



EDUCATION BULLETIN - OCTOBER 2015

Neonatal Diabetes

Neonatal diabetes is where a baby develops diabetes in the first six months of life. At this age, it is quite possible that the cause of diabetes is not type 1 (autoimmune diabetes). There are various monogenic (single-gene mutation) causes of diabetes that particularly present in the first few months of life. **Genetic testing should be undertaken because it may influence management.**

Monogenic diabetes is uncommon, accounting for ~1–4% of paediatric diabetes cases. There are various types, and they appear to occur all over the world – not just in Europe and the Americas but also in Africa and Asia.

The commonest form of monogenic diabetes occurring in the neonatal period is mutations of the KCNJ11 gene - this represents 30-40% of all cases. Infants with this diagnosis actually do much better when treated with sulphonylureas such as glibenclamide rather than insulin. However, the genetic diagnosis must be made before insulin is stopped, and the changeover from insulin to glibenclamide.

There are a number of other types, such as mutations of the INS gene and others. Some of the rarer types are associated with congenital abnormalities. All these other types require insulin.

Genetic testing for any baby diagnosed under six months of age can currently be done free-of-charge by a laboratory in Exeter in the United Kingdom. DNA from the baby and parents needs to be sent to the laboratory in a specific way. To arrange this testing, please see and follow the instructions on the website <http://www.diabetesgenes.org/content/genetic-testing-diabetes-presenting-infancy>

Note that it doesn't matter if the child is now older than six months of age – as long as the diagnosis was made in the first six months, the child should be tested, no matter what their age – the genes do not change.

If you have any young people who were diagnosed with diabetes in the first six months of life, we encourage all centres to make use of the free testing by this highly expert and very helpful English team.

Please contact us if you have any questions. If a transition to glibenclamide is advised for any of your patients, we can connect you with an expert who can help guide you through the process.

For further reading: [ISPAD Clinical Practice Consensus Guidelines 2014 The diagnosis and management of monogenic diabetes in children and adolescents](#)