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Dercum's Disease (Neurolipomatosis) as Concomitant Disease in Autopsy Practice

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Abstract: The article provides an overview of the associated disease in autopsy practice known as Dercum's disease or neurolipomatosis. This article reviews the clinical and pathological aspects of this disease, including its prevalence, symptoms, and tumor characteristics. Particular attention is paid to diagnostic and treatment methods, as well as their influence on autopsy results.

Keywords: Dercum's disease, neurolipomatosis, autopsy practice, tumors, diagnosis, treatment.

The relevance of the research. The relevance of the article on "Dercum's Disease (neural lipomatosis)" in autopsy practice is due to the need for a deeper understanding of this rare condition, its clinical manifestations, pathological features, diagnostic methods, and its impact on autopsy results.

Neural lipomatosis (Dercum's disease) is a rare condition of unknown etiology, characterized by subcutaneous fat deposits with various locations throughout the body. Histologically, these deposits consist of lipomas and are associated with excess weight or obesity, as well as various mental disorders (anxiety, depression, sleep disturbances). The classification of Dercum's disease is related to the size and location of the fat nodules (general diffuse, general nodular, localized nodular, and juxta-articular forms). Diagnosis is based on the clinical picture and the exclusion of several other diseases associated with lipomas. There is no universally accepted approach to treating patients. In some cases, successful liposuction or lidocaine application has been reported. Other treatment methods have also been reported, but their effectiveness is based only on anecdotal descriptions and has not been confirmed in clinical studies. Dercum's Disease, also known as neural lipomatosis, is a rare condition that is typically characterized by the presence of painful subcutaneous fat deposits of varying sizes, multiplicity, and localization. Associated symptoms include excess weight or obesity, fatigue or weakness, and a range of psychological manifestations (such as sleep disturbances, emotional instability, depression, and anxiety), although they are not always present in the clinical picture of the disease. The etiology of Dercum's disease remains unknown, and the classification of the disease, diagnostic criteria, and treatment strategy are subject to debate. The disease was first described by American neurologist Francis Xavier Dercum (1856–1931) [1]. He published two articles on the disease in 1888 [2] and 1892, using the term "adipose dolorosa." The disease was also described in Philadelphia by American physician James Mesther Anders (1854–1936) and British physician and medical biographer Sir William Hale White (1857–1949) from Guy's Hospital in London. In addition to neural lipomatosis, several synonyms for Dercum's disease have appeared in the medical literature. These include: adipose dolorosa, lipomatosis dolorosa, adipose rheumatism, lipalgia, and Anders syndrome. Due to its unclear etiology and lack of a clear definition, Dercum's disease is also known as Dercum's syndrome. Dercum's disease has been recognized by the World Health Organization in the ICD-10 and is classified as lipomatosis, not elsewhere classified (IV - E88.2). Orphanet and the National Organization for Rare Disorders have also included this condition in their lists. The total number of publications on Dercum's disease is estimated at approximately 140-160 references. The majority of these are case reports, and only a few articles provide patient analyses. An excellent review was published in 2012 by Hansson et al., which includes proposed classification criteria. This review aims to summarize the latest data on this disease with a focus on clinical presentation, differential diagnosis, and treatment.

Dercum's disease is a rare disorder, almost exclusively occurring in adults and predominating in individuals aged 35 to 50 years. Very few cases of the disease in children have been reported. It is predominantly a disease affecting women, with the female-to-male ratio estimated to be in the range of 5 to 30:1. Most reported cases involve patients of Caucasian race.

Some articles suggest that Dercum's disease predominantly occurs in postmenopausal women. In contrast, studies by Herbst and Asare-Bediako showed that four-fifths of female patients developed the disease before menopause. No studies on the prevalence or incidence of the disease have been conducted. Metabolic changes associated with Dercum's disease are the subject of interesting hypotheses.

Hansson and colleagues described the excess of glucocorticoids, caused by hormone intake, as a hypothetical cause of Dercum's disease. It is known that glucocorticoids cause dyslipidemia, and it is possible that Dercum's disease is related to altered lipid metabolism. Labuzek and colleagues described a case of Dercum's disease treated with metformin. This drug is primarily used in obese patients with diabetes and has various metabolic effects. They found favorable effects of the drug on adipokines, β-endorphins, and pro-inflammatory cytokines associated with pain reduction. Both metabolic and other mechanisms of action of metformin (e.g., influence on synaptic plasticity, microglial activation) have been hypothetically considered as mechanisms for pain reduction in patients. Hansson and colleagues measured various neuropeptides in the cerebrospinal fluid and plasma of 53 patients with Dercum's disease. They found a lower level of substance P in the cerebrospinal fluid of patients. The level of neuropeptide Y was also borderline decreased, while some increase in the level of β-endorphin in the cerebrospinal fluid of patients was detected. The role of these changes in the pathophysiology remains unknown. Kampen and colleagues proposed an inflammatory pathomechanism, possibly leading to the release of painful neuropeptides. The role of trauma was suggested in some patients. The role of impaired lymphatic vessel function was also hypothesized. According to Rasmussen and colleagues, impaired lymphovascular function leads to a deficit in lymphatic transport, and lipomas apparently formed due to dysfunction in lymphatic drainage. Medical literature contains a significant number of atypical manifestations of Dercum's disease. Some of them are described in various locations of adipose tissue deposits. It should be noted that patients with lipomas may experience chest pain [16]. Although this is very rare, it should be taken into account when diagnosing mastalgia, as well as when detecting tumors in the chest. Abdominal pain as the main symptom of the disease [17] and arthralgia as the first symptom of Dercum's disease [18] have been described in rare cases. A patient with Dercum's disease complicated by adipose tissue necrosis of the skin and septic shock was described by Haddad et al. [19]. Cavale et al. [20] described a patient with adipose formations on the scalp, with severe headaches.

