Vondráčková et al. Large copy number variations in combination with point mutations in the *TYMP* and *SCO2* genes found in two patients with mitochondrial disorders

Supplementary Tables

Table 1: Hemizygous genes on 22q13.33 found in our patients

Gene (MIM no.)	Desciption of the gene	Phenotype of the disease (MIM no.)	Type of inheritance	Hemizygous in patient
LMF2 (-)	lipase maturation factor 2	-	-	Patient 1,2
NCAPH2 (611230)	condensin II complex, non-SMC subunit 2	-	-	Patient 1,2
SCO2 (604272)	cytochrome c oxidase assembly protein	fatal infantile cardioencephalomyopathy due to cytochromec oxidase deficiency (604377)	AR	Patient 1,2
TYMP (131222)	thymidine phosphorylase	mitochondrial DNA depletion syndrome 1 (603041)	AR	Patient 1,2
ODF3B (-)	outer dense fiber of sperm tails 3B	-	-	Patient 1,2
KLHDC7B (-)	kelch domain containing 7B	-	-	Patient 1,2
c22orf41 (-)	synaptonemal complex central element protein 3	-	-	Patient 1,2
CPT1B (601987)	muscle carnitine palmitoyltransferase 1B	-	AR in mice	Patient 1,2
CHKB (612395)	choline kinase beta	muscular dystrophy, congenital megaconial type (602541)	AR	Patient 1 (partialy), patient 2
LOC10144603 (-)	-	-	-	Patient 2
MAPK8IP2 (607755)	mitogen-activated protein kinase 8 - interacting protein 2	-	-	Patient 2
ARSA (607574)	arylsulfatase A	metachromatic leukodystrophy (250100)	AR	Patient 2

Table 2: Clinical features of 12 Czech and 44 patients reported in the literature with SCO2 deficiency.

	Czech patients (n=9) non-neonatal presentation	**Czech patients (n=3) neonatal onset**	**Reported cases in the literature (n=25) non-neonatal presentation**	**Reported cases in the literature (n=19) neonatal onset**
Reference	current report and [1, 2]	[3]	[4, 5, 6, 7, 8¶]	[4, 6, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20]
Gender (male/female)	4/5	2/1	10/14£	10/9
Onset of symptoms	3.3 ± 1.5 months (1.5 - 6)	< 1 months	5 ± 2.2 months (2 - 10)	< 1 months
Survival	none	none	5/25§	1/19£
Age of death	11.7 ± 7 months (4 - 28)	< 3 months	17 ± 15 months (5 - 60 months)	2.7 ± 2.5 months (25 days - 11 months)
IUGR	0/9 (0)	0/2 (0)	1/4Ω	3/18 (17)
Prematurity	0/9 (0)	1/3 (33)	0/4Ω	1/18 (6)
Inspiratory stridor	5/9 (56)	2/3 (67)	13/21 (62)	7/11 (64)
Hypotonia	8/9 (89)∗	3/3 (100)	25/25 (100)Ψ	19/19 (100)
Leigh/Leigh-like syndrome	2/9 (22)	0/3 (0)	7/22 (32)	6/17 (35)
Psychomotor delay/regression	9/9 (100)	n.a	23/25 (92)	n.a
Movement disorder	3/9 (34)	n.a	0/20 (0)	n.a
Ataxia	1/9 (11)	n.a	0/20 (0)	n.a
Seizures	1/9 (11)	1/3 (33)	5/21 (24)	7/16 (44)
Ptosis	2/9 (22)	0/3 (0)	7/22 (32)	1/12 (8)
HCMP	4/9 (44)	3/3 (100)	14/23 (61)	19/19 (100)
SMA-like phenotype	4/9 (44)	n.d	12/21 (57)	7/11 (64)

clinical features are expressed in % in brackets

abbreviations: n.a: not applicable ; n.d: not determined; HCMP: hypertrophic cardiomyopathy; SMA: spinal muscular atrophy

¶ Sambuughin et al: all three patients suffered from infantile hyperthermia

£ not reported in one patient

§ survival or age of death is not reported in four cases; only one patient was still alive at the time of publication (25 months old girl)

Ω not reported in majority of published non-neonatal cases

∗ acral spasticity in one patient

Ψ in many patients axial hypotonia was combined with acral spasticity (the exact number of patients was not further specified in original paper - Pronicki et al. 2010)

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