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# Congenital lymphedema is not only an aesthetic problem (clinical cases)

Lymphedema is a chronic disease of the lymphatic system that can be associated with genetic mutations or iatrogenic (medically induced) effects. It results in structural changes in the lymphatic vessels, leading to obstruction and impaired lymph circulation. The progressive swelling of tissues manifests as enlargement of certain body areas, most often the lower limbs, primarily in people of working age. This condition significantly reduces the patient's quality of life due to functional limitations and psychosocial consequences.

**Objective** — to present clinical cases of congenital lymphedema in individuals of different ages and to summarize the available information on this pathology in order to increase clinicians' awareness. To highlight the tactics of a dermatologist and emphasize the importance of interdisciplinary cooperation in the management of such patients.

**Materials and methods.** We described clinical cases of congenital lymphedema in girls aged 16 and 12, and a 34-year-old woman. Common complaints were: onset of the disease without apparent cause, gradually increasing swelling, impaired motor function, pronounced aesthetic defect, and depressed mood.

**Results and discussion.** The most well-known forms of congenital lymphedema are Milroy disease and Meige disease, which have a genetic basis. In the differential diagnosis with acquired forms, it is important to consider family history, the onset of the disease, and its association with trauma or infection. Lymphoscintigraphy is considered the gold standard for diagnosing lymphedema, as it allows detection of aplasia or hypoplasia of lymphatic structures in congenital forms and, in acquired forms, obstruction or damage to them.

**Conclusions.** Lymphedema is a complex, chronic condition that involves more than simple swelling, and it requires continuous care and the coordinated involvement of various specialists. Patients often seek medical help primarily due to visible changes in their body shape and skin appearance. In this setting, the dermatologist's role extends beyond identifying skin alterations to guiding further management, ensuring proper skin care, and preventing infections such as mycoses, erysipelas, and cellulitis. Essential care components include daily hygiene with neutral-pH cleansers, intensive moisturizers to restore the skin barrier, and timely referral to physiotherapists, rehabilitation experts, or vascular surgeons for comprehensive therapy. Treatment strategies range from conservative measures — manual or device-assisted lymphatic drainage, compression therapy, and medication — to surgical options like lymph node transplantation or lymphovenous anastomosis, especially effective in early stages of the disease.

**Keywords**

Congenital lymphedema, aesthetic defect, clinical cases, clinical presentations, differential diagnosis, methods of treatment, role of dermatologists in the management of lymphedema.

Lymphedema (also known as elephantiasis) is a pathological condition characterized by the enlargement of particular body parts due to chronic progressive swelling caused by impaired lymphatic drainage [10]. Despite complex etiopathogenetic mechanisms, the primary impact is on the aesthetic function of the skin and the affected body areas. This disease has a profound impact on the patient's quality of life, leading to significant functional limitations and psychosocial consequences. Therefore, it is a consequential medical and social problem. The patient's age also underscores the importance of this. Most of them are of working age [8].

According to the World Health Organization (WHO), approximately 140 million people worldwide suffer from lymphedema, which, in its late stages, can lead to elephantiasis. Lymphatic filariasis, which is the leading cause of secondary lymphedema, affects about 100 million people, and over 882 million people in 44 countries remain at risk of

infection. In the developed countries, among women, the most common cause of lymphedema is upper limb lymphedema following mastectomy.

Depending on the causative factors, lymphedema is classified as primary (congenital) or secondary (acquired). The congenital type has a clear molecular and genetic basis, unlike the acquired form, which results from mechanical damage to the lymphatic vessels. The pathological process is more often unilateral. Significant swelling leads to deformity of the lower limb, less commonly the upper limb, face, large labia in women, or scrotum in men, and impairs their function.It limits precision and range of motion, potentially leading to a permanent loss of working capacity. Patients often complain of a feeling of heaviness and distension in the affected area. In addition, there is an increased risk of secondary infections (streptococcal lymphangitis, phlegmon, abscess), which can be life-threatening. Unfortunately, at later stages, comorbid life-threatening diseases of the heart, kidneys, and liver often develop [2, 8].

Regarding the psychosocial consequences, deformation of the limbs, genital organs, or face is a cosmetic defect that can cause psychological discomfort, social isolation, and decreased self-esteem. At the stage of decompensation, patients lose the ability to lead a full-fledged lifestyle.