Differential diagnosis of Dercum's disease is conducted with the following conditions: Fibromyalgia, Lipoedema, Lymphedema, Panniculitis, Proteus syndrome, Weber-Christian disease, Frohlich syndrome, Progressive lipodystrophy, Cushing's syndrome, Hypothyroidism, Benign symmetric lipomatosis (Madelung's syndrome, Launois-Bensaude syndrome), Familial multiple lipomatosis.

The aim of the study. The aim of the study on Dercum's disease (adiposis dolorosa) in autopsy practice is to investigate the clinical and pathological features of this rare condition, its impact on autopsy findings, and to search for more effective methods of diagnosis and treatment. This research also aims to improve understanding of this disease among medical pathologists and raise awareness among pathologists, which in turn may contribute to more accurate diagnosis and treatment of patients, as well as a more precise assessment of the cause of death in autopsy results. The study aims to summarize the latest data on Dercum's disease, including epidemiology, clinical presentation, differential diagnosis, and treatment. The article also aims to raise awareness of this rare condition and increase knowledge within the medical community.

Materials and methods. The study of articles on neurolipomatosis includes familiarizing oneself with available materials and using various research methods.

- ✓ Scientific articles published in journals and scientific publications

- ✓ Books and textbooks related to neurolipomatosis and related topics
- ✓ Clinical studies and research reports
- ✓ Conferences, symposiums, and other medical events

Research methods:

- ✓ Literature review and analysis of available articles and research on neurolipomatosis
- ✓ Systematic literature review, which includes the analysis and synthesis of results from multiple studies
- ✓ Clinical studies, including observation and analysis of the medical and disease history of patients with neurolipomatosis
- ✓ Molecular-genetic research aimed at studying the genetic basis of neurolipomatosis
- ✓ Immunological studies to investigate the immune response to neurolipomatosis
- ✓ Radiological studies, such as computed tomography (CT) and magnetic resonance imaging (MRI), for visualization and analysis of tumors
- ✓ Biopsy and histological examination of the tumor to identify and assess the characteristics of neurolipoma

These materials and research methods help to obtain a more comprehensive understanding of neurolipomatosis, its causes, manifestations, diagnosis, and possible treatment methods.

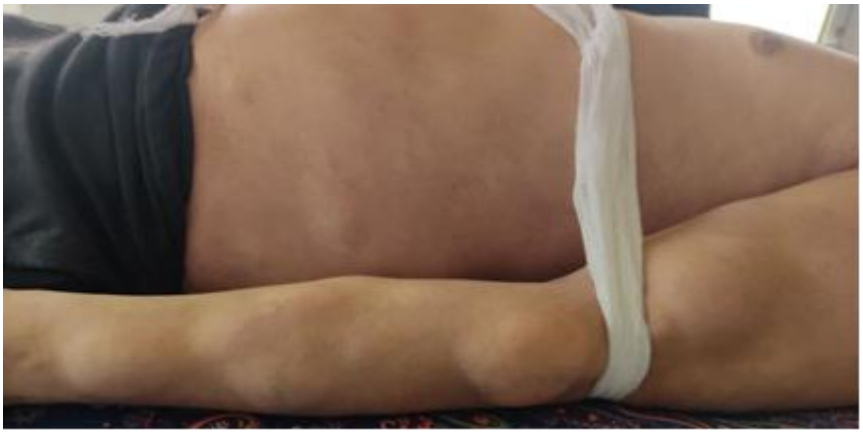

Fig.1 (a). Macroscopic appearance of neurofibromatosis.

