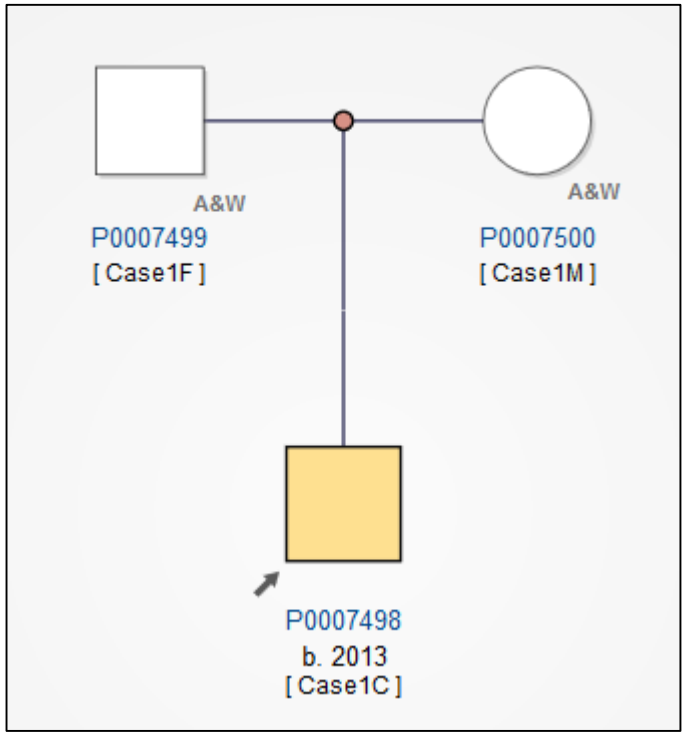
Case 1: description

RD-Connect identifier: Case1C

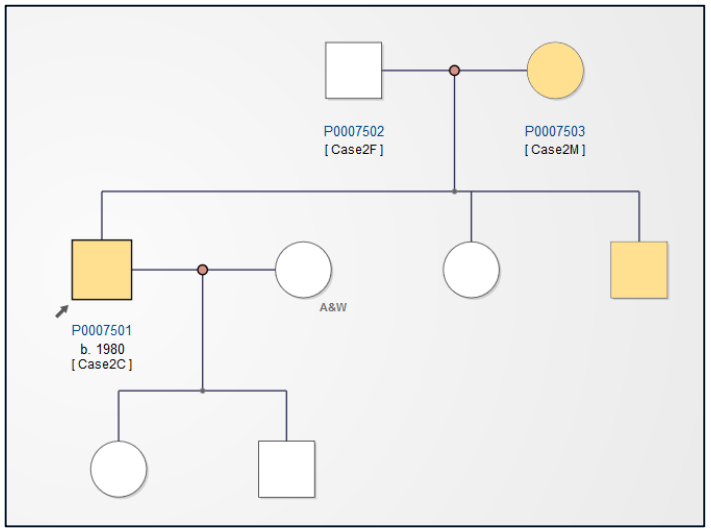
Gender	Male
Age	5 years
Referral	Congenital myasthenic syndrome
Onset	Congenital
Global pace of progression	Progressive (slow)
Main clinical features	• Neonatal hypotonia • Distal arthrogryposis • Inability to walk • Recurrent lower respiratory tract infections



