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Maturity Onset Diabetes of the Young (MODY) is the most common form of monogenic diabetes and represents 2-3 % of all diabetes cases in Norway. MODY patients confer sensitivity to sulfonylurea. Thus, it is important to diagnose subjects to install precision medicine. One challenge is to distinguish pathogenic from benign variants. Functional analyses have shown to be a valuable tool to correctly classify variants. In this project, we are using a robust functional pipeline to characterize all variants of unknown clinical significance identified in patients from the Norwegian MODY registry, aiming for an improved diagnosis and treatment of subjects with diabetes. In order to gain more knowledge of signaling pathways in the pancreatic beta cell, we have also investigated how new molecular mechanisms are involved in the regulation of hepatocyte nuclear factor -1 alpha (HNF-1A), which is important for controlling normal pancreatic beta cell function.

The project is taking place at the KG Jebsen Center for Diabetes Research, under the supervision of researcher Ingvild Aukrust (main supervisor), Prof. Pål R. Njølstad (co- supervisor) and Prof. Lise B. Gundersen (co-supervisor).