081 ADULT LEUKODYSTROPHY: MULTI-DISCIPLINARY STRUCTURED APPROACH

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Background Leukodystrophy refers to a rare group of conditions that are inherited and involve progressive destruction of previously acquired myelin. Diagnostic difficulty arises because individual disease prevalence is less than 1 in 20000, the presentation in adults (>16 years) may be different compared to the larger classical paediatric experience, and a number of acquired white matter conditions (eg multiple sclerosis and vascular disease) may have to be eliminated. In order to make the process more efficient a multi-disciplinary group was established using a structured approach. Our early experience is presented together with an illustrative case.

Methods The group has met since 2012. The central features of the diagnostic pathway were: family history, ethnicity, specific clinical features and MRI analysis.

Results 23 adult cases were reviewed, and new diagnoses made in 12 cases to date. The diagnoses included CADASIL, hereditary diffuse leukoencephalopathy with neuroaxonal spheroids (HDLS) (CSF1R mutation), hereditary sensory neuropathy with dementia and hearing loss (DNMT1 mutation), X-linked adrenoleukodystrophy/adrenomyeloneuropathy (ABCD1 mutation).

Summary A structured diagnostic approach is helpful. Most new diagnoses came from genetic testing, which looks to be more efficient than conventional investigations. A full review is given in Ahmed *et al*, JNNP, 2014. Referrals are welcome.