Case 2: description

RD-Connect identifier: Case2C

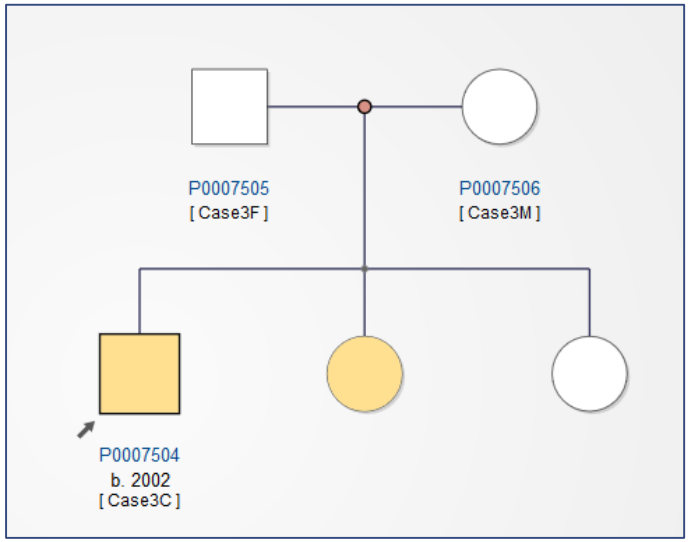
Gender	Male
Age	38 years
Referral	Macular dystrophy
Onset	Adult
Global pace of progression	Progressive
Main clinical features	• Progressive visual loss • Scotoma • Abnormality of retinal pigmentation



Case 3: description

RD-Connect identifier: Case3C

Gender	Male
Age	16 years
Referral	Muscular dystrophy
Onset	Juvenile
Global pace of progression	Progressive



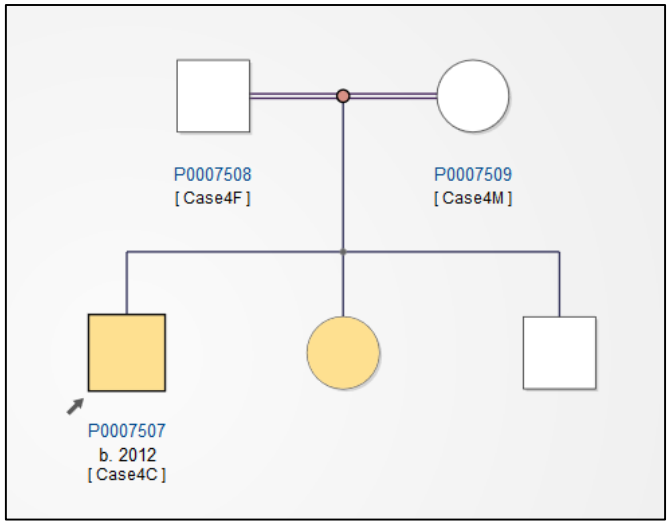
Main clinical features

- Muscle weakness
- Dystrophic muscle biopsy
- Quadriceps muscle atrophy
- Myalgia

Case 4: description

RD-Connect identifier: Case4C

Gender	Male
Age	6 years
Referral	Mitochondrial disorder
Onset	Infantile
Global pace of progression	Progressive
Main clinical features	• Microcephaly • Epileptic encephalopathy • Intellectual disability • Motor deterioration

