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Titolo	Alkaptonuria and Ochronosis // edited by Jozef Rovenský, Tibor Urbánek, Boldišová Oga, James A. Gallagher
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Descrizione fisica	1 online resource (152 p.)
Disciplina	610
Soggetti	Rheumatology Respiratory organs—Diseases Pathology Otolaryngology Dermatology Molecular biology Pneumology/Respiratory System Otorhinolaryngology Molecular Medicine
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Introduction -- Alkaptonuria and Ochronosis -- History -- Metabolism of Aromatic Amino Acids -- Tyrosine Metabolism Disorders -- Detection of Homogentisic Acid in Plasma and Urine -- Alkaptonuria Genetics -- Experimental Alkaptonuria in Animals -- Experimental Models of Alkaptonuria and Ochronosis -- Ochronotic Arthropathy -- Clinical Manifestation of Ochronotic Arthropathy in the Spine -- Analysis of X-Ray Symptomatology of Ochronotic Arthropathy in the Spine -- Clinical Manifestation of Ochronotic Arthropathy in the Peripheral Joints -- Clinical and Ultrasound Evaluation of Enthesopathy in Alkaptonuria in the Romani Community -- Analysis of X-Ray Symptomatology of Ochronotic Arthropathy in the Area of the Peripheral Joints -- Manifestations of Alkaptonuric Ochronosis in the Eye -- Symptoms of Alkaptonuria and Ochronosis in the Ear -- Manifestations of Alkaptonuria and Ochronosis in the Visceral Organs

-- Manifestations of Alkaptonuria and Ochronosis in the Respiratory Tract -- Pathological Anatomy and Histology of Ochronosis -- Cellular and Molecular Pathogenesis of Ochronosis -- Picture of Synovial Effusion in Ochronosis -- Coincidence of Alkaptonuric Ochronosis with Other Diseases -- Differential Diagnosis of the Disease -- Sine Syndromes in Alkaptonuria -- History of Nitisinone (NTBC) -- Case Reports -- References.

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## Sommario/riassunto

This book comprehensively describes alkaptonuria and ochronosis. Beginning with the history, genetics, pathophysiology and diagnostics of the disease, the authors subsequently present a detailed characterization of its clinical manifestation in the spine, peripheral joints, eyes, ears, visceral organs and respiratory tract, its pathological anatomy and histology, as well as differential diagnosis. This is complemented by the latest data on therapy and experimental models of alkaptonuria, and supported by several case reports. Numerous pictures and radiological images document the clinical symptoms, giving the reader a solid understanding of the disease. On the basis of the editor's and authors' own extensive observations, the book offers an analysis of protein metabolism and aromatic amino acids in the context of alkaptonuria. Written by international experts in the field, the book offers a valuable reference guide for healthcare professionals working in rheumatology, dermatology, pulmonology, otolaryngology and histopathology.

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