

Email: ukneqas@immqas.org.uk

Web Address: http://www.immqas.org.uk

## UK NEQAS for Alpha 1 Antitrypsin & Phenotype Identification

Distribution : 162

May 2016

Participant : 91339

44 out of 52 participants returned data for this distribution. 85% response rate.

### NII Responses

INFO2, 10162, 10711, 93470, 94405, ORG\_G, ORG, INFO

**162-1** is a sample from a 20 year old male with asthma.

**162-2** is a sample from a 49 year old female with a family history of liver disease.

Sample	Designated Response	Your Response	Score	OMIS
162-1	M, MM	MM	0	0
162-2	M, M1M2, MM	MM	0	0
			<b>Total MIS</b>	0

The current window of analysis comprises the previous four distributions

### Comments

Sample 162-1:

33 laboratories reported sample 162-1 as PI M or PI MM. Three laboratories reported PI M Null and 4 reported PI M with some degradation. Four laboratories reported an AAT quantitative level but did not report a phenotype.

Sample 162-1 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.

Sample 162-2:

32 laboratories reported sample 162-2 as PI M or PI MM and 4 reported PI M1M2. Three laboratories stated that there was PI M with some sample degradation and one laboratory reported PI degraded. Four laboratories reported an AAT quantitative level but did not report a phenotype.

This patient had previously been phenotyped as PI M1M2 and the electrophoretic pattern would be expected to show splitting of the M4 and M6 bands but this phenotype was only reported by four laboratories.

The Total Misclassification scoring (**MIS**) system for the **qualitative** element of the scheme gives an indication of the number of instances where a laboratory's response is at variance with that defined for each specimen in a window of **four** distributions.

A panel of reference laboratories (10070, 10656, 10776, 14477) were in agreement in providing the target responses.

Laboratories have been given misclassification score (MIS) for each sample where they are at variance with the target response as determined by the Reference Panel. The overall misclassification score (OMIS) now includes a running window of 8 samples over 12 months.

The categories of performance for Phenotype Identification are:

	<b>Total MIS</b>
● <b>Good</b>	Zero
○ <b>Adequate</b>	1-3
● <b>Poor</b>	> 3

For further performance criteria information please see our website at <http://www.immgas.org.uk>.

If laboratories require further assistance please contact the centre.