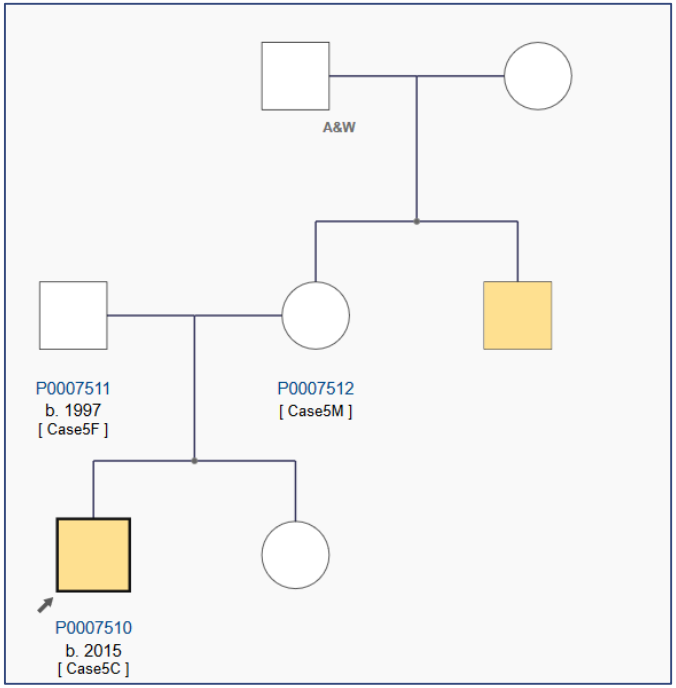


Consanguinity

Case 5: description

RD-Connect identifier: Case5C

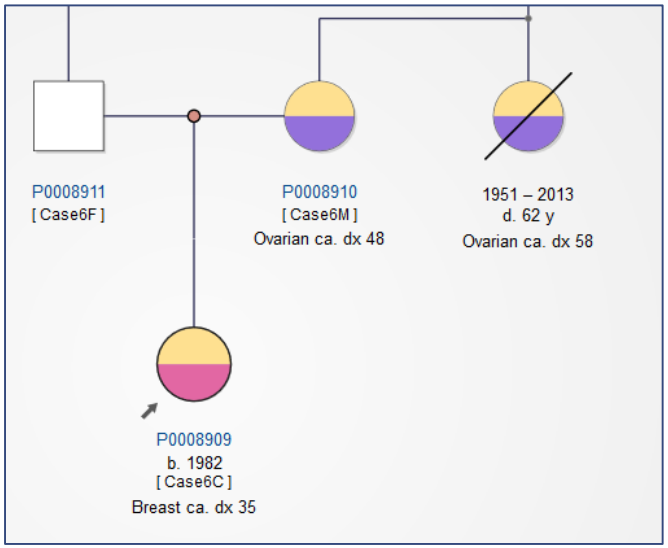
Gender	Male
Age	3 years
Referral	Muscular dystrophy
Onset	Childhood
Global pace of progression	Progressive
Main clinical features	• Muscle cramps • Seizures • Paresthesia • Renal insuficienty



Case 6: description

RD-Connect identifier: Case6C

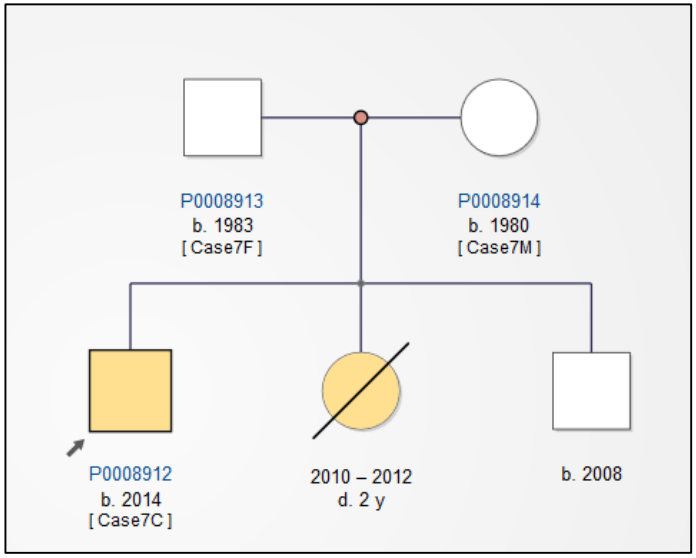
Gender	Female
Age	35 years
Referral	Breast Cancer
Onset	Adult
Main clinical features	• Breast carcinoma • Neoplasm of the breast • NO Decreased fertility • NO Colon cancer



Case 7: description

RD-Connect identifier: Case7C

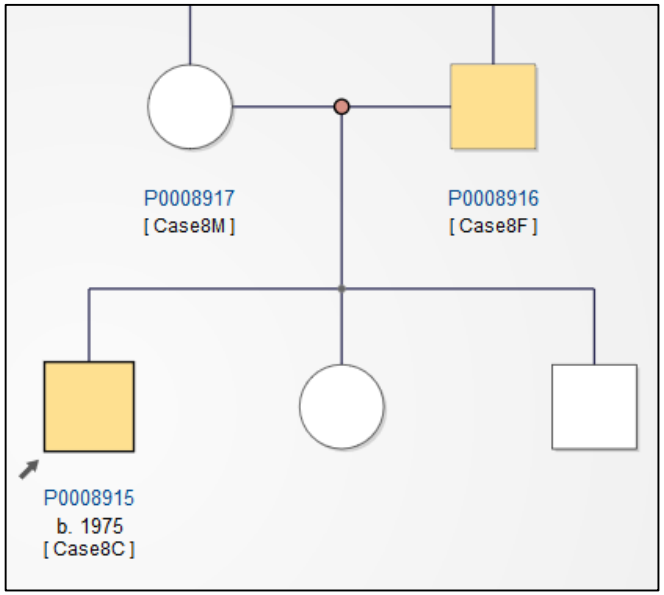
Gender	Male
Age	3 years
Referral	Inherited cancer-predisposing syndrome
Onset	Childhood
Main clinical features	• Neoplasm of the colon • Leukocytosis • Lymphoma • Basal cell carcinoma • Cafe-au-lait spot



