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# Poster P18

## A neuropathy-mitochondrial multidisciplinary team meeting: enhancing diagnosis

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Introduction

Next-generation sequencing has highlighted the clinical and genetic overlap of genetic neurological disorders. However, geographic and organisational distance between departments can limit interdisciplinary working. We hypothesised that a multidisciplinary team meeting (MDT) bridging neuropathy and mitochondrial teams could facilitate enhanced diagnostics and management of our patients.

Methods

Patients were recruited at a specialist inherited neuropathy centre or the Highly Specialised Services for rare mitochondrial disorders. Genetically undiagnosed families were discussed over three, one-hour meetings.

Results

Eighteen families were discussed and action plans generated based on a mitochondrial variant (4/18) or phenotype (14/18). These included reanalysis of existing genomic data, further clinical phenotyping, fadvanced analysis of existing histological samples e.g. muscle biopsies, and additional genetic testing. Nine families (50%) have a confirmed genetic diagnosis, accounting for at least part of their neurological syndrome, and a further two have a promising genetic candidate. These include POLG-related neuropathy, intellectual disability and retinitis pigmentosa syndrome due to recessive SCAPER variants, NDUFAF2-related neuropathy, ataxia and optic atrophy syndrome and MT-ATP6 myeloneuropathy. Diagnoses were made with testing available via the clinical service in 44% (4/9) of solved cases, as directed by clinical expertise.

Conclusions

An MDT combining the approaches of two specialist services has resulted in a 50% diagnostic rate for previously unsolved patients. This highlights the benefit of interdisciplinary working where complex phenotypes bridge classical sub-specialities.