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Alkaptonuria and Ochronosis

Alkaptonuria is a rare inherited genetic disorder of phenylalanine and tyrosine metabolism. This is an autosomal recessive condition that is due to a defect in the enzyme homogentisate 1, 2-dioxygenase, which participates in the degradation of tyrosine. As a result, homogentisic acid and its oxide accumulate in the blood and are excreted in urine in large amounts. The polymer of homogentisic acid called alkapton impregnates bradotrophic tissues. The first signs of deposition of the ochronotic pigment can be detected accidentally during professional examination of the anterior segment of the eye. The ochronotic pigmentation of the ocular structures is present in approximately 70% of patients. In addition to the sclera, lumps of the ochronotic pigment can be found in the conjunctiva and cornea. Since similar pigmentation of the cornea is not present in other medical conditions, this finding is regarded as pathognomic for alkaptonuricochronosis. The diagnosis of alkaptonuria is based on the characteristic findings in urine. Alkaptonuria patients do not seek medical help due to difficulties with vision—they are without subjective complications. In parallel with the ocular manifestations, ochronotic changes can be found in the hearing organ. Color changes of the auricle are visible in the 10th to 15th year of life. Also typical for alkaptonuricochronosis are changes in the skin, mainly brownish or bluish pigmentation of the skin under the arm, in the face, neck and hands, and rarely on the nails. Given their visibility they may be relevant for the early diagnosis alkaptonuricochronosis. Ochronotic pigment is deposited also on the internal organs. In the field of cardiovascular organs it is the myocardium and blood vessels. From a clinical point of view, the most serious process takes place in the joints and is called ochronoticarthropathy. Therapy of alkaptonuria with Nitisinone is underway.

Biography

Jozef Rovenský completed his MD in 1966 from Medical Faculty, Masaryk's University, Brno, Czech Republic. In 1970, he has undertaken specialization in Internal Medicine, in 1973 in Rheumatology and in 1983 in Clinical Immunology, all from Post-graduate Med. School, Bratislava, Slovak Republic. He completed DSc in 1993 from School of Medicine (Internal Medicine), Comenius University, Bratislava, Slovak Republic. In 1991, he worked as Associate Professor and in 1995 as Professor of Internal Medicine at School of Medicine (Internal Medicine), Masaryk's University Brno, Czech Republic. In 1990, he served as a Director for Research Institute of Rheumatic Diseases, Piešťany. He has over 200 papers *in extenso*. According to the Citation Index these papers were cited more than 2500 times with a H-index of 28.

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