Case 8: description

RD-Connect identifier: Case8C

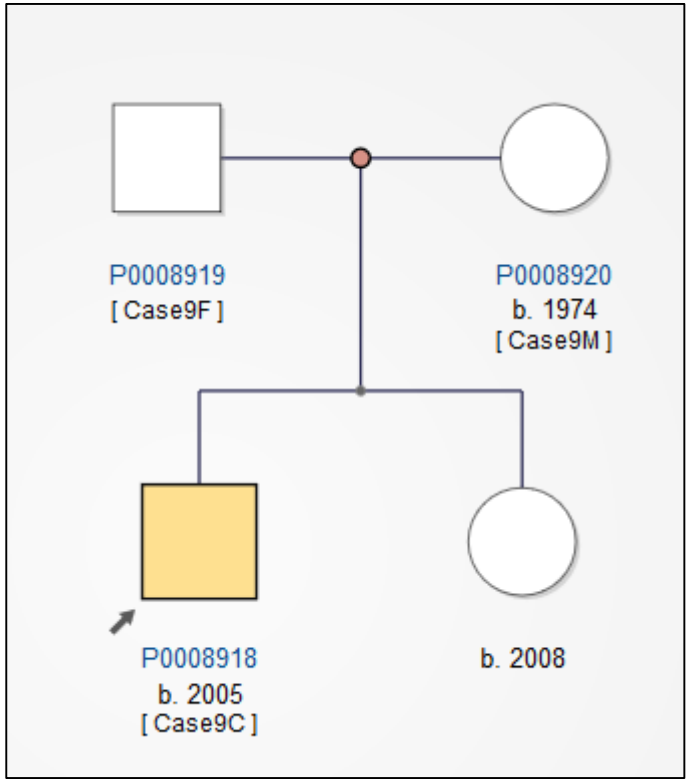
Gender	Male
Age	43 years
Referral	Inherited digestive cancer-predisposing syndrome
Onset	Adult
Main clinical features	• Multiple lipomas • Colon cancer • Multiple gastric polyps • Adenocarcinoma of the colon • Fibrosarcoma



Case 9: description

RD-Connect identifier: Case9C

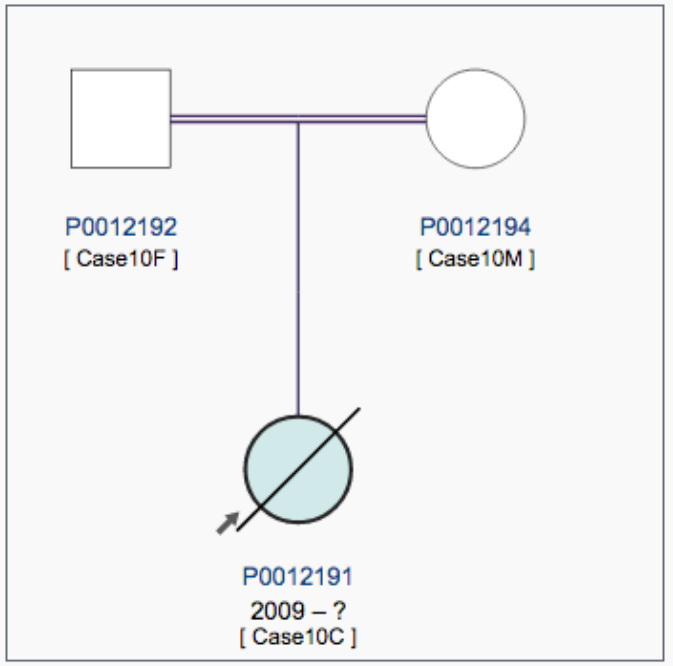
Gender	Male
Age	13 years
Referral	Inherited cancer-predisposing syndrome
Onset	Juvenile
Main clinical features	• Neurofibromas • Neurofibrosarcoma • Cafe-au-lait spot • Hypertelorism



