LABORATORY:
Centre de Génétique Humaine - Erasme ULB

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EQA:	2016 DNA Sequencing – NGS (vGermline) EMQN 2017 DNA Sequencing – NGS (vGermline) EMQN 2018 DNA Sequencing – NGS (vGermline) EMQN 2019 DNA Sequencing – NGS (vGermline) EMQN 2020 DNA Sequencing – NGS (vGermline) EMQN 2021 DNA Sequencing – NGS (vGermline) EMQN 2022 DNA Sequencing – NGS (vGermline) EMQN 2023 DNA Sequencing – NGS (vGermline) EMQN 2015 Postnatal constitutional CNV detection (array) EMQN 2016 Postnatal constitutional CNV detection (array) EMQN 2017 Postnatal constitutional CNV detection (array) EMQN 2018 Postnatal constitutional CNV detection (array) EMQN 2019 Postnatal constitutional CNV detection (array) EMQN 2020 Postnatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2022 Postnatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2016 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2017 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2018 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2019 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2021 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2023 Prenatal constitutional CNV detection GenQA (Genomics Quality Assessment) 2015 Blood -postnatal GenQA (Genomics Quality Assessment) 2016 Blood -postnatal GenQA (Genomics Quality Assessment) 2017 Blood -postnatal GenQA (Genomics Quality Assessment) 2018 Blood -postnatal GenQA (Genomics Quality Assessment) 2019 Blood -postnatal GenQA (Genomics Quality Assessment) 2020 Blood postnatal GenQA (Genomics Quality Assessment) 2021 Recurrent miscarriage karyotyping GenQA (Genomics Quality Assessment) 2022 Postnatal karyotyping GenQA (Genomics Quality Assessment) 2023 Postnatal karyotyping GenQA (Genomics Quality Assessment) 2015 Rapid Prenatal Aneuploidy-FISH GenQA (Genomics Quality Assessment) 2016 Rapid Prenatal Aneuploidy-FISH GenQA (Genomics Quality Assessment) 2017 Rapid Prenatal Aneuploidy-FISH GenQA (Genomics Quality Assessment) 2017 Molecular Rapid aneuploidy QF-PCR/MLPA/BoBs GenQA (Genomics Quality Assessment) 2018 Molecular Rapid aneuploidy QF-PCR/MLPA/BoBs GenQA (Genomics Quality Assessment) 2019 Molecular Rapid aneuploidy QF-PCR/MLPA/BoBs GenQA (Genomics Quality Assessment) 2020 Molecular Rapid aneuploidy QF-PCR/MLPA/BoBs GenQA (Genomics Quality Assessment) 2021 Rapid prenatal testing for common aneuploidies GenQA (Genomics Quality Assessment) 2022 Rapid prenatal testing for common aneuploidies GenQA (Genomics Quality Assessment)

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RELATED CONTENT

Related Genetic Tests

- Achondroplasia (hot spot mutation - p.Gly380)
- Albright hereditary osteodystrophy
- Algrove syndrome (Triple A syndrome)
- Alzheimer disease (gene panel)
- Amyloidosis hereditary (gene panel)
- Angelman / Prader Willi Syndrome
- Ataxia (autosomic dominant and recessive / except expansion of triplets) (gene panel - 722 genes)
- Azoo-/oligozoospermia (microdeletion of 3 regions of Y-chromosome AZFa, b and c)
- Beta-globin hemoglobinopathies
- Beta-globin hemoglobinopathies, Sickle cell anemia, Sickle cell disorder (hot spot mutation - p.Glu6Val, p.Glu6Lys)
- Beta-globin hemoglobinopathies, phenotype modifiers (hot spot mutations - rs7482144 (Xmn1) at promoter 158 bp 5? upstream of HBG2 / 32C-T in the 5' UTR of the HBS1L)
- Brain malformations (gene panel)
- Breast and Ovarian cancer, HBOC, familial (gene panel - 17 genes)
- Charcot-Marie-Tooth (CMT1A, GJB1)
- Congenital malformation (gene panel - 1721 genes)
- Cystic Fibrosis / Congenital absence of the vas deferens / CFTR-related disorders (88 hot spot mutations)
- Cystic Fibrosis, newborn screening (12 hot spot mutations; CFTR)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Dyslipidemia (gene panel)
- Early onset epileptic encephalopathy (gene panel - 845 genes)
- Endocrine Disorders - Hyper(Hypo)parathyroidism (gene panel - 24 genes)
- Endocrine Disorders - Hypothyroidism (gene panel - 42 genes)
- FSHR - Ovarian Hyperstimulation Syndrome
- Fragile X syndrome/POF/FXTAS - CGG repeat expansion
- Frequent hearing deficiency (4 genes)

