

**P032** Mutations in the *proinsulin (INS)* gene are a common cause of neonatal diabetes

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Heterozygous mutations in the *proinsulin (INS)* gene represent a novel cause of neonatal diabetes. We aim to determine the prevalence of *INS* mutations in 286 patients diagnosed with diabetes before two years of age. Mutations in the *KCNJ11* gene had been excluded in all cases and a further 121 had also been tested for *ABCC8*.

*INS* mutations were identified in 31/142 probands diagnosed under 6 months of age, 2/86 diagnosed between 7-12 months, and none in those diagnosed between 1 and 2 years. All *INS* mutation carriers were insulin treated and the median age at diagnosis was 9 weeks (0-35). In keeping with a lack of insulin *in utero*, birth weight was reduced (median 10<sup>th</sup> centile, (<1-86)). Three probands inherited the mutation from an affected parent, two of which were diagnosed at 7 and 22 years.

In conclusion mutations in the *INS* gene are a common cause of neonatal diabetes, with the vast majority presenting in the first 6 months of life, although they can rarely result in diabetes in older children or adults.