LATE CUTANEOUS FORM OF PORPHYRIA CUTANEA TARDA IN 26-YEAR-OLD MAN: CLINICAL CASE REPORT

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Relevance. Porphyria cutanea tarda (PCT) is a rare form of enzymatic defect disorder that occurs due to deficiency of an enzyme called Uroporphyrinogen decarboxylase (UROD). Well-known risk factors for PCT is mainly HCV infection and alcohol abuse.

Target. To provide information about this rare form of disease for clinicians for better understanding and early diagnosis, treatment and prevent severe complications.

Research methods. Laboratory tests and genetic tests.

Results and their discussion. Case presentation.

26-year-old male, sportsman (hockey player), height 192 cm, weight 96 kg presented to Grodno hospital with complaints of blisters and wounds on the skin of his hand. He noticed the appearance of wounds, micro-cuts, and blisters that were only on the skin of his hands. Those wounds were mildly painful and healed very poorly, leaving red scars at the site. Initially, elevated liver enzymes ALT, AST and hepato-splenomegaly were detected. And after accumulation of intermediates of heme-biosynthesis pathway and iron overload were prominent. Rash on his hands consisting of blisters (left thumb), crusted lesion and hyperpigmentations. Increased skin fragility and hypertrichosis is also visible. Laboratory findings showed increased levels of Pentacarboxylporphyrins 174.72 nmol/day (normal <= 10), Hexacarboxyporphyrins 17.33 nmol/day (normal<= 8), Heptacarboxyporphyrins 14.17 nmol/day (normal<= 9). Patient T. denied any consumption of alcohol or tobacco smoking and he never had hepatitis or any liver infection that he knew of. He also denied the role of the sun in PCT symptoms and prognosis. He does not recall any chronic or severe disease in the past (including liver infection). Treatment included multiple courses of phlebotomy with low-dose hydroxychloroquine. After treatment with multiple courses of phlebotomy and low-dose hydroxychloroquine patients' condition improved but not significantly. PCT can be type 1 and type 2. Type 1 of PCT is an acquired disorder and it is triggered by iron overload or viral infection. Type 2 is autosomal dominant [1]. Diagnosis of PCT depends on the history of the patient, clinically and characteristics of the appearance of wounds and blisters on the skin of the back of the hand but main diagnosis still depends on laboratory evaluation and increased level of porphyrin intermediates is helpful for definitive diagnosis [2].

Conclusions. Porphyria cutanea tarda, though rare in young males and often linked to identifiable hepatic risk factors, should remain in the differential diagnosis of photosensitive blistering disorders across age groups.

Primary healthcare clinicians and Dermatologists and should maintain vigilance for PCT in younger patients with compatible cutaneous phenotypes, pursue a comprehensive diagnostic workup.

LITERATURE

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NEUROBLASTOMA GENETIC AND MOLECULAR ASPECTS

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Relevance. Neural crest cells are found in areas like the adrenal glands, chest, and abdomen, which are common sites for neuroblastoma. The maturation, movement, and development of these cells are controlled by various genes, with MYCN playing a major role. Problems with these genes, like overexpression, can lead to tumors. From a genetic perspective, neuroblastoma is linked to the overexpression of the MYCN gene, mutations in genes like ALK and PHOX2B, and changes in the ATRX gene and TERT gene rearrangements, which are more common in high-risk cases [1]. In 85% of cases, mutations in the ALK gene are found at positions F1174, F1245, and R1275. These mutations occur in both types of neuroblastoma – those that happen by chance and those that are inherited. Studies suggest that sometimes these ALK mutations are connected with changes in the MYCN gene [2].

Molecular mechanisms

The MYCN gene helps neuroblastoma cells get energy through a process called aerobic glycolysis. This gene also controls many processes that are important for normal cell development and the formation of cancer. MYCN increases the use of glucose and glutamine, which helps the tumor grow in cancer like neuroblastoma. The RAS-MAPK pathway is active in the primary form of neuroblastoma. This pathway is linked to a worse outlook and poorer results. Mutations in genes such as ALK are connected with the activation of this pathway [2].

Target. To understand the genetic and molecular changes that cause neuroblastoma. This includes looking at variations in the MYCN gene, its overexpression, and changes in other genes like ALK (anaplastic lymphoma kinase), PHOX2b, and others.

Research methods. The information was gathered from existing research and studies.