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Review Article

Assessment Genetic Mutations in Gene VPS113A in Indicate Chorea-Acanthocytosis Syndrome

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Abstract

Chorea-acanthocytosis syndrome is primarily a neurodegenerative disorder that affects many parts of the body. In addition to Chorea, another common characteristic of Chorea-acanthocytosis syndrome is dystrophy in various muscles such as organs, face, mouth, tongue and throat. Chorea-acanthocytosis syndrome is caused by the mutation of the VPS13A gene, which is independent in the long arm of chromosome 9, 9q21.2.

Keywords: Chorea-Acanthocytosis Syndrome; VPS113A Gene; Blood Cells

Generalized Chorea-Acanthocytosis Syndrome

Choura-acanthocytosis syndrome is primarily a neurodegenerative disorder that affects many parts of the body. Chorea refers to the involuntary movements created by people with this disorder. People with this disease also have a strange abnormal red blood cell (acanthocytosis). This condition is one of the groups of diseases known as neurocancontosis, which includes neurological problems and abnormal red blood cells [1].

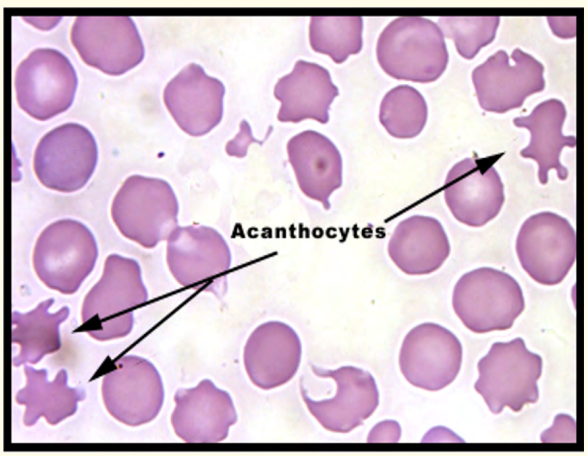


Figure 1: Microscopic Image of Stellar Abnormal Red Cells in Chorea-Acanthocytosis Syndrome.

Symptoms of Chorea-Acanthocytosis Syndrome.

In addition to Chora, another common characteristic of Choras-Acanthocytosis is dystrophy in various muscles such as organs, face, mouth, tongue and throat. These muscle shifts can cause vocal sounds, involuntary sputum, and spasm of the limbs. Eating food may also be disturbed as the language and throat can interfere with chewing and swallowing food. People with chorea-acanthocytosis may wound their lips, lips and mouths with an uncontrollable scratch. Approximately half of people with Chorea-acantocytosis have seizures [2].

People with Chorea-Acanthocytosis syndrome may experience mental problems, learning and remembering information (cognitive impairment). They may have allergies and weakness in the arms and legs (peripheral neuropathy) and muscle weakness (myopathy). Musculoskeletal and dysmorphic disorders usually cause speech problems in people with this condition and can lead to inability to speak [3].

Behavioral changes are a common feature of Chorea-Acanthocytosis syndrome and may be the first sign of this disease. These behavioral changes may include changes in personality, obsessive-compulsive disorder (OCD), self-avoidance, and inability to take care of yourself [3].

Symptoms and Symptoms of Chorea-Acanthocytosis usually begin in the early to mid-adult years. The kinetic problems of this

