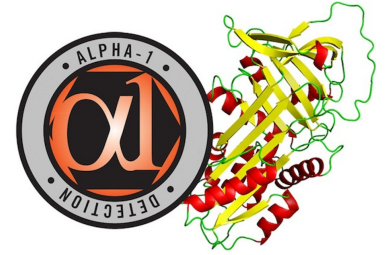




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PLEASE READ. IMPORTANT INFORMATION REGARDING THE CONFIRMATORY TESTING.

Dear Healthcare Provider,

We would like to thank you for screening your patients through GeneAidyx. Our priority is to provide you and your patients with the most accurate genetic diagnosis for alpha-1-antitrypsin deficiency possible. The impact of the diagnosis of alpha-1-antitrypsin deficiency to patients and their families is life changing. Because this diagnosis is so important we adhere to the American College of Medical Genetics (ACMG) guidelines for testing.

The ACMG guidelines emphasize 'the importance of retesting a positive specimen... this practice not only guards against communication errors, historical changes in the numbering of exons, and inconsistency in the numbering of nucleotides and codons, but also allows the laboratory to absolutely rule out dropout of an abnormal allele which can occur through family-specific polymorphisms in primer sites. This approach provides a level of customized QC not otherwise available. Although allele dropout is rare, the ease of doing this simple QC step makes it compelling.' (URD 9.2.4.2)

The ACMG defines a confirmation as 'A repeat analysis on a sample from an individual....this may be done: 1) using a new independent sample; 2) using another aliquot of DNA prepared from the original submitted sample; or 3) using another method of analysis from the one used originally to identify the sequence variation.' (URD 2.4)

Confirmation testing is a service we encourage and provide free of charge (including return shipping). By submitting a whole blood sample on your patients who have tested positive for a deficient allele, we are able to confirm the original genotype, perform a plasma AAT level and perform high quality reflex testing by determining the PI type by Isoelectric Focusing.

We encourage you to offer this additional confirmation testing to your patients with abnormal allele(s). If phlebotomy is not a service you provide, you can refer your patients, with our custom confirmation kit, to a local lab for a blood draw.

If you would like to discontinue receiving our confirmation kits automatically, please have your office call us and we will place a note in our database to only send confirmation kits when you specifically request them.

If you have any questions or concerns, please feel free to contact us by phone at 866-522-0692 or email us at alpha1lab@geneaidyx.com.

Sincerely,

Mark L. Brantly, MD
Medical Director
GeneAidyx, Alpha-1 Antitrypsin Genetics Laboratory

ACMG reference URL:
https://www.acmg.net/Pages/ACMG_Activities/stds-2002/URD.htm