Parkinson Disease 13, Autosomal Recessive
Parkinson Disease 2, Autosomal Recessive Juvenile
Parkinson Disease 5, Autosomal Dominant
Parkinson Disease 6, Autosomal Recessive Early-Onset
Parkinson Disease 7, Autosomal Recessive Early-Onset
Parkinson Disease 8, Autosomal Dominant
Parkinson Syndrome
Parkinson Syndrome - NGS
Pelizaeus-Merzbacher Disease
Pendred Syndrome
Peutz - Jeghers Syndrome
Prader-Willi Syndrome
Primary familial brain calcification
Pseudoxanthoma Elasticum
Renal Tubular Acidosis, Distal, Autosomal Dominant
Renal Tubular Acidosis, Distal, With Hemolytic Anemia
Rendu - Osler - Weber Syndrome
Rett Syndrome
SHOX Deficiency Disorders
Spastic Ataxia, Charlevoix - Saguenay Type
Spastic Paraplegia
Spastic Paraplegia 11, Autosomal Recessive
Spastic Paraplegia 15, Autosomal Recessive
Spastic Paraplegia 17, Autosomal Dominant
Spastic Paraplegia 20, Autosomal Recessive
Spastic Paraplegia 3, Autosomal Dominant
Spastic Paraplegia 4, Autosomal Dominant
Spastic Paraplegia 44, Autosomal Recessive
Spastic Paraplegia 5A, Autosomal Recessive
Spastic Paraplegia 7, Autosomal Recessive
Spherocytosis, Type 4
Spinal And Bulbar Muscular Atrophy, X-Linked 1
Spinal Muscular Atrophy, Type I
Spinocerebellar Ataxia Type 1
Spinocerebellar Ataxia Type 10
Spinocerebellar Ataxia Type 12
Spinocerebellar Ataxia Type 14
Spinocerebellar Ataxia Type 15
Spinocerebellar Ataxia Type 17
Spinocerebellar Ataxia Type 2
Spinocerebellar Ataxia Type 27
Spinocerebellar Ataxia Type 3
Spinocerebellar Ataxia Type 36
Spinocerebellar Ataxia Type 5
Spinocerebellar Ataxia Type 6
Spinocerebellar Ataxia Type 7
Spinocerebellar Ataxia Type 8
Spinocerebellar Ataxia, Autosomal Recessive 1
SUFU Gene Associated Syndromes
Thrombophilia
Thrombophilia Due To Activated Protein C Resistance
Thyrotoxic Periodic Paralysis Type 1
Thyrotoxic Periodic Paralysis Type 2
TP53 Associated Diseases
TP63 Associated Diseases
Tuberous Sclerosis
Tuberous Sclerosis - NGS
Waardenburg Syndrome, Type I
Wilms tumor-aniridia-genital anomalies-retardation syndrome
Wilson Disease
Wiskott-Aldrich syndrome-2
Wolfram Syndrome Type 2
Yellow Nail Syndrome

In order to send samples for genetic analyses, contact the institute at the following email address:

consulenze@birdfoundation.org

Detailed information will be provided regarding the type of test requested, the analytical methods, the required samples and their handling and shipping conditions, the medical prescriptions (number and indications), and the estimated turnaround time.
In order to schedule a medical consultation or blood sampling, please contact the institute at the following phone number: ***(+39) 0444 555557***

We offer appointments from Monday to Friday from 8:30 to 12:30 and from 14:30 to 17:30.

Support with your 5x1000 our fight against rare diseases by marking the fiscal code 02767330240 in the box relative to the financing of scientific research in your tax form. Your aid will help improving our services and the research on rare diseases..

"Mauro Baschirotto" Institute for Rare Diseases

"Mauro Baschirotto" Institute for Rare Diseases
B.I.R.D. Foundation n.p.o.

Operates within the national healthcare system, licensed with the Decree of the Regional Executive No. 149 of the 03/08/1999 and accredited with the Decree of the Regional Executive No. 769 of the 12/07/2002 as Out-of-hospital Center for the Diagnosis, Treatment and Cure of the Rare Diseases, and currently accredited with the Decree of the Regional Executive No. 2711 of the 29/12/2014 as Medical Genetics Laboratory.

