Table S2. Individuals excluded from comparison

Individual	**15**	**16**	**17**
Reference	Del Rizzo et al., 2013	Garret et al., 1998	Barbot et al., 1995
General information			
Ethnicity	Italian	Dutch	Portuguese
***SUOX* variant (NM_000456.3)**	c.[427C>A];[427C>A]	c.[479G>A];[479G>A]	NA
Effect on protein (NP_000447.2)	p.[(His143Asn)];[(His143Asn)]	p.[(Arg160Gln)];[(Arg160Gln)]	NA
Genetic testing method	NA	NA	NA
Sulfite oxidase activity in fibroblasts	NA	absent	absent
Sulfitest	positive	NA	1) negative, 2) positive (6 y)
Plasma homocysteine	< 1 µmol/L	NA	NA
Gender	F	F	F
Age at onset	1 m	5 m	3 m
Age at diagnosis	12 m	NA	6 y
Age at last evaluation	2 y 6 m	5 y	7 y
Death	NA	NA	NA
Family history	none	none	none
Consanguineity	no	yes	yes (first cousins)
Birth			
Gestational week	NA	NA	term
Neonatal period	unremarkable	NA	unremarkable
Central Nervous System			
Symptoms that led to specialist evaluation (age)	nystagmus (since 1 m), acute left hemiparesis (12 m)	two seizures (5 m)	loss of head control, irritability, sleep disorder during otitis media (3 m, but first evaluation at 9 m)
Global DD / ID after acute onset	yes (since first month of life)	yes (regression starting at 21 m)	DD / ID
Abnomal muscle tone	left hemiparesis	hypertonia	hypotonia
Movement disorder (Hyperkinesia or dystonia)	no	choreo-athetoid movements, dystonia	dystonia, hyperkinetic movements
Nystagmus	yes (since first month of life)	NA	no
Ataxia	no	yes	yes
Seizures	no	yes	no
EEG anomalies (at age)	normal (NA)	NA	normal (NA)
Neuroradiological abnormalities (at age)	MRI: mild cerebral atrophy and asymmetric stroke-like lesions of the globus pallidus (12 m); later, no significant brain atrophy, globus pallidus lesions well delineated and reduced in size (2 y)	globus pallidus changes, basal ganglia calcification, vermian hypoplasia, cerebellar atrophy (NA)	symmetrical hyperintensity of the globus pallidus, enlarged cerebello-medullary cystern with inferior vermis hypoplasia (6 y)
Behavioral anomalies	no	irritability at night	no
Organs and Systems			
Visual anomalies	no	ectopia lentis (2 y)	no
THERAPY	clinical and biochemical improvement under dietary treatment	NA	diet treatment with low levels of organic and inorganic sulphur for 2 months