ERNDIM Qualitative Blood Spot Acylcarnitine Scheme

Annual Report London 2011

Two circulations (17 & 18) were sent out to the 53 laboratories assigned to the London centre of the ERNDIM dried blood spot acylcarnitine scheme. The first was sent out in June, with a return date of 15th July 2011 and the second in May 2012 with a return date of 8th July 2012. The extreme delay to the second circulation, for which the scheme organisers apologise, was due to problems of sample supply. This qualitative scheme depends on the goodwill of patients donating "spare" blood for QA purposes to allow the distribution of real clinical blood samples. This may also mean that blood spots distributed may be of sub-optimal size & quality.

Returns for circulation 17 were received from 47 (89%); all of these arrived by the initial due date. For circulation 18 valid returns were received from 45 (85%); 5 of these arrived after the initial due date of 8/6/2012 but results were included as there had been delays in sample receipt .

There were 5 laboratories who failed to make a return on either circulation. Three of these did not submit results in 2009 or 2010. Two laboratories reported on Circulation 17 only, and one on circulation 18 only.

Participants were asked to respond via email using a supplied report template, and to send a scan and/or table of quantitative results if possible. All laboratories responded by email. All laboratories provided a suggested/differential diagnosis. Most suggested some form of appropriate follow-up to confirm a putative diagnosis. A summary of the samples sent and the number of respondents suggesting the definitive diagnosis as part of the differential is given in the table below.

Sample	Enzyme/transporter defect	Diagnostic Acylcarnitine	Respondents
17a	Normal acylcarnitine profile		44/47
17b	Methylmalonic acidaemia (MMA, MIM251000)	C3	45/47
17c	Medium chain acyl CoA dehydrogenase deficiency (MCADD, MIM 201450)	C8, C6, C10:1	47/47
18a	Multiple acyl CoA dehydrogenase deficiency (MADD, MIM 231680)	C8, C10	23/45
18b	Normal		40/44
18c	Treated Carnitine uptake disorder (CUD/CTD MIM 212140)	borderline low C0	29/44

Sample 18a proved particularly difficult to interpret. This was from an adult patient, currently well, on vitamin supplements, with well controlled multiple acyl CoA

dehydrogenase deficiency (MADD, MIM 231680). The C8 & C10 were clearly abnormal but the other acylcarnitine species considered diagnostic for MADD were not significantly raised. There was a clear division of respondents into those who suggested a diagnosis of MCADD based on the raised C8 acylcarnitine and those who considered MCADD unlikely due to the raised C10 and proportionately lower C10:1. Most laboratories suggested appropriate follow-up tests which would have informed definitive diagnosis.

Sample 18c gave an essentially normal profile. Interpretation relied heavily on the clinical details given. Laboratories who did not suggest CUD have not been marked down.

Once again, we are extremely grateful to the centres that have provided informative material for circulation. If any participants can provide samples in the future it would enormously facilitate this scheme, providing, as it does, genuine clinically derived samples for assay and interpretation. 3-4ml of lithium heparin anticoagulated whole blood or 65-70 30-50μl blood spots on Whatman (Schleicher & Schuell) 903 or Ahlstrom 226 paper would provide sufficient material for one circulation. Samples for use in the scheme should be accompanied by a short clinical history and confirmation that informed consent/local ethical approval (as required in the referring centre) for use of the sample has been obtained.

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