

Diagnostic odyssey, a deep dive into patient symptomology and disease state pathophysiology is often performed in children with undiagnosed diseases of suspected genetic origin, who face uncertainty in daily life. The goal of Diagnostic odyssey is to uncover possible diagnoses in patient (Proband) cases where top medical specialists do not identify a diagnosis. The ultimate goal is not to provide appropriate medical care, but rather to provide the healthcare team with a rational approach to otherwise hidden, genetic mutation-based pathophysiology, to augment their treatment strategy options.

In the case of a young patient with a recurrent undiagnosed respiratory illness, iVariant Guide, an application produced by Advaito Bioinformatics, was utilized to map out the exome of the patient and the patient's parents. Utilizing filtering techniques within the iVariant Guide system, data was gathered regarding genetic mutations present in immune function-related pathways. Analysis was conducted on the list of genetic variants provided in iVariant Guide, and a list of pathways of interest was developed based upon those findings.

The most notable findings show multiple potential pathologies for the Proband's respiratory illness. These pathways include genetic mutations present in the Proband which, based upon current knowledge, would increase possibly pathological immune response and lead to similar symptomology. Our findings are still ongoing, but support the promise of whole exome sequencing as a viable method for diagnostic odyssey resolution.