Polygenic risk for triglyceride levels in the presence of a high impact rare variant

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In our 2022 publication, we reported that having a 22q11.2 deletion and being male increase the risk of having high levels of triglycerides (a condition called hypertriglyceridemia). Chromosome 22q11.2 does not contain any genes that are directly related to hypertriglyceridemia. So, what makes this condition to be more likely in adults with 22q11.2DS?

In this study, our team checked if certain common genetic variations outside of the chromosome 22q11.2 region can predict the risk of having higher levels of triglycerides. In 157 adults with 22q11.2DS and detailed genetic data, we found that these common variants, together with obesity and being male, could predict the risk of high triglyceride levels quite well.

Of course, lipid levels (including triglycerides) can easily be measured in a blood test. In the future, however, children found to have the variants that increase their risk of hypertriglyceridemia may be encouraged to adopt healthy diets and active lifestyles early on in life to lower the risk.