Case 10: description

RD-Connect identifier: Case10C

Gender	Female
Age	Deceased 2 years
Referral	Lipoic acid synthesis defect?

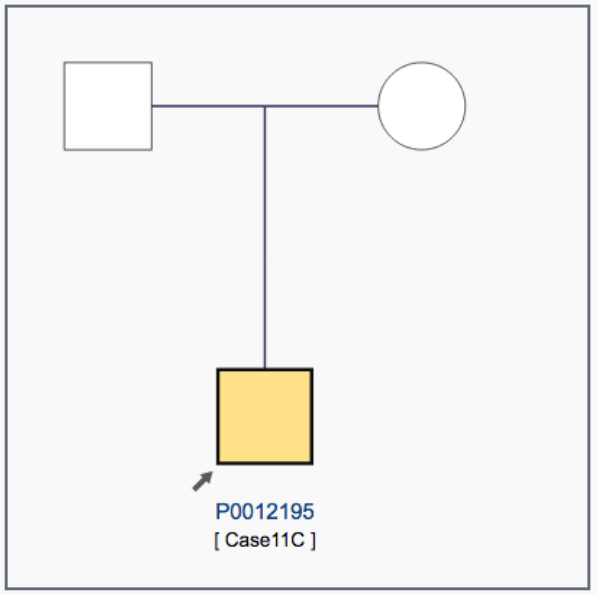


Onset	2 months
Main clinical features	• Growth retardation • Hypotonia • Microcephaly • Encephalopathy • Lactic acidosis • MRI: white matter atrophy and demyelination • Low PDH activity • No microscopic alterations (muscular biopsy)

Case 11: description

RD-Connect identifier: Case11C

Gender	Male
Age	2 months
Referral	Hyperammonemia

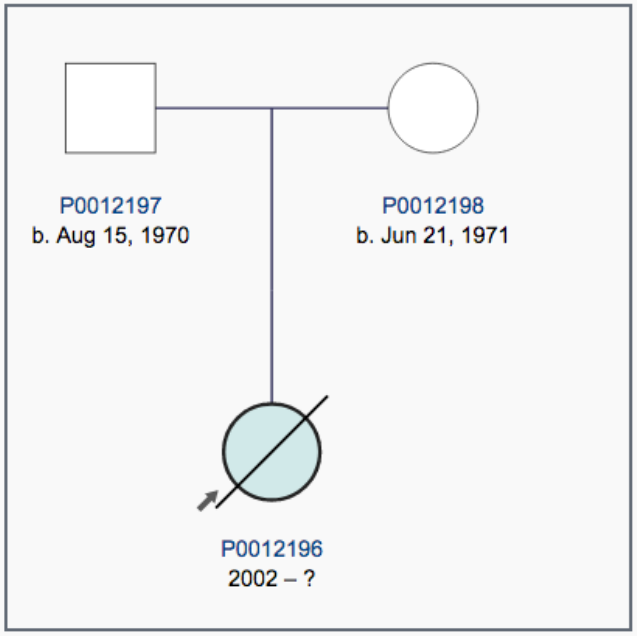


Onset	2 months
Main clinical features	• Hyperammonemia • Vomiting • Diarrhoea • Aminoacids and orotic acid suggestive of urea cycle disorder • Previous episodes of vomiting and diarrhoea

Case 12: description

RD-Connect identifier: Case12C

Gender	Female
Age	Died at 1 year of age due to apnoea
Referral	Mitochondrial disease

