Tests performed through our international collaboration

Molecular Studies for Inborn Errors of Metabolism

B	Inborn Errors of Metabolism			
1	PKU (Phenylketonurie)	Phenylalaninie hydroxylase	PAH	
2	SCAD	Acyl – CoA-dehydrogenase, short chain	ACADS	
3	LCHAD	Mitochondrial trifunctional protein, alpha subunit	HADHA	
4	MCAD	Acyl – CoA-dehydrogenase, Medium chain	ACADM	
5	VLCAD	Acyl – CoA-dehydrogenase, Very Long chain	ACADVL	
6	Glutaricacidaemia 2	Electron – transferring – flavor – protein, alpha polypeptide	ETFA	
7	Glutaricacidaemia 2	Electron – transferring – flavor – protein, beta polypeptide	ETFB	
8	Glutaricacidaemia 2c	Electron – transferring – flavor – protein, dehydrogenase	ETFDH	
9	Carnitine – palmtoyltrans-ferase 2 deficiency	Carnitine – palmitoyltrans – ferase 2	CPT 2	
10	X – ALD		ABCD1	
11	MVK	Mevalonatkinase	MVK	