Supplemental table 2: SCO2 variants-phenotypes

allel 1	allel 2	phenotype	additional condition(s)	CNS features	PNS features	death (age)	publication
E140K	R6Qfs	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1				3 mo	Kairit et al
E140K	W36X	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1				< 1 mo	Verdijk et al
E140K	Q53X	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Lactic acidosis	Gliosis, atrophic brain		2 mo	Papadopoulou et al
E140K	R90X	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Severe metabolic acidosis			0.5-1 mo	Jacksch et al
E140K	C133Y	Spinal muscular atrophy type I	Cardiomyopathy, hypotonia, persistent lactic acidosis , respiratory distress			1 mo	Tarnopolsky et al
E140K	E140K	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Stridor and respiratory insufficiency, ptosis, strabismus	Spasticity and psychomotor retardation	Axonal degeneration and demyelination	~10 mo	Pronicki et al Versela et al Jaksch M et al
E140K	L151P	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1				birth	Sacconi et al
E140K	V160G	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Severe neutropenia	Seizures		4 mo	Knuf et al
E140K	R171Trp	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Respiratory insufficiency, enlarged liver, edema	Brain edema		4 mo	Jacksch et al
E140K	M177T	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Ptosis, strabismus	Spasticity, psychomotor retardation		4 yo	Pronicki et al
G193S	G193S	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Hepatomegaly, metabolic acidosis, respiratory insufficiency, decreased tone and muscle bulk, wrist flexion, decreased grasp strength, weak suck reflex, significant head lag, dystonic arching of the back			1 mo	Mobley et al
E140K	10-BP DUP, NT1302	Spinal muscular atrophy	Resembled Werdnig-Hoffmann disease	Denervation due to loss of the anterior horn cells of the spinal cord		1.5 mo	Salviati et al
V160A	P233T	Fatal infantile hyperthermia				5-10 mo	Sambuughin et al
E140K	S225F	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1		Brain atrophy	Nerve conduction studies revealed a mixed axonal and demyelinating sensory motor neuropathy	6 mo	Papadopoulou et al
E140K	P169T	Charcot-Marie-Tooth disease	Ptosis and strabismus		Motor predominant polyneuropathy	alive (5)	
D135G	R171Q	Charcot-Marie-Tooth disease			Motor and sensory polyneuropathy	alive (22)	