Public Service Charter

"Mauro Baschirotto" Institute for Rare Diseases

(synthetic)

Mauro Baschirotto" Institute for Rare Diseases – B.I.R.D. Foundation
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The "Mauro Baschirotto" Institute for Rare Diseases (B.I.R.D.) was established by the nonprofit Foundation and Association for Rare Diseases “Mauro Baschirotto”. The institute operates within the national healthcare system, recognized by the Decree of the Venetian Regional Executive No. 149 of the 03/08/1999 and accredited with the Decree of the Regional Executive No. 769 of the 12/07/2002 as Out-of-hospital Center for the Diagnosis, Treatment and Cure of the Rare Diseases and currently with the Decree of the Regional Executive No. 2711 of the 29/12/2014 as Medical Genetics Laboratory.

The main goal of the institute is to offer an answer to the needs of the patients affected by a scarcely known disease. With time, our structure, situated in Costozza di Longare near Vicenza (Italy), is becoming a highly appreciated reference center as demonstrated by the numerous requests from patients coming from all over Italy and from abroad.
The Institute in particular offers prenatal and postnatal diagnoses in the Molecular Genetics Laboratory of the Institute that offers a wide range of molecular tests for rare diseases and research projects that space form basic to clinical research and include the implementation of new diagnostic approaches as well as the discovery of new potential cures for rare diseases.

Out-patient rehabilitation programs are performed by several professional figures necessary for the novel protocols developed for individuals and groups of patients with the same pathology; Furthermore the institute offers poly-specialist outpatient visits involving different medical specialists with in-house hospitality facilities for patients and their family members / accompanying persons; Finally an education program is running to train doctors and healthcare professionals to help patients and their families in the management of the rare diseases.

Our institute offers a variety of services for patients with rare disease and their families.

Our Services include:
- *Genetic diagnosis of the listed diseases within the Italian National Healthcare System;*
- *Ambulatory visits and genetic counselling for the diagnosis of genetic diseases and the determination of reproductive risks;*
- *Out-house physiatrist visits for the definition of rehabilitation treatments;*
- *Out-house rehabilitation treatments;*

Ultrasound scans for internal medicine, joints, thyroid gland and soft parts of the body.