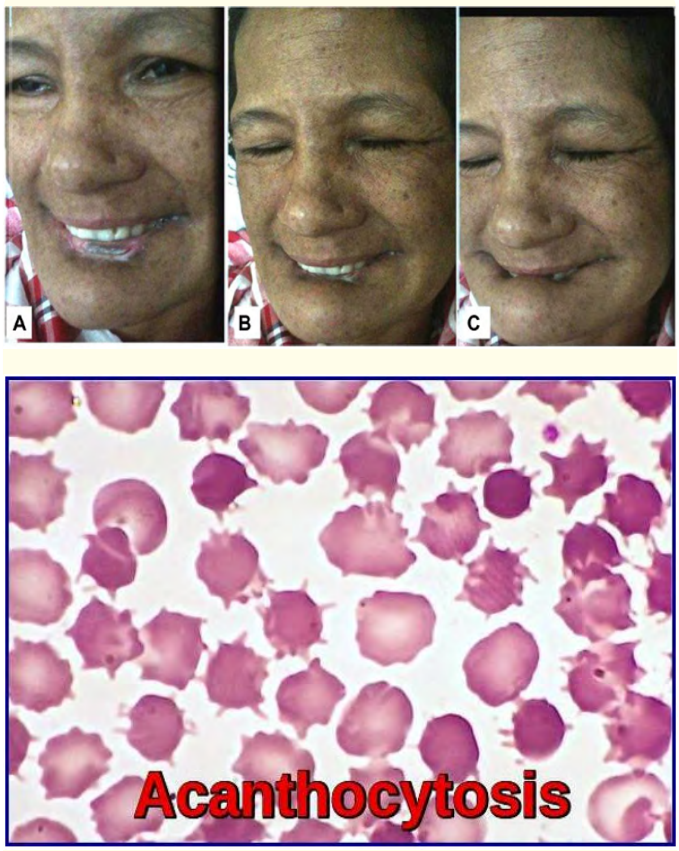


Figure 2: Another view of Chorea-Achanthocytosis syndrome disorders.Symptoms of Chorea-Acanthocytosis Syndrome.

situation increase with age. The loss of cells (atrophy) in specific regions of the brain is the main cause of neurological problems in people with Chorea-Acanthocytosis syndrome [4].

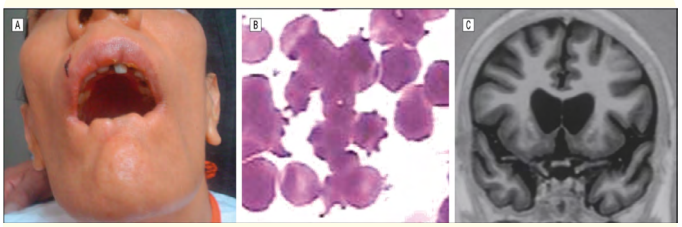


Figure 3: Images of lumbar lesion disorders (A), stellate red blood cells (B), and cerebral impairment (C) in Chorea-Acanthocytosis syndrome.

Etiology of Chorea-Acanthocytosis syndrome

Chorea-Acanthocytosis syndrome is caused by the mutation of the VPS13A gene, which is independent in the long arm of chromosome 9, 9q21.2. The VPS13A gene provides instructions for the production of a protein called chorein, whose function is unknown in the body. Some researchers believe that chorein plays an important role in intracellular protein transport. Most of the mutations in the VPS13A gene result in the production of an abnormal, nonfunctional version of the chorein protein. The VPS13A gene is active throughout the body (expressed). It is unknown why the mutation in the gene affects only the brain and red blood cells [5].

Chorea-Acanthocytosis syndrome follows an autosomal recessive hereditary pattern. Therefore, in order to create this syndrome, two versions of the mutated gene of VPS13A (one parent and one of the mother) are needed and the chance of having a child with autosomal recessive syndrome is 25% for each pregnancy [5].

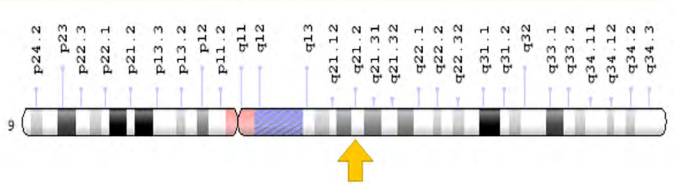


Figure 4: Schematic view of chromosome 9, in which the VPS13A gene is located in the long arm of this chromosome 9q21.2.

Frequency of Chorea-Acanthocytosis syndrome

Chorea-Acanthocytosis syndrome is a genetic disorder that is estimated to be about 500 to 1000 people in the world with this syndrome [6].

Diagnosis of Chorea-Acanthocytosis syndrome

Chorea-Acanthocytosis syndrome is diagnosed based on the clinical and clinical findings of the patients and some pathological examinations. The most accurate method for diagnosing this syndrome is the molecular genetic testing of the VPS13A gene to investigate the presence of possible mutations [7].