Although this problem is considered surgical, such patients often turn to a dermatologist. Focusing on the skin, the manifestations of lymphedema progress through distinct stages. In the early stage, the skin is smooth, elastic, and soft, with no indentation left when pressed. As it progresses, hyperkeratosis and papillomatosis develop, causing the skin to become rough and thickened, with folds and wart-like growths appearing («elephant skin» symptom). Over time, the skin and subcutaneous tissue harden due to fibrosis, and the swelling becomes irreversible, with the development of trophic changes and ulcer formation. Skin changes such as hyperpigmentation also appear, giving it a mottled appearance.

Moreover, small lymphatic vesicles or blisters may form, from which lymph leaks. A patognomonic sign when the lower limb is affected is Stemmer's sign: the inability to pinch the skin on the dorsal side of the second toe [2, 6, 10].

The lymphatic system plays a crucial role in immune defense. Lymph stagnation creates an ideal environment for bacterial growth. A damaged skin barrier, due to cracks or ulcers, serves as a «gateway» for infection (mostly *Streptococcus*). The primary dangers associated with elephantiasis is the risk of infection, the development of recurrent cellulitis or erysipelas, and the potential for eczematization. It, in turn, further damages the lymphatic vessels, creating a vicious cycle of elephantiasis progression.

Since lymphedema is a chronic disease, treatment is often largely ineffective. Currently, most researchers assert that the success of treatment depends on the early and accurate differentiation of the form of lymphedema. A multidisciplinary approach is crucial. Coordinated collaboration among surgeons, rehabilitation specialists, dermatologists, and other relevant professionals is essential. The treatment goal is to control symptoms, reduce swelling, restore mobility, prevent complications, and improve quality of life. The effectiveness of treatment depends on the stage of the disease. In the early stages, when the skin is soft and irreversible changes have not yet occurred, therapy yields good results [1].

**Objective** — to present clinical cases of congenital lymphedema in individuals of different ages and to summarize the available information on this pathology in order to increase clinicians' awareness. To highlight the tactics of a dermatologist and emphasize the importance of interdisciplinary cooperation in the management of such patients.

## Materials and methods

**Clinical Case 1.** A 16-year-old female patient, a resident of a rural area, came for a consultation regarding the enlargement of the labia majora. It causes problems with movement and hypochondria. Her mother noticed these changes since the girl was 2 years old, and there were no triggering factors. The swelling gradually increased. She did not seek medical attention for a long time, believing that the condition would normalize on its own. At the age of 14, the girl underwent surgery; however, over the past 2 years, the swelling and skin thickening have spread to the inner thighs and buttocks. According to her mother, her second pregnancy was uncomplicated, and delivery was on time. The child was born weighing 3200 g, measuring 48 cm, with no pathological changes.

On examination (Fig. 1), the labia majora are significantly enlarged, covering the vaginal cleft. The skin is of normal color, shiny in some areas, and wrinkled in certain spots, with two longitudinal atrophic scars (resulting from surgical interventions). On palpation, the skin is dense and firm; no depressions remain after pressing. The pathological process extends to the inguinal areas and buttocks. During thermometry of these areas, the local temperature is 35.4 °C. Ultrasound of the pelvic lymphatic vessels revealed aplasia of the valves. We diagnosed Milroy's disease.

Due to complicated financial circumstances, the girl's mother refused genetic counseling and consultation with a plastic surgeon. During joint examination with a phlebologist, urologist, and gynecologist, we decided to proceed with observation. Mandatory

recommendations include daily thorough hygiene of the intimate area, avoiding prolonged periods of physical inactivity, and exercises to improve drainage function. If discomfort increases or dysuric symptoms appear, surgical intervention is indicated.

**Clinical Case 2.** *Patient M.*, the mother of a 12-year-old girl, came for a consultation with complaints of marked swelling of the child's legs, which complicates walking and limb movements. The child was born full-term, weighing 3800 g, measuring 40.5 cm in length. It was the third pregnancy, and the course of pregnancy and delivery was uncomplicated. From the age of 2, the mother noticed thickening of the child's lower limbs, which she attributed to being overweight. In 10, the circumference of her legs increased, and the swelling spread to the buttocks. The mother consulted doctors, who prescribed antihistamines, diuretics, and topical angioprotective agents. However, prolonged treatment did not yield noticeable results. Moreover, a diagnosis of lymphedema was not suspected, the lymphatic vessels were not thoroughly examined, and medical genetic counseling was not recommended.

