Fig. 2. ***Patient M.*** **Dermoscopic images captured in polarised mode at 10 × magnification**
A — papular elements with blood-filled crusts and scales and irritation of the surrounding tissues; B — yellowish papules with isolated vascular inclusions.

found. He was referred for consultation to our clinic. Based on the medical history, clinical findings and laboratory results, the boy was diagnosed with eruptive congenital hereditary xanthomatosis against the background of familial hypertriglyceridemia. To confirm the diagnosis and adjust treatment, the patient was referred to the Okhmatdyt National Children's Specialized Hospital. The child remained there for one month, during which the diagnosis was confirmed clinically and histopathologically. PCR testing for EBV, CMV and HAV-G was negative. Infusion therapy with glucose-saline solutions was administered. Antibiotic and antifungal therapy, gastroprotective agents, vitamins and lipid-lowering agents were prescribed. Topical glucocorticosteroids and emollients were applied to the rashes. The boy was discharged with significant improvement; the number of rashes decreased, and no new ones appeared during treatment. It was recommended to continue a low-fat diet, lipid-lowering agents and vitamins. Topical therapy, as previously prescribed, was continued. The patient remains under the supervision of a pediatrician and a dermatologist.

Conclusions

Primary xanthomatosis is a rare hereditary condition that manifests in early childhood. It presents as skin rashes, against the background of abnormal lipid metabolism, and can lead to the development of severe systemic complications. Timely diagnosis, based on awareness of the condition among a wide range of specialists and the use of additional diagnostic methods, in particular dermoscopy of the rashes, and correction of biochemical abnormalities, enables the prevention of further progression of the disease and improves patients' quality of life and life expectancy.

There is no conflict of interest.
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Клінічний випадок еруптивного вродженого ксантоматозу

Ксантоми — це локалізовані інфільтрати гістіоцитарних пінистих клітин, що містять ліпіди, які зазвичай наявні в дермі чи сухожиллях. Ксантоми виникають переважно у разі порушень обміну ліпопротеїнів. З огляду на прояви на шкірі дерматолог часто першим виявляє цю проблему. За наявності множинних елементів використовують термін «ксантоматоз». Розрізняють первинний ксантоматоз, пов'язаний із сімейними випадками гіперхолестеролемії, і вторинний, що виникає внаслідок набутого порушення фізіології ліпідного обміну. Поява еруптивного ксантоматозу, що проявляється множинними припіднятими висипаннями з ознаками запалення, які часто спричиняють неприємні відчуття, може бути первинною ознакою гострого панкреатиту і цукрового діабету 2-го типу та зазвичай виникає через високий рівень тригліцеридів у крові.

Презентація випадку. *Хворий М.*, 1 міс, госпіталізований з підвищеною температурою тіла та поширеним висипом. Об'єктивно на шкірі обличчя, тулуба і кінцівок помітні множинні плоскі та горбикові елементи діаметром від 0,1 до 0,5 см, що мали жовтуватий колір. Проведено фізикальні та лабораторні обстеження, за результатами яких виявлено високі показники ліпідного обміну. Відповідно до даних генеалогічного анамнезу верифіковано сімейну гіперліпідемію V типу. Встановлено діагноз еруптивного вродженого успадкованого ксантоматозу.

Висновки. Первинний ксантоматоз — рідкісна сімейна хвороба, що маніфестує в ранньому віці. Проявляється появою висипань на шкірі на тлі порушення ліпідного обміну та може призводити до розвитку тяжких системних ускладнень. Вчасна діагностика дає змогу вжити профілактичних заходів, щоб запобігти подальшому розвитку хвороби, а також покращити якість та збільшити тривалість життя пацієнтів.

Ключові слова: ксантоматоз, метаболізм ліпідів, хвороби шкіри, діагностика, візуалізація шкіри.

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