Thema 7 Project Proposal

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# The project

This sequencing project focused on a certain frameshift mutation which changes the transcription rate of a gene. The gene in question is called SP1. The frameshift causes a heterozygous variant going by the name of c. 1995delA. This study has observed that this pathogenic variant may play an important role in causing a multitude of genetically inherited syndromes, or surely heightening the chance of acquiring a syndrome. This set of syndromes falls under a group named BMF, bone marrow failure. Inherited, in the case of this project.