Upon examination, the lower limbs (Fig. 2) were enlarged, with unchanged skin color, a swollen and firm texture, and a tender response to palpation, while maintaining preserved tactile and temperature sensation. Electrothermography of the lower limb skin showed a local temperature of 35.5 °C. Ultrasound of the lower limb vessels, ECG, echocardiography — no pathological changes detected. The complete blood count, urine, and blood biochemistry analyses results, except for slightly elevated cholesterol levels, are within normal limits. After consultation with a vascular surgeon and geneticist, we diagnosed congenital early lymphedema. Recommended: periodic diuretics, Detralex 1000 daily for 2—3 months, moderate exercises, lymphatic drainage device pneumatic compression PULSTAR S2 weekly.

**Clinical Case 3.** *Patient K.*, a 34-year-old, was referred for consultation by a family doctor. She complains of excessive weight, attributed to swelling of the skin on her lower limbs, buttocks, and lower back. She has been suffering since early childhood. According to her mother, she was born weighing 5.2 kg with a height of 47.5 cm and had thick legs. Over time, during puberty, the skin swelling increased, becoming more pronounced on the right side, making movement difficult and causing discomfort. The patient denies any injuries.

On examination (Fig. 3): weight 135 kg, height 162 cm. The patient moves with the help of crutches and is in a depressed and irritable state due to severe psycho-emotional discomfort. Both lower limbs are significantly enlarged in volume. Above the right popliteal fossa, a round, firm-elastic formation, up to 15 cm in diameter, hangs medially. The lumbar region is deformed. The skin shows no signs of integrity violation but is dry, firm, and patchily pigmented. Sensation is preserved. Ultrasound of the vessels shows normal arterial and venous blood flow. Diagnosis: Meige disease. We refer the patient to a vascular surgeon.

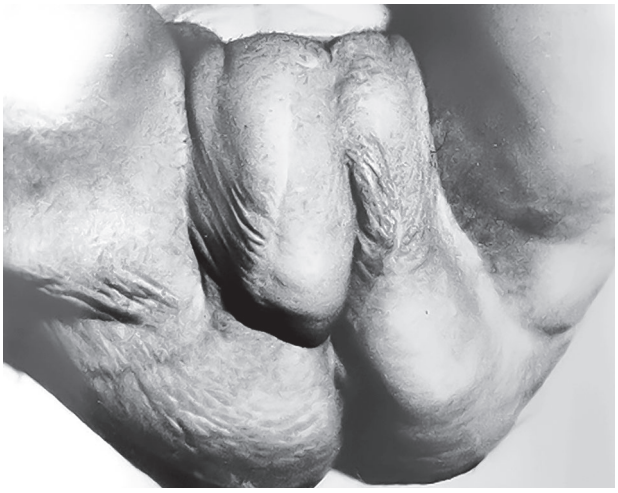

Fig. 1. **Significant enlargement of the labia majora, that covers the vaginal cleft**

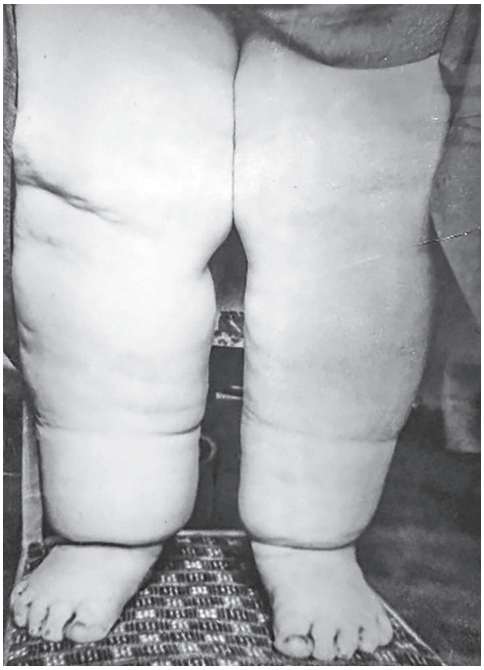

Fig. 2. **The lower limbs are sharply enlarged in volume. The skin color is unchanged**

## Results and discussion

Primary lymphedema is associated with intrauterine developmental disorders of the lymphatic system. Genetic mutations are mainly concentrated in genes that encode growth factors and receptors necessary for the development of lymphatic vessels. As a result,

