Ataxia telangiectasia review pdf

Ataxia telangiectasia, also known as ataxia-telangiectasia (A-T), is a rare genetic disorder characterized by neurological and ophthalmological symptoms. The clinical features of A-T are diverse and include cerebellar ataxia, oculocutaneous telangiectasias, immunodeficiency, and frequent infections. The disorder is caused by mutations in the ATM gene, which encodes a serine-threonine kinase involved in the DNA damage response. This review highlights the molecular mechanisms underlying A-T, including the role of ATM in cellular responses to DNA damage and the impact of ATM deficiency on the development of cancer. It also discusses the clinical manifestations, diagnosis, and management of A-T, emphasizing the importance of early detection and intervention. Illustrations are not included in this text summary. The information is based on the latest research and clinical practices in the field of genetics and oncology, providing a comprehensive overview of A-T for healthcare professionals and researchers.