ESM Results – Accuracy of Clinical diagnoses

The accuracy of clinical diagnosis, defined as a positive genetic test result in a patient with a clinical suspicion of MD for each of its types, was: 84/258 (33%) for *GCK*-MODY, 4/28 (14%) for *HNF1A/HNF4A*-MODY, 2/4 for *HNF1B*-MODY, 6/15 for neonatal diabetes (five with a heterozygous dominant mutation in *KCNJ11* and one in a previously described case with a mutation in *GCK* [1]), 2/3 for Wolfram syndrome and 1/1 for Alström syndrome. The patient with neonatal diabetes and congenital heart defect had a deletion of a fragment located on the short arm of chromosome 8 detected using FISH (fluorescent in-situ hybridisation) and SNP-array approaches (data not shown). *GCK*-MODY was therefore the most commonly observed form, detected in 83% of children referred to genetic screening, followed by neonatal diabetes (7%), *HNF1A/HNF4A*-MODY (4%), *HNF1B*-MODY (2%) and diabetes in Wolfram and Alström syndromes (2% and 1%, respectively). Genetic tests were withheld in one referred patient after reconsideration of the phenotype.

Reference:

[1] Borowiec M, Mysliwiec M, Fendler W, et al. (2011) Phenotype variability and neonatal diabetes in a large family with heterozygous mutation of the glucokinase gene. Acta Diabetol 48: 203-208