	Our patient	Pollitt *et al*. [1] Gray *et al.* [20] Chambliss *et al*. [2]	Shield *et al*. [21] Sass *et al*. [3]	Sass *et al.* [3]
Patient information	Female Mixed European ancestry Parents non-consanguineous	Male Pakistani ancestry Consanguinity not specified	Female Pakistani ancestry Parents consanguineous	Male Mixed European ancestry Parents consanguineous
***ALDH6A1* mutations**	c.514T>C c.1603C>T	c.1336G>A c.1336G>A	c.184C>T c.184C>T	c.785C>A c.785C>A
Biochemical abnormalities	↑ plasma/urine MMA ↑ lactate ↑ HIBA ↑ AIBA ↑ β-alanine	↑ methionine ↑ 3-hydroxyproprionate ↑ HIBA ↑ AIBA ↑ β-alanine	↑ 3-hydroxypropionate ↑ methylcitrate Mild ↑ lactate ↑ HIBA ↑ AIBA	Mild ↑ 3-hydroxyproprionate ↑ HIBA ↑ β-alanine
CNS findings	Severe developmental delays, dystonia and microcephaly. Delayed myelination and thin corpus callosum on MRI	Normal development No imaging reported	Severe developmental delays, hypotonia and microcephaly. Delayed myelination and thin corpus callosum on MRI	Early delays, corrected by 25 months. Relative microcephaly. No imaging reported. Frontal cortex microcalcifications on autopsy.
Clinical findings	Tall forehead, epicanthal folds, mild hypertelorism, short philtrum, broad halluces, right single palmar crease.	Healthy No reported dysmorphisms	Microphthalmia and cataracts, diagnosed as Warburg Micro Syndrome. Narrow, down-slanting palpebral fissures, short nose, depressed nasal bridge.	Bulbous nose, long philtrum. Died at 26 months from hepatoencephalopathy and liver failure following a febrile illness.