Dr Elisa De Franco is a molecular geneticist at the University of Exeter. She has been working at understanding the genetics of neonatal diabetes for 10 years. Elisa obtained an undergraduate degree and a master's degree in medical biotechnologies at the University of Turin, Italy, followed by an EU-funded PhD fellowship at the University of Exeter. Elisa's research has also been funded by the Naomi Berrie Foundation for the Study of Diabetes, the Diabetes Research and Wellness Foundation and the European Foundation for the Study of Diabetes in partnership with Novo Nordisk. Elisa was the Society for Endocrinology Early Career Science Prize lectura in 2021. She received the ISPAD Young Investigator Award in 2020, the European Association for the Study of Diabetes Rising Star award in 2018 and in 2019 she has been awarded an RD Lawrence fellowship by Diabetes UK. She has delivered over 15 international invited lectures and published over 50 papers, including first-author publications in the Lancet, Nature Genetics, the American Journal of Human Genetics, the Journal of Clinical Investigation and Diabetes. Elisa is the scientific lead for the neonatal diabetes genetic testing service at the University of Exeter. Her research uses genome sequencing to discover genes essential for beta-cell development and function. She has so far discovered and published 7 novel genetic causes of neonatal and early onset diabetes. Elisa believes that the integration of genetic research and clinical care is fundamental to improve treatment and care for children with rare forms of diabetes and can lead to important insights into the biology of type 1 and type 2 diabetes.

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Title: Using gene discovery in rare disease to gain new insights into pancreas development and function

Abstract: Understanding how pancreatic beta-cells develop during human development is essential to advance current protocols aimed at developing insulin-producing beta-cells in vitro and highlight therapeutic targets for diabetes treatment. Identifying the single-gene mutations which result in individuals developing diabetes in the first 6 months of life (a condition called neonatal diabetes) has the potential to give unique insights into the genes regulating human beta-cells which would never be discovered by studying animal models alone.

By performing genome sequencing analysis of >100 individuals with neonatal diabetes, we have identified mutations in genes which were not previously thought to be important within beta-cells. These include genes essential for preserving beta-cell function (like YIPF5 which encodes a regulator of endoplasmic reticulum to Golgi transport) and genes crucial for human pancreatic development (such as the gene encoding for the negative regulator of transcription and stem cell pluripotency factor, CNOT1).

These results highlight the power of human genetic studies to pinpoint genes which are essential for human beta-cell function and development, improving our knowledge of the biological mechanisms leading to diabetes and highlighting new promising targets to be further investigated to develop better therapies for individuals living with diabetes.