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Test ID - 67632

Final Report

Summary of Patient Information

Name:	Date of Birth:	Gender:	Viafet ID:
Mahmoud Abdullah Alhunaiti - HC07100204	23 Apr 2019	Male	BEY020728

Referring Clinic Information

Center:	Clinician:
Hamad Medical Center	Dr. Moza Al-Bader

Test Requested

Pre-PGT Workup

Summary of Result

Patient	Zygosity	Variant	Gene	Condition	Mode of Inheritance
Mahmoud Abdullah Alhunaiti - HC07100204	Hemizygous	NM_000500.7: c.1A>G; p.Met1?	CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	Autosomal Recessive

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Patient Information

Name:	Date of Birth:	Gender:	Viafet ID:
Mahmoud Abdullah Alhunaiti - HC07100204	23 Apr 2019	Male	BEY020728

Referring Clinic Information

Center:	Clinician:	External ID:
Hamad Medical Center	Dr. Moza Al-Bader	HC07100204-New

Sample Information

Specimen:	Collection Date:	Receipt Date:	Report Date:
Peripheral Blood in EDTA Tube	May 18, 2025	May 26, 2025	July 01, 2025

Test Requested Pre-PGT Workup

Clinical Information: Sample submitted as part of the PGT-M Setup in order to confirm the external laboratory result and perform internal quality control.

Summary of Result:

Hemizygous for NM_000500.7: c.1A>G; p.Met1?

Result and Interpretation: The proband is hemizygous for NM_000500.7: c.1A<G; p.Met1? likely pathogenic variant in the CYP21A2 gene established in association with autosomal recessive Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency. See Test Order ID 67630 and 67631 for testing performed concurrently.

Methods:

DNA was extracted using the Qiagen DNA Extraction Mini Kit. To evaluate the presence of NM_000500.7: c.1A<G; p.Met1? in the CYP21A2 gene, the targeted sequence and at least 150bp of flanking regions were amplified using PCR. After cleaning the PCR products, cycle sequencing was carried out using the ABI Big Dye Terminator v.3.0 kit. Products were resolved by electrophoresis on an ABI 3500 capillary sequencer. The patient's sequences were aligned and compared with the reference sequences (GRCh37). Nomenclature used in this report follows the guidelines as outlined by the Human Genome Variation Society (HGVS) v15.11. Test reports contain no information about other portions of the gene. This test was developed and its performance characteristics determined by Viafet Genomics Laboratory. The US Food and Drug Administration (FDA) has determined that clearance or approval of its method is not necessary and thus neither have been obtained. This test was developed for clinical purposes. The above result and interpretation assumes that samples received by the laboratory were correctly labeled and that family relationships and clinical diagnoses are as stated. Any remaining DNA is retained indefinitely as per local regulations. DNA may be used anonymously in appropriate quality control of this variant. In order to avoid error and/or misinterpretation, it is inadvisable to transcript any portion of this report.

Recommendation: Genetic counseling is recommended to explain test results to the patients and to discuss reproductive or medical options.

Report electronically signed by:

Dr. Ali Hellani, PhD, MHGSA
Laboratory Director