Genetic tests available in the Molecular Genetics Laboratory

Adrenoleukodystrophy
Alpha-1 Antitrypsin Deficiency
Alzheimer Disease Type 1
Alzheimer Disease Type 2 Linked To Apoe
Alzheimer Disease Type 3
Alzheimer Disease Type 4
Alzheimer Syndrome and Frontotemporal Dementia
Amyotrophic Lateral Sclerosis
Amyotrophic Lateral Sclerosis 1
Amyotrophic Lateral Sclerosis 4, Juvenile
Amyotrophic Lateral Sclerosis With Frontotemporal Dementia 1
Andersen-Tawil Syndrome
Angelman Syndrome
Aniridia
Ataxia With Oculomotor Apraxia Type 2
Ataxia, Early-Onset, With Oculomotor Apraxia And Hypoalbuminemia
Autoimmune Polyendocrine Syndrome Type 1
Autosomic Recessive Spinocerebellar Ataxias
Bartter Syndrome Type 3
Basal Cell Nevus Syndrome
Beckwith-Wiedemann Syndrome
Berardinelli-Seip Congenital Lipodystrophy Type 2
Bietti Crystalline Corneoretinal Dystrophy
Brugada Syndrome
CADASIL/CARASIL - NGS, Syndrome
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy
Charcot - Marie - Tooth Disease, type 2J
Charcot - Marie - Tooth Disease, Type 1B
Charcot - Marie - Tooth Disease, Type 2A2
Charcot - Marie - Tooth Disease, Type 2D
Charcot - Marie - Tooth Disease, X-Linked, 1
Charcot-Marie-Tooth Disease, Demyelinating, Type 1a
Chorea, Benign Hereditary
Corneal Dystrophy Linked To TGFBI
Corneal Dystrophy, Avellino Type
Corneal Dystrophy, Groenouw Type I
Corneal Dystrophy, Reis - Bucklers Type
Corneal Dystrophy, Thiel - Behnke Type
Creutzfeldt - Jakob Disease
Creutzfeldt-Jakob Disease
Cystic Fibrosis
Darier - White Disease
Deafness, Aminoglycoside-Induced
Deafness, Autosomal Recessive
Deafness, Autosomal Recessive 1A
Deafness-Dystonia-Optic Neuronopathy Syndrome
Dentatorubral-Pallidoluysian Atrophy
Diabetes Insipidus, Nephrogenic, Autosomal
Diabetes Insipidus, Nephrogenic, X-Linked
Dystrophia Myotonica 1
Epileptic Encephalopathy, Early Infantile, 2
Familial Cerebral Cavernous Malformation - NGS
Farber Lipogranulomatosis
Fatal Familial Insomnia
Friedreich Ataxia 1
Frontotemporal Dementia
Gardner Syndrome
Gitelman Syndrome
Glycogen Storage Disease Type V
Hemochromatosis
Hemochromatosis, Juvenile
Hemochromatosis, Type 3
Hemochromatosis, Type 4
Hepatic Venoocclusive Disease With Immunodeficiency
Hereditary Hemorrhagic Telangiectasia
Hereditary Hemorrhagic Telangiectasia - NGS
Hereditary Hemorrhagic Telangiectasia Type 1
Hereditary Hemorrhagic Telangiectasia Type 2
Hereditary Neuropathy With Liability To Pressure Palsies
Hereditary Spastic Paraplegia, Pannel 1
Hereditary Spinocerebellar Ataxias 1,2,3,6,7,8,10,12,17,36,DRPLA
Heritable Pulmonary Arterial Hypertension
Homocysteinemia
Homocystinuria Due To Deficiency Of N(5,10)-Methylenetetrahydrofolate Reductase Activity
Huntington Disease
Huntington Disease-Like 2
Hyperferritinemia - Cataract Syndrome
Hyperimmunoglobinemia D with recurrent fever
Hyperkalemic Periodic Paralysis Type 1
Hypokalemic Periodic Paralysis
Hypokalemic Periodic Paralysis Type 1
Hypokalemic Periodic Paralysis Type 2
Hypomyelinating Leukodystrophy 2
Indirect test using STRs
Krabbe Disease
Leber Optic Atrophy
Lesch-Nyhan Syndrome
Leukodystrophy type 2, Hypomyelinating
Leukoencephalopathy with mild cerebellar ataxia and white matter edema
Li-Fraumeni Syndrome 1
Li-Fraumeni Syndrome 2
Lipodystrophy, Congenital Generalized, Type 1
Lymphedema-Distichiasis Syndrome
Malignant Hyperthermia - NGS, Susceptibility To
Malignant Hyperthermia, Susceptibility To, 1
Malignant Hyperthermia, Susceptibility To, 5
Mast Syndrome
Menkes Syndrome
Mental Retardation, X-Linked, ARX-Related
Metachromatic Leukodystrophy
Mevalonic Aciduria
Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes
Multiple Sulfatase Deficiency
Myoclonic Epilepsy Associated With Ragged-Red Fibers
Myotonia Congenita (Becker), Autosomal Recessive
Myotonia Congenita (Thomsen), Autosomal Dominant
Myotonia, Potassium-Aggravated
Neuroferritinopathy type 3
Neuropathy, Ataxia, And Retinitis Pigmentosa
Niemann-Pick Disease
Niemann-Pick Disease, Type A
Niemann-Pick Disease, Type B
Niemann-Pick Disease, Type C1
Niemann-Pick Disease, Type C2
Ornithine Aminotransferase Deficiency
Paramyotonia Congenita Of Von Eulenburg
Paraplegia Spastica Tipo 2
Parkinson - Dementia Syndrome
Parkinson Disease 1, Familial