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UK NEQAS for Alpha 1 Antitrypsin & Phenotype Identification

Distribution : 182	May 2018	Participant : 91339
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43 out of 52 participants returned data for this distribution. 83% response rate.

Nil Responses

INFO2, 10711, 91347, 94474, NOKLUS, YVSOLAB, ORG_G, ORG, INFO

182-1 is a sample from a 40 year old female with chronic obstructive pulmonary disease (COPD).

182-2 is a sample from a 20 year old male with asthma.

Sample	Designated Response	Your Response	Score	OMIS
182-1	M, MM	MM	0	0
182-2	M, MM	MM	0	2
			Total MIS	2

The current window of analysis comprises the previous four distributions

Comments

Sample 182-1:

36 laboratories reported either PI M or PI MM for sample 182-1. Single responses were also submitted of phenotypes; PI MS, PI MZ and PI M Degraded. 4 laboratories reported an AAT quantitative level but did not report a phenotype and 3 laboratories only reported a phenotype. Laboratories that have reported results that are out of consensus should recheck their gels.


Sample 182-2:

33 laboratories reported sample 182-2 as PI M or PI MM. Four laboratories reported PI M Null and 1 laboratory reported PI MZ. Five laboratories reported an AAT quantitative level but did not report a phenotype. 3 laboratories only reported a phenotype. Laboratories that have reported results that are out of consensus should recheck their gels.

Sample 182-2 had a low antitrypsin quantitation that could be due to the presence of a null allele or alternatively by antitrypsin loss, such as occurs in a protein losing enteropathy. The presence of a homozygote (e.g. MM) can not be differentiated from a null heterozygote (e.g. M null) unless family studies have taken place. 15 laboratories provided additional comments to indicate that the antitrypsin quantitation is lower than expected for a homozygote and that either protein loss or the presence of a null allele cannot be excluded.

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