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Degree Project in Bioinformatics

Masters Programme in Molecular Biotechnology Engineering,
Uppsala University School of Engineering

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Author	Jessika Nordin	
Title (English)	Assessment of variant load in an idiopathic autoinflammatory index patient	
Title (Swedish)		
Abstract	<p>An index patient with an idiopathic autoinflammatory disease was sequenced for over ~1900 immunological genes, and their regulatory elements, in a Targeted Sequence Capture Library. This data was used for creating a bioinformatics pipeline for all projects that use the same library. The pipeline was built from the GATK best practices framework and goes from raw sequence data to a list with ranked variants.</p> <p>To receive a list of interesting variants, the index patient was compared to his immediate family and a cohort of Swedish controls. This was done since it is probable that the disease causing variants in the index patient is private to him (the family do not have the variant). The controls were used to be sure that the variants are not common in the Swedish population.</p>	
Keywords	Autoinflammatory disease, bioinformatics pipeline, GATK, index patient, variation load	
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