Diagnosis of Fatty Acid Oxidation Disorders by Quantitative Determination of Carnitine and Acylcarnitine

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List of Abbreviations

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ACP(s) Acylcarnitine profile(s)

AC(s) Acylcarnitine(s)

Arb Arbitary unit (flow unit)

4-AP 4-aminophenazone

BBB Blood brain barrier

CAT Carnitine acetyl transferase

CACT Carnitine/acylcarnitine translocase

CK Creatine kinase

CMR Cardiac magnetic resonance

CoQ10 Complex Q10 (respiratory chain)

CPT Carnitine palmitoyl transferase

DCs Dicarboxylic acid(s)

DBS Dried blood spot

DHA Docosahexaenoic acid

DNPH Dinitrophenylhydrazine

ESI Electrospray ionisation

ETF Electron transfer flavoprotein

FAB-MS/MS Fast atom bombardment mass spectrometry

FAODs Fatty acid oxidation disorders

FFAs Free fatty acids

GA Glutaric aciduria

GLDH Glutamate dehydrogenase

GO Glucose oxidase

HMG-CoA Hydroxy-3-methylglutaryl-coenzyme A

IEM Inborn error(s) of metabolism

ISTD Internal standard

LCADD Long chain acyl-CoA dehydrogenase deficiency

LCHAD Long chain L-3 hydroxyacyl CoA dehydrogenase deficiency

LDH Lactate dehydrogenase

LO Lactate oxidase

m/z Mass to charge ratio

MADD Multiple acyl CoA dehydrogenase deficiency

MCAD Medium chain acyl CoA dehydrogenase deficiency

MCD Muscle carnitine deficiency

MMA Methylmalonic aciduria

MS/MS Mass/mass spectrometry

MCT Medium chain triglycerides

OCTN2 Organic cation/ carnitine transporter novel 2

PCA Principal component analysis

PDH Pyruvate dehydrogenase

POD Peroxidase

RI Renal insufficiency

RF Radio frequency

RPE Retinal pigment epithelium

RT Retention time

SCADD Short chain acyl CoA dehydrogenase deficiency

SCD Secondary carnitine deficiency

SCHAD Short chain hydroxyacyl CoA dehydrogenase deficiency

SIDS Sudden infant death syndrome

SPSS Statistical package for social science

TFP Trifunctional protein

TIC Total ion current

VLCADD Very long chain acyl CoA dehydrogenase deficiency

VPA Valproic acid

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