

S009 Single-strand break repair and neurodegenerative disease
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Recent work has highlighted a connection between defects in chromosomal single-strand break repair (SSBR) and hereditary neurodegenerative disease. Spinocerebellar ataxia with axonal neuropathy-1 (SCAN1) results from a defect in TDP1, a polypeptide that associates with the SSBR machinery and processes a variety of modified or damaged 3'-termini including abortive intermediates of topoisomerase-1 activity. Ataxia oculomotor apraxia-1 (AOA1) results from mutations in Aprataxin, a second component of the SSBR (and DSBR) machinery with a role in processing damaged DNA termini. Recent advances in our understanding of the role of these and other proteins in DNA strand break repair and in the relationship of this process to human neurological disease will be presented.