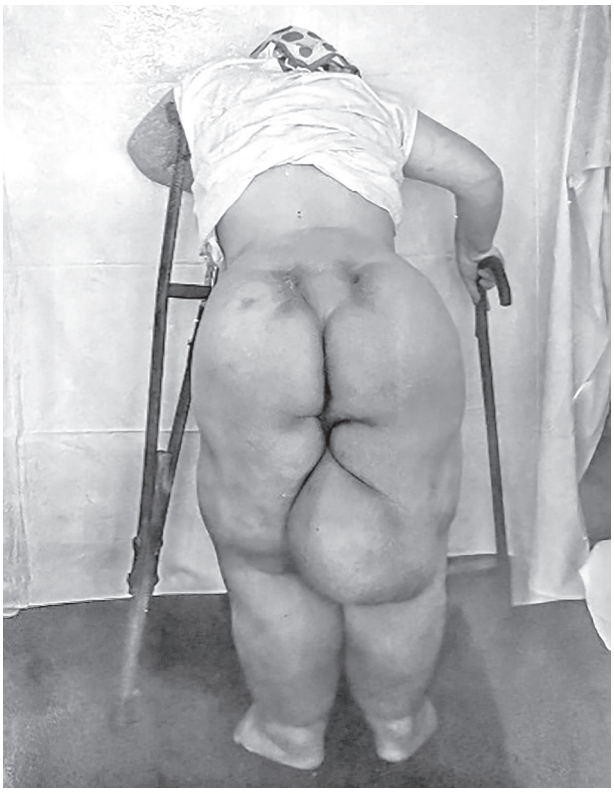

Fig. 3. **The patient is leaning on crutches. Both lower limbs are enlarged. On the right, a rounded 15 cm mass protrudes medially above the popliteal fossa**

this leads to aplasia, hypoplasia, or dysplasia. Depending on the age of manifestation, early (praecox) congenital lymphedema typically appears at birth or by the age of 35, whereas late (tarda) congenital lymphedema typically occurs after 35 years.

The most common congenital form is Milroy's disease. It is often caused by a mutation in the FLT4 gene (FMS-like tyrosine kinase 4), which encodes the VEGFR-3 tyrosine kinase receptor (vascular endothelial growth factor receptor 3). This receptor is critical for lymphangiogenesis during embryonic development. A mutation leads to impaired formation of valves and lymphatic vessel collectors. The inheritance type is autosomal dominant.

Another manifestation of genetic changes is Meige's disease (lymphedema praecox), which begins to develop during adolescence. The disease is inherited in an autosomal dominant manner. It is associated with mutations in the FOXC2 gene (Forkhead box C2), which leads to improper formation or insufficiency of valves in the lymphatic vessels. It causes lymph reflux and progressive swelling.

Additionally, lymphedema can be part of complex syndromes. For example, in Turner syndrome, in addition to swelling of the feet and/or hands at birth, an extra skin fold on the neck is observed. In Noonan syndrome, neck lymphedema, short stature, and heart defects are noted. Limb lymphedema can also be associated with ptosis and deafness in cases of GATA mutations.

In diagnostics, genetic testing and lymphoscintigraphy (as the gold standard for visualizing disorders) will be helpful.

Congenital lymphedema should be distinguished from acquired lymphedema. A detailed history will show a clear connection with surgery, trauma, or infection in the acquired form. In the congenital form, early onset and family history are crucial.

Secondary (acquired) lymphedema, is associated with external and iatrogenic causes that lead to damage or obstruction of previously normally functioning lymphatic pathways. Among iatrogenic causes in developed countries, the most common are lymph node dissections and radiation therapy for oncological diseases. Removal of lymph nodes in breast cancer leads to the development of elephantiasis of the upper limb on the affected side [3]. In tropical regions, the primary cause of acquired elephantiasis is parasitic invasion-filariasis, which is caused by the nematode Wuchereria bancrofti [2].

Another factor of lymphedema may be trauma or chronic varicose insufficiency. The disease begins with slight swelling, which gradually increases in width as the lymphatic vessels dilatate and become more dense. Fibrotic and polycystic changes in the skin often lead to its necrosis. Recurrent erysipelas (erysipeloid) can also lead to fibrosis and limb enlargement.

Diagnostic lymphoscintigraphy will reveal the absence or marked reduction in the number of lymphatic vessels in congenital lymphedema, as well as obstruction or damage to lymphatic vessels in a specific area in acquired cases. Soft tissue ultrasound will provide an assessment of fibrosis, skin thickness, and rule out deep vein thrombosis [1, 2, 6, 8].

## Conclusions

Lymphedema is not just a swelling. It is a complex disorder that requires constant care and coordinated actions of a multidisciplinary team. The distorted aesthetic appearance of certain areas of the body is the primary reason patients visit a doctor.