Therapeutic pathways for Chorea-Acanthocytosis syndrome

The strategy of treatment and management of Chorea-Acanthocytosis syndrome is symptomatic and supportive. Treatment may be carried out by a team of experts, including orthopedic surgeons, neurologists, psychiatrists and other health care professionals. There is no valid treatment for this syndrome, and all clinical interventions are designed to reduce the suffering of the sufferers. Genetic counseling is also important for all parents who want a healthy baby [8].

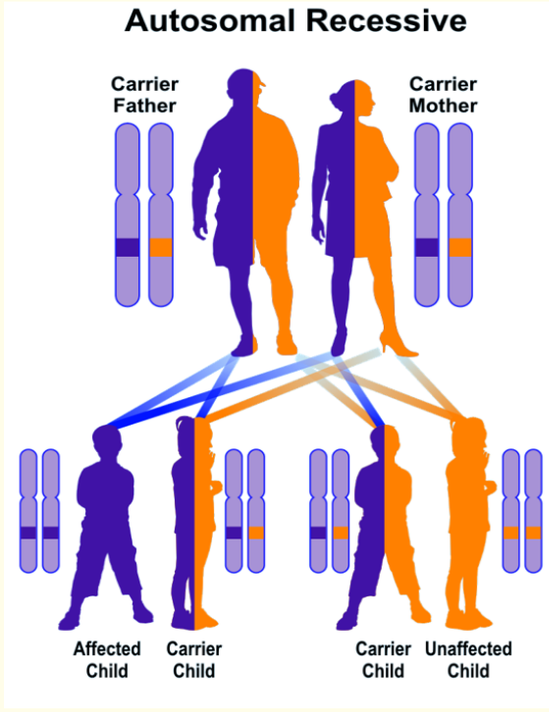
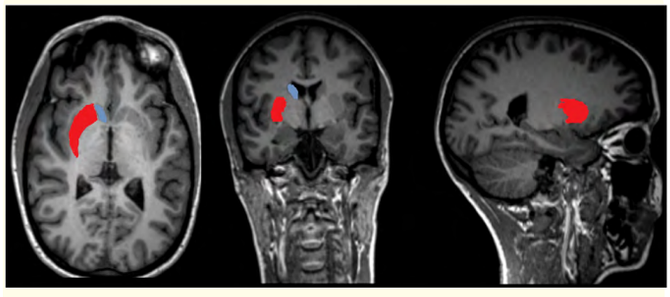


Figure 5: Schematic view of an autosomal recessive hereditary pattern that follows Chorea-Acanthocytosis syndrome.



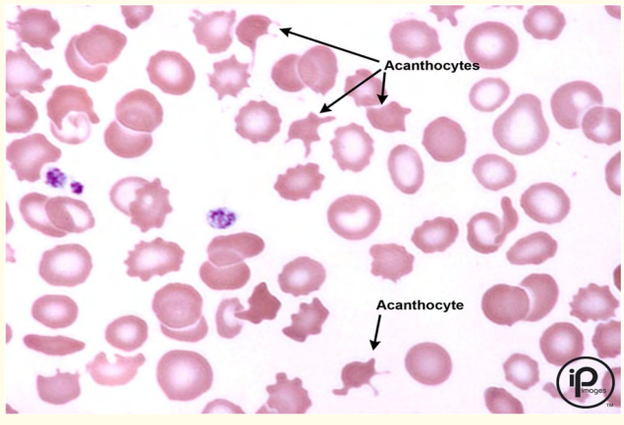


Figure 6: Radiological imaging of cerebrovascular and microscopic imaging of the star-shaped red blood cell in Chorea-Acanthocytosis syndrome.

Discussion and Conclusion

Choura-Acanthocytosis syndrome is primarily a neurodegenerative disorder that affects many parts of the body. In addition to Chorea, another common characteristic of Chorea-Acanthocytosis is dystrophy in various muscles such as organs, face, mouth, tongue and throat. Chorea-Acanthocytosis syndrome is caused by the mutation of the VPS13A gene, which is independent in the long arm of chromosome 9, 9q21.2. There is no valid treatment for this syndrome, and all clinical interventions are designed to reduce the suffering of the sufferers. Genetic counseling is also important for all parents who want a healthy baby.

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