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Opinion Article

A Short Note on Erdheim Chester Disease

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1. Introduction

Erdheim Chester Disease (ECD) is a rare multiple systemic disorder in adolescents. It is characterized by high production and accumulation of histiocytes in multiple tissues and organs. Histiocytes are large phagocytic cells (macrophages) that usually play a role in responding to infection and injury. The phagocytic cell is any "scavenger cell" that surrounds and destroys invading microorganisms or cellular debris. Related symptoms and findings and course of the disease depend on the specific location and extent of such involvement. The specific underlying cause of ECD is unknown.

ECD affects certain areas of the long bones of the legs, including the shafts (diaphysis) and the ends of the shafts (epiphysis) (*i.e.,* metaphysis). The ends of the long bones are usually avoided or there may be mild changes. Infiltration by histiocytes usually leads to extensive or conges tive increase in bone density as well as hardening (osteosclerosis) and bone hardening. In some rare cases, other bones may be involved, such as the lower jaw bone (mandible) or some of the bones in the spinal column (vertebrae). In most affected individuals, the initial symptom of the disorder is associated with bone pain, usually affecting the knees and legs, which are similar on both sides of the body (symmetrical). In some cases, more common symptoms may develop, including weight loss, fever, muscle and joint pain; and general feeling of discomfort, weakness and fatigue (sickness).

2. Description

ECD is also characterized by the involvement of the skin, the tissue behind the eyebrows (retrobulbar area); lungs; brain; the pituitary gland, the area containing the organs behind the abdominal cavity (retroperitoneum) and/or other sites. The relevant symptoms and course of the disease may vary individually, depending on the site and the degree of involvement. Some people with ECD may develop soft, yellow, fatty plaques or nodules on the eyelids (xanthelasma) or skin (catenius sintomas). In addition, the involvement of the retrobulbar region may lead to a marked protrusion of eyeballs (exophthalmos) and other features and discoveries. In those with pulmonary involvement, progressive scarring and hardening of the lung tissue (pulmonary fibrosis) can lead to dry cough, prolonged breathing (dyspnea), insufficient oxygen supply to the blood, and impaired heart ability t o pump adequately blood to lungs and other parts of the body (heart failure), and fatal complications.

In some affected individuals, there may also be an infiltration of the pituitary gland, which can lead to diabetes insipidus. It is a metabolic condition in which insufficient secretion of Antidiuretic Hormone (ADH) by the pituitary gland results in large amounts of dilute urine (polyuria) and excessive thirst (polydipsia). ADH usually reduces the amount of water lost in the urine. The pituitary gland produces many hormones, including ADH; which is regulated and connected to an area called the hypothalamus in the brain. In some rare cases, ECD is also characterized by the involvement of the lower part of the brain (brain stem) and other areas of the brain, such as the cerebellum, which are involved in coordinating voluntary movement,

balance, and posture. Associated neurological symptoms may vary from case to case. However, such abnormalities often include impaired muscle coordination (ataxia); abnormally inconsistent pattern of gait (gait); vague speech (dysarthria); and/or involuntary, rhythmic, rapid eye movements (nystagmus).

ECD is also characterized by infiltration of tissue within the retroperitoneal region (retroperitoneal fibrosis) and associated scarring. In some cases, such changes can lead to obstruction of the tubes (*i.e.,* ureters) that carry urine from the kidneys to the bladder, leading to urinary (hydronephrosis), impaired kidney (kidney) function, and abnormal inflammation of the kidneys failure. Some cases of retroperitoneal fibrosis involving the main artery (aorta) and its branch blood vessels (pediatric fibrosis) have also been described.

As mentioned above, the course of the disease varies depending on the extent of involvement outside the bone (extra social involvement) and affecting the internal organs (visceral involvement). In some cases, disease progression and related organ dysfunction can lead to potentially fatal complications such as pulmonary fibrosis, heart failure and/or kidney failure.

The exact cause of the ECD is unknown. However, the disease is thought to indicate an abnor-mal inflammatory process, accompanied by excessive proliferation and accumulation of certain cells, associated scarring or growth of fibrous connective tissue (fibrosis). Histiocytic cells, which contain large amounts of lipid (xanthomatous histiocytes) can penetrate extensively into affected tissues; some lymphocytes; and distinctive, large cells with multiple nuclei (touton giant cells). In people with ECD, these fatty, nodular (xanthogranulomatous) cell deposits can infiltrate multiple tissues and organs, leading to impaired organ function. It has been found that approximately 50% of patients with ECD are tested positive for the BRAF V600E gene muta-tion, indicating that it may be a histolytic neoplasm.

The diagnosis of ECD is made on the basis of a comprehensive clinical evaluation, detailed patient history, identification of symptomatic features and a variety of specialized tests. Such studies may include plain X-rays advanced imaging techniques including Computed Tomography (CT) scanning, Magnetic Resonance Imaging (MRI) and/or bone scan (bone scintigraphy); and/or other tests. Plain X-rays of the bone involved usually reveal symmetrically increased hardening and hardening, mainly leaving the epiphysis in the metaphysis and diaphysis, which is considered typical of ECD. In addition, the diagnosis can be confirmed by biopsy and micro-scopic evaluation of tissue samples, which exhibit the infiltration of fat (lipid) folate histocytes with distinct, large cells with some non-Langerhans cellular properties and multiple nuclei. The BRAF V600E gene mutation was found in approximately half of the patients with ECD.

3. Conclusion

Reports indicate that various treatments have been used with limited success. Such recommended measures may include the administration of corticosteroid drugs (e.g., prednisone); some chemotherapeutic drugs that control or prevent abnormal cell proliferation (e.g., vinblastine); the use of high energy rays that preferably destroy or injure rapidly expanding cells (radiation therapy); immunotherapy; and/or surgery. Further research is needed to determine the appropriate treatments for this disorder. Additional treatment for people with ECD may be symptomatic and supportive. In 2017, Zelboraf (vemurafenib) was approved by the FDA to treat specific adult patients with ECD who have the BRAF V6000 genetic mutation made by Zelboraf Hoffman-LaRoche, Inc.