BM1207

Neuromuscular diseases
DMD
Myotonic dystrophy
Spinal muscular atrophy
Facioscapulohumeral muscular dystr

Neurodegenerative diseases
Parkinson's disease
Huntington's disease
Spinocerebellar ataxia
Frontotemporal dementia

Metabolic diseases
Lysosomal storage disorders
Hyperphenylalaninemias
Organic acidemias
Congenital defects of glycosylation
Familial hypercholesterolemia

Retinal dystrophies
Retinitis pigmentosa
Leber Congenital Amaurosis

Other disorders
Dystrophic Epidermolysis Bullosa
Ataxia telangiectasia
CADASIL
X-linked a-gammaglobulinemia

CA1703

Neuromuscular diseases
DMD
Myotonic dystrophy
Spinal muscular atrophy
Facioscapulohumeral muscular dystrophy
Limb-girdle muscular dystrophy
Congenital myasthenic syndrome

Neurodegenerative & neurological diseases
Parkinson's disease
Huntington's disease
Spinocerebellar ataxia
Frontotemporal dementia
Leukoencephalopaties with ataxias
Dementias
Fragile X syndrome
Epilepsy
Rett syndrome

Metabolic diseases
Lysosomal storage disorders
Hyperphenylalaninemias
Organic acidemias
Congenital defects of glycosylation
Type I diabetes

Retinal dystrophies
Retinitis pigmentosa
Leber Congenital Amaurosis
Inherited retinal disease

Cancer
B-cell lymphoma
B-cell leukemia
Lung Cancer
Colorectal cancer

Other disorders
Dystrophic Epidermolysis Bullosa
Inherited cardiomyopathies
Liver pathology
Hepatic encephalopathy
Inflammatory and autoimmune diseases
Chronic obstructive pulmonary disease
Circadian rhythms/sleep disorders
Osteogenesis Imperfecta
Infectious diseases
Dermatitis