LABORATORY REPORT

Patient Information	Laboratory
Patient no.: IKHS200000256 **Patient Name**: Sanad A. Dmour **Date of Birth**:06/30/2014 **Gender**: Male **Ethnicity**: Mediterranean	**Physician Name**: Dr. Ali Alhawamdeh **Specimen**: DNA **Received Date**: **Prepared by**: **Report Date**: 04/05/2020

TEST REQUESTED: Clinical Whole Exome Sequencing

CLINICAL INFORMATION*

A 5 years and 8-month old boy born to non-consanguineous parents via IVF manifested with global developmental delay since age of 2 years old, delayed speech and communication and lack of social activity. Basic metabolic screening was negative for inborn errors of metabolism and no proximal discharges or epileptiform activity. Family history: This individual's twin brother with similar manifestations and hyperactive .

*: Clinical information indicated above follows HPO nomenclature.

TEST RESULT

POSITIVE RESULT

A 'VARIANT OF UNKNOWN SIGNIFICANCE(VUS)' WAS IDENTIFIED.

INTERPRETATION

By whole-exome sequencing, a heterozygous variant, c.1847A>T (p.Lys616Met) in the *ATAD3A* gene was identified. A clear genetic explanation for the disease in this individual could not be found as only a VUS was identified.

RECOMMENDATIONS

1. Genetic counseling is recommended to discuss the implications of this test report and test results should be interpreted in the context of the patient's medical evaluation, family history, and racial/ethnic background.
2. Reinterpretation of exome sequencing data is recommended on an annual basis and may be requested by the referring clinician and one should be cautious about that variant classification and/or interpretation may change over time if more information becomes available and identification of new variants associated with disease phenotype during the re-assessment.
3. Further studies (clinical/functional) may help in ascertaining the role (if any) of these variants in the disease predisposition.