Leukodystrophies with late disease onset: an update.
Köhler W.

Source
Fachkrankenhaus Hubertusburg, Klinik für Neurologie und Neurologische Intensivmedizin, Wermsdorf, Germany.
wolfgang.koehler@kh-hubertusburg.de

Abstract
PURPOSE OF REVIEW:
Knowledge of the metabolic and genetic basis of known and previously unknown leukodystrophies is constantly increasing, opening new treatment options such as enzyme replacement or cell-based therapies. This brief review highlights some recent work, particularly emphasizing results from studies in adulthood leukodystrophies.

RECENT FINDINGS:
Evidence from recent studies suggests increasing importance of metabolic dysfunctions, for example, in peroxisomal lipid metabolism or energy homeostasis, influencing axonal integrity and oligodendrocyte function and leading to white matter demyelination. In addition, diagnostic and therapeutic progress in metachromatic leukodystrophy, X-linked adrenoleukodystrophy, Krabbe diseases and other rare leukodystrophies with late onset are summarized.

SUMMARY:
Better understanding of leukodystrophies in neurological routine practice is of crucial importance for differentiating between other white matter diseases such as toxic, inflammatory or vascular leukoencephalopathies. Many leukodystrophies are particularly important to recognize because specific treatments already exist or are currently under investigation. The article also provides an overview of currently known leukodystrophies in adulthood.