Conclusion In children and adolescents with T1DM DPN is highly prevalent, but in the majority of patients it is subclinical. Sensitivity and negative predictive values of the clinical neurological examination are low. Therefore, routine NCV measurement for the assessment of DPN appears warranted in these patients.

Introduction Molecular and genetic advances have changed the way we look at associations of signs and symptoms or miscellaneous syndromes. A recent report has hypothesized that KCNJ10 mutations, affecting potassium channels present in the brain, ear and the kidneys, are responsible for the constellation of epilepsy, ataxia, sensorineural deafness, and tubulopathy (EAST or SeSAME syndrome). We present six patients belonging to three families with similar findings.

Case Series We describe three Asian siblings, two Caribbean siblings, and one Caucasian child who have epilepsy, ataxia, sensorineural deafness, and tubulopathy. Consanguinity was present only in the Asian family. Seizures were a presenting symptom in four of the cases with onset as early as 3-7 months of age. Development delay and learning difficulties were present in all of the cases. Ataxia was evident from early on. Sensorineural hearing loss was identified at different ages and in some cases was asymptomatic. In some cases, tubulopathy was an incidental finding. Two of the children were being followed up by nephrologists and neurologists before a unifying diagnosis was determined. Five children had previously been extensively investigated with metabolic and mitochondrial investigations, magnetic resonance images, and electroencephalograms all normal. All six children had biochemical evidence of a tubulopathy with hypokalaemia, hypomagnesaemia, and alkalosis. KCNJ10 DNA mutations have been identified in all the children.

Conclusion Recent advances in genetics have enabled us to determine the likely unifying cause for hitherto puzzling signs and symptoms in six children under our care.