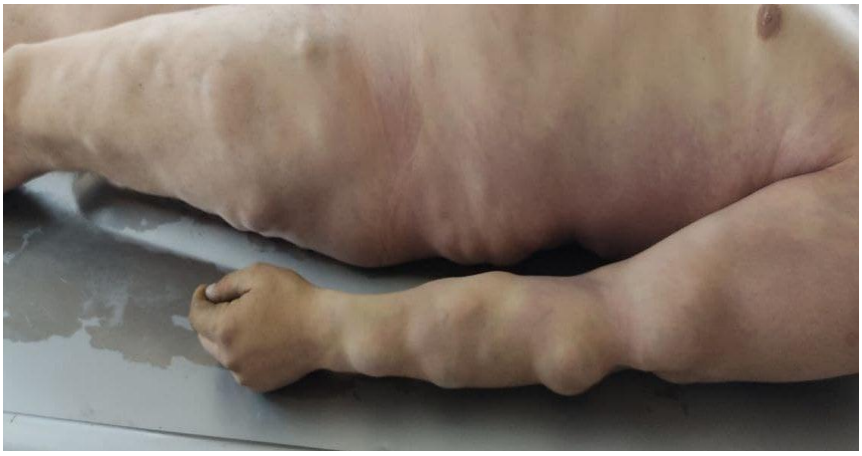

Fig.1 (b). Macroscopic appearance of neurofibromatosis.

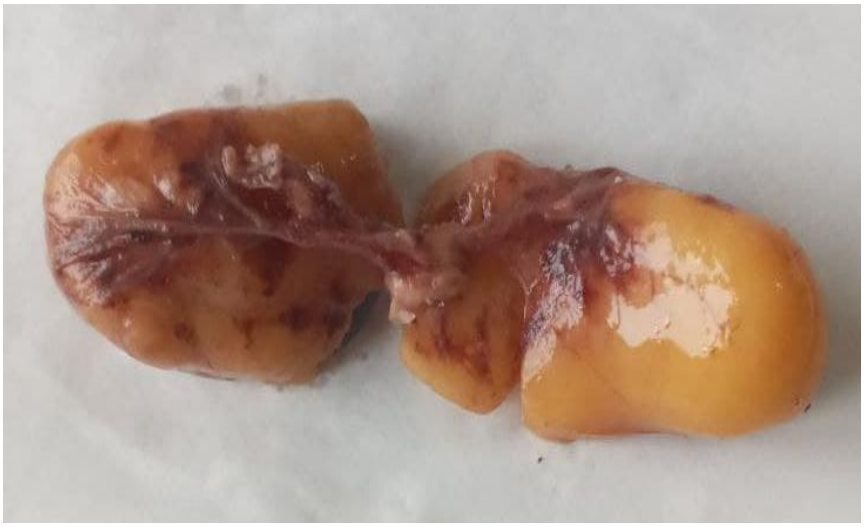

Fig.2 (a). Material obtained during autopsy.

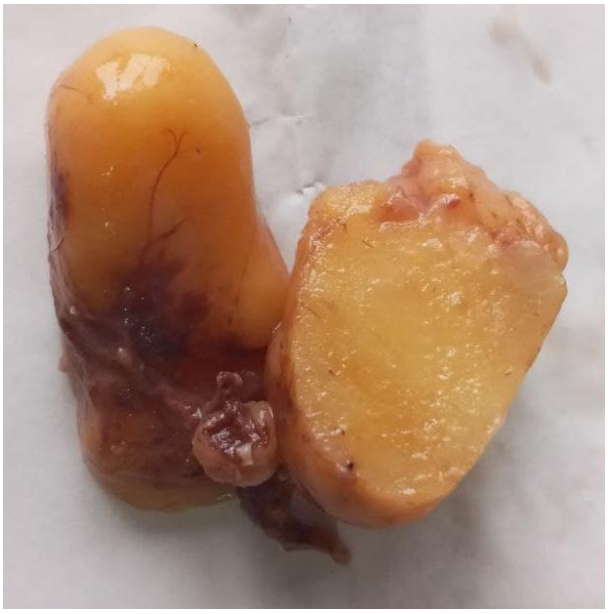

Fig.2 (b). Material obtained during autopsy.

Result and discussion. We provide more information about the disease dercum's disease as an accompanying condition in autopsy practice. We reported a case of dercum's disease in a troop during an autopsy as an accompanying condition. In the department of pathological anatomy of the pathology bureau of the Bukhara region, when examining a deceased body, an accompanying disease called neurolipomatosis was discovered, also known as dercum's disease, which occurs in 1 case out of 295 in the last 2-3 years of our autopsy practice. Subcutaneous lipomas can appear in various parts of the body and are more prevalent in the legs, arms, and torso (front or back). Approximately every fifth case affects the face, and in every third case, lipomas develop in the scalp and neck of patients with dercum's disease.

Conclusions. Neurofibromatosis is a genetic disorder that is characterized by the presence of neurofibromas, which are benign tumors that arise from the nerve sheaths. In addition to neurofibromas, other neurological symptoms such as delayed psychomotor and intellectual development, speech and motor impairments, epilepsy, and other neurological symptoms can be observed in individuals with neurofibromatosis. Morphologically, neurofibromas can contain significant amounts of fatty tissue and can vary in size and shape. Furthermore, structural anomalies can be detected in the tumors of individuals with neurofibromatosis.

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