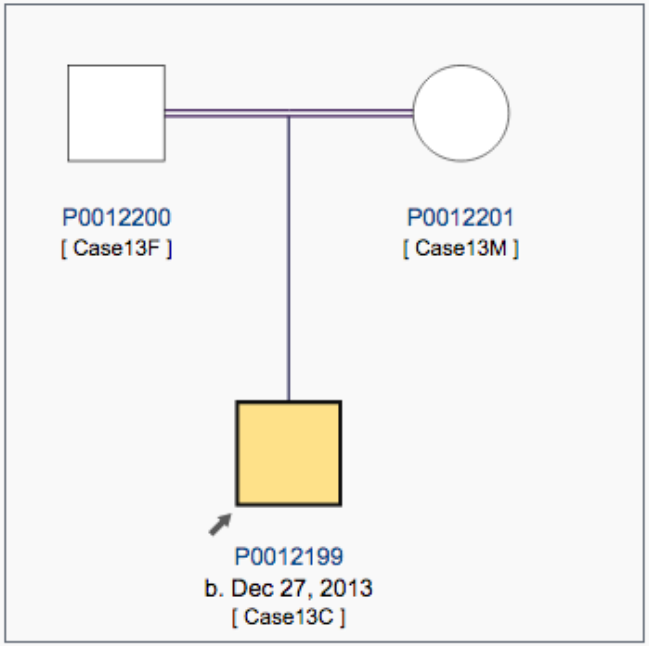


Onset	First month of age
Main clinical features	• Microcephaly • Retinopathy • Dilated cardiomyopathy • Generalyzed hypotonia • Myopathy • Seizures • Cataracts • High lactate and slight elevation of 3-methylglutaconic acid • Low complex IV activity and normal complexes II and III activities • MRI: white matter microhemorrhages

Case 13: description

RD-Connect identifier: Case13C

Gender	Male
Age	1,5 years of age
Referral	Energy metabolism alteration

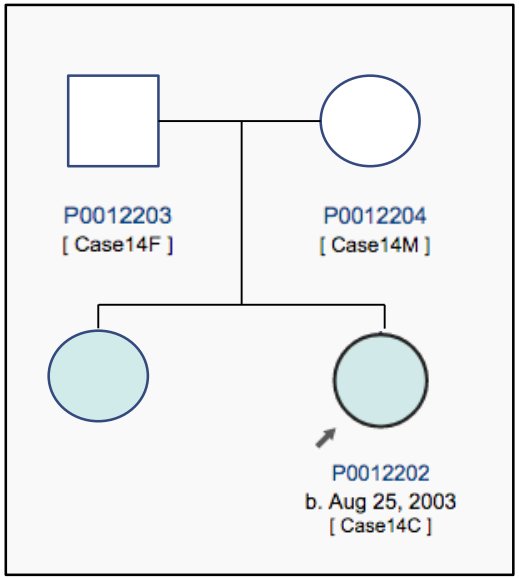


Onset	Neonatal
Main clinical features	• Neonatal hypotonia • Lower limbs spasticity • Nuclear punctate cataracts both eyes • Poor head control • Generalized developmental delay with psychomotor regression. Absence of language • High lactate in LCR, normal organic acids, Aminoacids, acylcarnitines, neurotransmitters. CoQ10 deficiency • Low pyruvate oxidation rates, low PDH

Case 14: description

RD-Connect identifier: Case14C

Gender	Female
Age	13 years
Referral	Glucogenosis or mitochondrial beta-oxidation deficiency
Onset	Childhood
Main clinical features	• Rhabdomyolysis • Lower limb pain • Myoglobinuria after exercise • HyperCKemia



Case 15: description

RD-Connect identifier: Case15C

Gender	Male
Age	4 years
Referral	Metabolic diseases with epilepsy
Onset	Infantile onset
Global pace of progression	Progressive
Main clinical features	• Rhabdomyolysis • Metabolic acidosis • Seizures • Global developmental delay • Ventricular tachycardia