| 030 | THE CLINICAL AND GENETIC SPECTRUM OF HEMIPLEGIC MIGRAINE |

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Objectives Hemiplegic migraine (HM) is a rare form of migraine with aura with a prevalence of 0.01–0.002%. Familial and sporadic forms are described and associated with ataxia and seizures. Causative genes include; CACNA1A, ATP1A2, SCNA1 for FHM types 1, 2 and 3 respectively. Recently PRRT2 mutations have been reported. However, significant numbers of patients remain genetically-undefined. The objectives are (i) Phenotype characterisation and (ii)Molecular investigations.

Methods 50 probands were screened on a targeted gene panel designed for the Illumina TruSeq Custom Amplicon platform. Amplicons defined coding regions of known paroxysmal genes; CACNA1A, ATP1A2, SCN1A, KCNK18, KCNA1, ATP1A3, SLC2A1, SLC1A3, PRRT2, MR1, CACNB4. Variants were validated with Sanger sequencing.

Results All patients reported classical symptoms; hemiplegic and/or hemisensory aura (100%), focal headache (100%), and complex aura (53%). Other features included epilepsy (13%), ataxia (10%), migraine with or without typical aura independent of HM (33%) and dyskinesia (2%). Variants were identified in: CACNA1A (12%), SCN1A (6%) and ATP1A2 (4%), PRRT2 (2%).

Conclusions CACNA1A is a major disease gene but HM shows significant clinical and, genetic heterogeneity. The paroxysmal disorders gene panel will be an important diagnostic tool in investigating HM, related disorders and the role of ion channels. Whole-exome sequencing is underway for unresolved cases.