Supplementary table 2: Diagnoses of patients with an inborn error of metabolism, neuromuscular or malformation syndrome.

			Number of patients
Inborn error of metabolism (n=64)	**Storage disorders**	GSD III	9
		GSD IIb (Danon disease)	6
		GSD II (Pompe disease)	9
		Mucopolysaccharide storage disorder	12
	Mitochondrial disorders	MELAS	2
		Leigh syndrome	1
		Sengers syndrome	2
		Congenital lactic acidosis	3
		Mitochondrial cytopathy	7
		Mitochondrial respiratory chain disorders	1

			Number of patients
	Disorders of fatty acid metabolism	Carnitine transporter deficiency	2
		VLCADD	2
	Disorder of glycosylation	Carbohydrate glycoprotein deficiency	2
	Disorder of amino acid metabolism	Glutaric acidemia type 3	1
	Other	Molybdenum cofactor deficiency	2
		Purine nucleoside phosphorylase deficiency	1
		Aicardi-goutieres syndrome	1
		Kabuki Syndrome	1
Neuromuscular disease (n=64)	**Friedreich's Ataxia**	Friedreich’s Ataxia	59
	Other neuromuscular disease (n=5)	Becker muscular dystrophy	1
		Emery Dreifuss muscular dystrophy	1

			Number of patients
RASopathy (n= 126)	**Noonan Syndrome**	Myofibrillar myopathy	1
		Downs syndrome	1
		Unknown	1
		Noonan syndrome	110
	Other RASopathy (n=16)	Costello syndrome	5
		Noonan syndrome with multiple lentigines	10
		Cardiofaciocutaneous syndrome	1

GSD = Glycogen Storage Disorder, MELAS= Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes, VLCADD= Very long-chain acyl-coenzyme A dehydrogenase deficiency