LABORATORY REPORT

Redcliffe labs

Patient Name	**: Mrs Dummy**		
DOB/Age/Gender	: 18 Y/Female	Sample Collected	: XXX
Patient ID / UHID	: XXX	Report Date	: XXX
Referred By	: Self	Barcode No	: XXX
Sample Type	: Whole blood EDTA	Report Status	: Final Report

Freidreichs Ataxia Mutation Analysis (FRDA)

TEST NAME	Friedreich Ataxia Molecular Diagnosis
SPECIMEN INFORMATION	Peripheral Blood collected on 13/12/2023

CINICAL HISTORY:-
C/o primary amenorrhea and reduced breast development.
MRI ABDO PLUS Pelvis - ABSENCE of UTERUS and OVARIES
PROVISIONAL Diagnosis- ?Hypergonadotropic Hypogonadism(?Mullerian AGENESIS AND Ovarian dysgenesis) with Gait and limb ataxia (sensory plus mild cerebellar) with Hammer toes with Pes cavus with AREFLEXIA and extensor planters.
Differentials- ? Gordon HOLMES syndrome, ? Boucher Neuhauser

METHODOLOGY:-
TP-PCR/ Fragment Analysis
This assay detects GAA expansion in the first intron of Frataxin gene.
1• PCR Amplification flanking GAA Repeats/ Fragment Analysis - This assay determines the accurate size of the triplet repeat up to approximately 60 - 70 repeats
2• Triplet primed PCR (TP-PCR) – This assay indicates the absence or presence of large expansion. It identifies large GAA repeats that may have been missed using current PCR methods.

Combination of GAA PCR and Triplet primed PCR helps in distinguishing cases of FRDA versus carriers.

TEST RESULTS

GAA Repeats	**TP PCR**	**Interpretation**
8 on allele 1 and 9 on allele 2	**Normal alleles**	**Diagnosis of FRDA excluded**

INTERPRETATION

Normal alleles	5-33 GAA repeats
Premutated alleles	34-65 GAA repeats
Mutated alleles	66-1700 GAA repeats

CLINICAL BACKGROUND
1• Friedreich ataxia (also called FA or FRDA), an autosomal recessive neurodegenerative disease, is associated with an unstable expansion of a GAA trinucleotide repeat in the first intron of the frataxin gene on chromosome 9q13.
2• Friedreich ataxia is a slowly progressive ataxia with typical onset between ages 10-25 years. It is characterized by depressed tendon reflexes, speech disturbance (dysarthria), muscle weakness, visual disturbance due to optic nerve atrophy and spasticity. Cardiomyopathy is the predominant cause of death.
3• Although rare, Friedreich's ataxia is the most common form of hereditary ataxia, affecting about 1 in every 50,000 people in the United States. Both male and female children can inherit the disorder.
4• FA is caused by a GAA expansion in the first intron of Frataxin gene. Normal GAA repeat length is 5-33 GAA repeats. 34-65 uninterrupted GAA repeats are mutable normal alleles. They do not cause disease, but may expand during transmission to the next generation and become disease-causing alleles. Disease-causing alleles have 66-1700 repeats.
5• 96% of affected individuals are homozygous for a triplet repeat expansion. About 4% of affected individuals will have a GAA expansion on one allele and a point mutation on the other allele.

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6• Appropriate genetic counseling for affected families requires accurate genetic diagnosis of Friedreich's ataxia that helps in differentiating it from other ataxias. Once the diagnosis of FRDA is confirmed, extended family screening followed by genetic counseling can be provided.

Indications for Testing:
1• Confirmation of Clinical Diagnosis of FRDA
2• Test is useful in the differential diagnosis for autosomal recessive cerebellar ataxia.
3• Carrier Risk Assessment- Positive Family History

Limitation of the Assay:
Presence of PCR inhibitors in the sample may prevent DNA amplification. Paradoxical results may arise due selection of inappropriate specimens and contamination during specimen collection.

References:
1• Potdar et al., 2013, Annual Review & Research in Biology, 3(4): 659-677
2• Ryan, 2000, EMQN guidelines
3• Muthuswamy et al., 2013, Hippokratia; 17(1): 38–41

*** End Of Report ***

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