





REGISTRIES



Ring 14 International
 Marco Crimi
ICD10 category Q93.2 - Q07.9
 Coverage: International



The European Huntington's Disease Network Registry
 Michael ORTH, Christian NERI
ICD-10 Category: G10
 Coverage: European
 Contact: michael.orth@uni-ulm.de and christian.neri@inserm.fr



Spanish Rare Diseases Registries Research Network - SpainRDR
 Manuel POSADA
ICD-10 Category: NA
 Coverage: Spain
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The NIH and ORDR Registries
 Yaffa RUBINSTEIN
ICD-10 Category: NA
 Coverage: USA
 Contact: Yaffa.Rubinstein@nih.gov



RDCRN Registry for Nephrotic Syndrome or Glomerular Disease
 Matthias KRETZLER
ICD-10 Category: N04
 Coverage: USA
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European Registry and Network for Intoxication Type Metabolic Diseases – E-IMD
 Stefan KÖLKER
ICD-10 Category: E71.1, E72.2, E72.3, E72.4
 Coverage: International
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Australian National Spinal Muscular Atrophy Registry
 Caroline GRAHAM
ICD-10 Category: G12.0, G12.1
 Coverage: Australia
 Contact: Caroline.Graham@health.wa.gov.au



Australian National Duchenne Muscular Dystrophy Registry
 Caroline GRAHAM
ICD-10 Category: G71.0
 Coverage: Australia
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Global FKRP Registry
 Agata ROBERTSON, Volker STRAUB
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 Coverage: International
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The International Dysferlinopathy Registry

Gaëlle BLANDIN
ICD-10 Category: G71.0
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The European SCA (spinocerebellar ataxia) registry, EUROSCA-R

Thomas KLOCKGETHER
ICD-10 Category: G11
Coverage: European
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International Registry of Recurrent and Familial Hemolytic Uremic Syndrome (HUS) and Thrombocytopenic Purpura (TTP)

Marina NORIS, Erica DAINA
ICD-10 Category: D59.3, M31.1
Coverage: International
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European Cystic Fibrosis Society - Patient Registry

Jacqui VAN RENS
ICD-10 Category: E84
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Italian Cystic Fibrosis Registry

Domenica TARUSCIO, Maria Antonietta STAZI
ICD-10 Category: E84
Coverage: Italy
Contact: domenica.taruscio@iss.it marco.salvatore@iss.it



LeukoFrance Database and Biobank

Odile BOESPFLUG-TANGUY
ICD-10 Category: E75
Coverage: France
Contact: odile.boespflug-tanguy@inserm.fr



Paroxysmal Nocturnal Hemoglobinuria Registry

Domenica TARUSCIO, Paola TORRERI
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BIOBANKS



Biobanking and Biomolecular Resources Research Infrastructure – BBMRI

Roxana MERINO MARTINEZ
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EuroBiobank

Marina MORA
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Telethon Network of Genetic Biobanks

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Covered diseases: D50-D89: Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism; E00-E90: Endocrine, nutritional and metabolic diseases; G00-G99: Diseases of the nervous system; M00-M99: Diseases of the musculoskeletal system and connective tissue; N00-N99: Diseases of the genitourinary system.

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