- Gilbert syndrome / Irinotecan sensitivity (homozygous A(TA)7TAA allele) - Pharmacogenetics
- Hemochromatosis (17 genes)
- Hemochromatosis hereditary type 1 (hot spot mutations - p.Cys282Tyr; p.His63Asp)
- Hereditary Hemolytic Anemias due to unknown or doubtful origin (gene panel - 52 genes)
- Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)
- Hereditary spastic paraplegia (gene panel - 249 genes)
- Huntington disease - CAG repeat expansion
- Hyperthyroidism (familial gestational or familial nonautoimmune, hypothyroidism, thyrotropin) - TSHR
- Hypochondroplasia (full sequencing)
- Leydig cell hypoplasia or Precocious puberty, male-limited
- Mc Cune Albright syndrome
- Multiple Endocrine Neoplasia type 2A and 2B / Familial medullary thyroid carcinoma
- Neurodevelopmental disorders (1300 genes)
- Neuromuscular disorders (548 genes)
- Pancreatic cancer (12 genes)
- Pancreatitis, hereditary (7 genes)
- Periodic Fever (88 genes)
- Phenylketonuria
- Porphyria (10 genes)
- Prostate cancer susceptibility (7 genes)
- Pulmonary Arterial Hypertension / Rendu Osler Weber disease (gene panel - 24 genes)
- Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4
- Thalassemia Alpha (2 genes)
- Thiopurine S-Methyltransferase deficiency - TPMT genotyping c.238G>C (rs1800462); c.460G>A (rs1800460); c.719A>G (rs1142345)) - Pharmacogenetics
- Torsion dystonia 1 (hot spot mutation - c.907_909delGAG)

Related Diseases

- Aceruloplasminemia
- Achondroplasia
- Acquired schizencephaly

- Acrocallosal syndrome
- Acrodermatitis continua of Hallopeau
- Acute intermittent porphyria
- Alobar holoprosencephaly
- Angelman syndrome due to imprinting defect in 15q11-q13
- Angelman syndrome due to maternal 15q11q13 deletion
- Angelman syndrome due to paternal uniparental disomy of chromosome 15
- Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation
- Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis
- Autosomal dominant hypocalcemia
- Autosomal dominant preaxial polydactyly-upperback hypertrichosis syndrome
- Autosomal recessive primary microcephaly
- Autosomal recessive sideroblastic anemia
- Azathioprine or 6-mercatopurine toxicity or dose selection
- Behavioral variant of frontotemporal dementia
- Behçet disease
- Beta-thalassemia intermedia
- Beta-thalassemia major
- Blau syndrome
- CANDLE syndrome
- CINCA syndrome
- Charcot-Marie-Tooth disease type 1A
- Cherubism
- Cholesteryl ester storage disease
- Classic phenylketonuria
- Combined pituitary hormone deficiencies, genetic forms
- Complete hydatidiform mole
- Congenital atransferrinemia
- Congenital bilateral absence of vas deferens
- Congenital communicating hydrocephalus
- Congenital dyserythropoietic anemia type I
- Congenital dyserythropoietic anemia type II
- Congenital erythropoietic porphyria

