

**P034** Identifying novel diabetes disease genes in patients with neonatal diabetes by homozygosity mapping

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The advent of SNP chip technology has made genome wide scans accessible and affordable for disease linkage studies. The standard methodology for homozygosity mapping uses the SNP data to identify genetic regions shared by multiple affected members of a family and has enabled the identification of a wide range of autosomal recessively inherited disease loci.

Our aim is to use homozygosity mapping to identify novel diabetes genes in small families and non-related individuals, from consanguineous or isolated populations. Investigations into the characteristics of mutation containing homozygous regions have been undertaken by using a training set of 15 individual probands with known recessively inherited mutations across 4 genes (*GCK*, *EIF2AK3*, *ABCC8* and *KCNJ11*). The proof of principle has been achieved with the genetic aetiology for 10 small families and individuals having been identified by sequencing of known diabetes genes within homozygous regions indicated by the studies. Our collection of probands with an unknown aetiology includes 7 small families and 32 individuals with diabetes diagnosed before 6 months of age. Our studies demonstrate the usefulness of this method to identify novel diabetes genes within our large group of patients.