### *Dermatologist's algorithm for suspecting lymphedema*

The dermatologist plays a role not only as a diagostician of skin changes but also as a coordinator of further treatment and prevention of complications. The primary directions for managing skin manifestations of the disease are:

1. Skin care. Daily hygiene involves using soap with a neutral pH. To restore the skin's barrier function, including hydration and prevention of

cracks, intensely moisturizing lotions with low pH are recommended.
2. Timely diagnosis and treatment of onychomycosis and foot mycoses, erysipelas, or cellulitis, which are familiar companions to this condition.
3. Reference and Coordination. The dermatologist refers the patient to key specialists for comprehensive therapy. A physiotherapist and rehabilitation specialist will develop a comprehensive anti-stasis therapy program [4, 7]. A vascular surgeon will determine the advisability of surgical intervention.

Comprehensive anti-stasis therapy, a conservative treatment method, encompasses manual or device-assisted lymphatic drainage massage, the use of compression garments, skin care, and specialized physical exercises [9]. Medications to improve lymphatic drainage, phlebotonics, and diuretics also have significant therapeutic value. Modern surgical treatment methods include lymph node transplantation and the creation of lymphovenous anastomoses to restore lymphatic drainage [10]. These methods are particularly effective in the early stages of both congenital and acquired lymphedema.

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# Вроджена лімфедема — це не тільки естетична проблема (клінічні випадки)

Лімфедема — хронічне захворювання лімфатичної системи, зумовлене генетичними мутаціями чи ятрогенним впливом. Наслідком лімфедеми є структурні зміни лімфатичних судин, що призводять до обтурації та порушення циркуляції лімфи. Наростаючий набряк тканин проявляється збільшенням окремих ділянок тіла, найчастіше нижніх кінцівок, переважно в осіб працездатного віку. Цей стан різко знижує якість життя пацієнтів через функціональні обмеження та психосоціальні наслідки.

**Мета роботи** — представити клінічні випадки вродженої лімфедеми в осіб різного віку та узагальнити наявну інформацію щодо цієї патології для підвищення обізнаності клініцистів. Висвітлити тактику дерматолога та підкреслити важливість міждисциплінарної співпраці у веденні таких пацієнтів.

**Матеріали та методи.** Описано клінічні випадки вродженої лімфедеми у дівчат віком 16 і 12 років та жінки 34 років. Спільними скаргами були: початок захворювання без видимих причин, набряк, що поступово наростав, порушення рухової функції, виражений естетичний дефект та пригнічений психологічний стан.

**Результати та обговорення.** Найвідомішими формами вродженої лімфедеми є хвороба Мілроя та хвороба Мейджа, що мають генетичне підґрунтя. Проводячи диференційну діагностику з набутими формами, важливо враховувати сімейний анамнез, початок захворювання та його зв’язок з травмою чи інфекцією. Лімфосцинтиграфію вважають золотим стандартом діагностики лімфедеми, оскільки вона дає змогу виявляти аплазію чи гіпоплазію структур лімфатичної системи у разі вроджених форм, а за набутих — їхню обструкцію чи пошкодження.

**Висновки.** Лімфедема — це складний хронічний стан, який характеризується не лише набряклістю, він потребує постійного догляду та координації між різними фахівцями. Пацієнти часто звертаються по медичну допомогу насамперед через видимі зміни форми тіла та стану шкіри. У цьому випадку роль дерматолога виходить за межі тільки виявлення змін на шкірі — він також визначає подальше лікування, допомагаючи забезпечувати належний догляд за шкірою та запобігати розвитку інфекцій, таких як мікози та бешиха, а також целюліту. Основні компоненти догляду включають щоденну гігієну з використанням засобів із нейтральним pH, інтенсивне зволоження для відновлення бар'єрної функції шкіри, а також своєчасне направлення до фізіотерапевтів, реабілітологів або судинних хірургів для проведення комплексної терапії. Стратегії лікування варіюються від консервативних заходів (ручного або апаратного лімфатичного дренажу, компресійної терапії та призначення препаратів) до хірургічних втручань, таких як трансплантація лімфатичних вузлів або встановлення лімфовенозних анастомозів, що особливо ефективно на ранніх стадіях захворювання.

**Ключові слова:** лімфедема вроджена, естетична проблема, клінічні випадки, клінічні прояви, диференційна діагностика, методи лікування, роль дерматолога у веденні лімфедеми.

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