- Congenital non-communicating hydrocephalus
- Cystic fibrosis
- DITRA
- Delta-beta-thalassemia
- Desmoplastic/nodular medulloblastoma
- Diamond-Blackfan anemia
- Disseminated superficial actinic porokeratosis
- Dowling-Degos disease
- Ear-patella-short stature syndrome
- Early-onset autosomal dominant Alzheimer disease
- Early-onset generalized limb-onset dystonia
- Encephalocraniocutaneous lipomatosis
- FTH1-related iron overload
- Familial GPIHBP1 deficiency
- Familial Mediterranean fever
- Familial apolipoprotein A5 deficiency
- Familial apolipoprotein C-II deficiency
- Familial cold urticaria
- Familial gestational hyperthyroidism
- Familial hyperthyroidism due to mutations in TSH receptor
- Familial hypocalciuric hypercalcemia type 1
- Familial hypocalciuric hypercalcemia type 2
- Familial hypocalciuric hypercalcemia type 3
- Familial lipase maturation factor 1 deficiency
- Familial lipoprotein lipase deficiency
- Familial medullary thyroid carcinoma
- Familial multiple meningioma
- Familial pancreatic carcinoma
- Familial peripheral male-limited precocious puberty
- Familial porphyria cutanea tarda
- Familial prostate cancer
- Fragile X syndrome
- Fragile X-associated tremor/ataxia syndrome

- Generalized pustular psoriasis
- Genetic hyperferritinemia without iron overload
- Gilbert syndrome (NON RARE IN EUROPE)
- Gorlin syndrome
- HJV or HAMP-related hemochromatosis
- Hartsfield syndrome
- Hb Bart's hydrops fetalis
- Hemochromatosis type 1 (NON RARE IN EUROPE)
- Hemochromatosis type 4
- Hemoglobin C disease
- Hemoglobin C-beta-thalassemia syndrome
- Hemoglobin D disease
- Hemoglobin E disease
- Hemoglobin E-beta-thalassemia syndrome
- Hemoglobin H disease
- Hemoglobin Lepore-beta-thalassemia syndrome
- Hemoglobin M disease
- Hemoglobinopathy Toms River
- Hepatoerythropoietic porphyria
- Hereditary breast cancer
- Hereditary chronic pancreatitis
- Hereditary coproporphyria
- Hereditary hyperferritinemia-cataract syndrome
- Hereditary neuropathy with liability to pressure palsies
- Hereditary pediatric Behçet-like disease
- Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome
- Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome
- Heritable pulmonary arterial hypertension
- Homozygous familial hypercholesterolemia
- Huntington disease
- Hyperimmunoglobulinemia D with periodic fever
- Hyperzincemia and hypercalprotectinemia
- Hypochondroplasia

- Hypothyroidism due to TSH receptor mutations
- IRIDA syndrome
- Idiopathic bronchiectasis
- Immune dysregulation-inflammatory bowel disease-arthritis-recurrent infections syndrome
- Infantile-onset periodic fever-panniculitis-dermatosis syndrome
- Intermittent hydrarthrosis
- Irinotecan toxicity
- JMP syndrome
- L-ferritin deficiency
- Leydig cell hypoplasia due to complete LH resistance
- Leydig cell hypoplasia due to partial LH resistance
- Lobar holoprosencephaly
- MASA syndrome
- Majeed syndrome
- McCune-Albright syndrome
- Meningioma
- Microcephalic osteodysplastic primordial dwarfism type II
- Microcytic anemia with liver iron overload
- Microform holoprosencephaly
- Midline interhemispheric variant of holoprosencephaly
- Muckle-Wells syndrome
- Multiple endocrine neoplasia type 2A
- Multiple endocrine neoplasia type 2B
- NLRP12-associated hereditary periodic fever syndrome
- Nakajo-Nishimura syndrome
- Neuroferritinopathy
- Non-syndromic metopic craniosynostosis
- Ovarian hyperstimulation syndrome
- PAPA syndrome
- PLCG2-associated antibody deficiency and immune dysregulation
- Partial chromosome Y deletion
- Partial hydatidiform mole
- Periodic fever-infantile enterocolitis-autoinflammatory syndrome

