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Abstract: Xeroderma pigmentosum (XP), trichothiodystrophy (TTD) and Cockayne syndrome (CS) are rare genetic diseases characterized by a large range of clinical symptoms. However, they are all associated with defects in nucleotide excision repair (NER), the system responsible for removing bulky DNA lesions such as those generated by UV light: cyclobutane pyrimidine dimers (CPDs) and pyrimidine-pyrimidone photoproducts (6-4 PPs). Over the past years, detailed structural and biochemical information on NER-associated proteins has emerged. In the first part of the article we briefly present the main steps of the NER pathway with an emphasis on the precise role of certain proteins. Further, we focus on clinical manifestations of the disorders and describe the diagnostic procedures. Then we consider how current therapy and advanced technology could improve patients' quality of life. Although to date the discussed diseases remain incurable, effective sun protection, a well thought out diet, and holistic medical care provide longer life and better health. This review summarizes the current state of knowledge regarding the epidemiology of NER-associated diseases, their genetic background, clinical features, and treatment options.

Keywords: Xeroderma pigmentosum, trichothiodystrophy, Cockayne syndrome, nucleotide excision repair, photosensitivity, neurological abnormalities