- Pfeiffer syndrome type 1
- Pituitary stalk interruption syndrome
- Pityriasis rubra pilaris
- Polydactyly of a triphalangeal thumb
- Polyglucosan body myopathy type 1
- Porokeratosis of Mibelli
- Porphyria due to ALA dehydratase deficiency
- Porphyria variegata
- Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome
- Prader-Willi syndrome due to imprinting mutation
- Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15
- Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1
- Prader-Willi syndrome due to paternal deletion of 15q11q13 type 2
- Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome
- Primary ovarian failure (NON RARE IN EUROPE)
- Progressive non-fluent aphasia
- Proximal spinal muscular atrophy type 1
- Proximal spinal muscular atrophy type 2
- Proximal spinal muscular atrophy type 3
- Proximal spinal muscular atrophy type 4
- Pseudohypoparathyroidism type 1A
- Pseudohypoparathyroidism type 1C
- Pseudopseudohypoparathyroidism
- Pustulosis palmaris et plantaris
- Radial hemimelia
- Rare autosomal recessive non-syndromic sensorineural deafness type DFNB
- Rendu Osler Weber
- Schilbach-Rott syndrome
- Seckel syndrome
- Semantic dementia
- Semilobar holoprosencephaly
- Septopreoptic holoprosencephaly
- Severe congenital hypochromic anemia with ringed sideroblasts

- Sickle cell anemia
- Sickle cell-beta-thalassemia disease syndrome
- Sickle cell-hemoglobin C disease syndrome
- Sickle cell-hemoglobin D disease syndrome
- Sickle cell-hemoglobin E disease syndrome
- Sitosterolemia
- Situs ambiguus
- Situs inversus totalis
- Sterile multifocal osteomyelitis with periostitis and pustulosis
- Symptomatic form of fragile X syndrome in female carriers
- Symptomatic form of hemochromatosis type 1
- Syndactyly type 4
- Systemic lupus erythematosus
- TFR2-related hemochromatosis
- Triphalangeal thumb-polysyndactyly syndrome
- Triple A syndrome
- Vasculitis due to ADA2 deficiency
- Wolman disease
- X-linked Charcot-Marie-Tooth disease type 1
- X-linked complicated corpus callosum dysgenesis
- X-linked complicated spastic paraplegia type 1
- X-linked erythropoietic protoporphyria
- X-linked sideroblastic anemia

Related Gene Panels

- Amyloidosis (3 genes) - ULB
- Ataxia (348 genes) - ULB
- Beta-globin hemoglobinopathies, phenotype modifiers (3 genes) - ULB
- Brain malformations (34 genes) - ULB
- Breast/Ovarian cancer (17 genes) - ULB
- Congenital malformation (1721 genes) - ULB
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB

- Dyslipidemia (13 genes) - ULB
- Early onset epileptic encephalopathy (845 genes) - ULB
- Endocrine Disorders - Hyper(Hypo)parathyroidism (24 genes) - ULB
- Endocrine Disorders - Hypothyroidism (42 genes) - ULB
- Hemochromatosis (17 genes) - ULB
- Hereditary Hemolytic Anemias (52 genes) - ULB
- Hereditary spastic paraplegia (188 genes) - ULB
- Neurodevelopmental disorders (1300 genes) - ULB
- Neuromuscular disorders (548 genes) - ULB
- Pancreas cancer (12 genes-) - ULB
- Pancreatitis (7 genes) - ULB
- Periodic Fever (88 genes) - ULB
- Porphyria (10 genes) - ULB
- Prostate cancer susceptibility (7 genes) - ULB
- Pulmonary Arterial Hypertension (24 genes) - ULB
- Thalassemia Alpha (2 genes) - ULB
