CIBERER'S DECENTRALIZED COLLECTIONS:

Node	RD	Number of samples/patients	Sample type
1	Lowe syndrome (LS); Congenital disorder of glycosilation (GDG).	LS: 7 CDG: 20	Primary fibroblasts, DNA, serum.
2	Rare ocular diseases (Norrie, retinal dystrophies, retinosis pigmentaria, Angelman S., alpha-1-antitrypsin, among others); rare cancers; inherited metabolic diseases; neuromuscular and peripheral neuropathies (CMT, Steinert, Duchenne/Becker, Kennedy, among others); neurodegenerative (ataxia, Huntington, Lafora); bone dysplasias (achondroplasia, Holt-Oram); dermatological (Epidermolysis bullosa, incontinentia pigmenti) and others.	Over 40,000 samples	DNA
3	Hereditary hemorrhagic telangiectasia; von Hippel Lindau syndrome (VHL)	HHT: 638 VHL: 140	**HHT:** DNA, plasma; BOECs (blood outgrowth endothelial cells) and HUVECs (Human umbilical vein endothelial cells) primary cultures. **VHL:** DNA, plasma samples, primary cultures.

Node	RD	Number of samples/patients	Sample type
4	Rare diseases and familiar cancer	25,000 samples of familial cancer and genetic diseases.	DNA
5	a): Leber Hereditary Optic Neuropathy, MERRF syndrome, mitochondrial sensorineural deafness, mitochondrial-diabetes; b): Leigh syndrome, severe mitochondrial encephalopathy, autosomal dominant optic atrophy plus syndrome, defects of intergenomic communication with multiple deletions of mtDNA.		Transmitochondrial cybrids (RDs within group **a)**; iPS Cells (RDs within group **b)**
6	Homocystinuria, Krabbe disease, hereditary multiple exostoses, Costello syndrome, Gaucher disease, GM1 gangliosidosis, Sanfilippo disease type C, Niemann-Pick disease types A/B , Niemann-Pick disease type C.	50-200 samples of each pathology except for Opitz C Syndrome (10 samples)	DNA, fibroblasts.
7	Muscular fiber type 2 atrophy, dermatomyositis, muscle distrophy, oculopharyngeal muscular dystrophy, myoadenylate deaminase deficiency, Mc Ardle disease, congenital myopathy, critical illness myopathy, distal myopathy, neutral lipid storage disease, myofibrillar myopathy, mitochondrial myopathy, autoimmune necrotizing myopathy, neuropathic pattern, polymyositis, sporadic inclusion body myositis, muscle toxicity (Sunitinib), vasculitis.	1-50 patients/RD	PBMC, plasma, serum, DNA, tissue.
8	Mitochondrial diseases	2,000 samples aprox.	DNA

Node	RD	Number of samples/patients	Sample type
9	Fragile X and ID		DNA, RNA, tissue
10	Prostatic cancer, Wolfram syndrome and renal diseases	>200 patients/total	DNA, RNA from lymphocytes and tissue
11	Gaucher disease; Fabry disease; Nieman-Pick disease A/B; Nieman-Pick disease C; Lysosomal acid lipase deficiency.	1-300 patients/RD	DNA, plasma, leukocytes, fibroblasts, monocyte-derived macrophages.
12	Rare genetic diseases, including an important collection of overgrowth disorders	35,800 samples of patients with rare genetic diseases. Includes a collection of 1,800 patients with overgrowth disorders (From the Spanish Overgrowth Syndrome Registry).	DNA
13	Hereditary angioedema due to C1-inhibitor deficiency; Acquired angioedema; Angioedema with normal C1 inhibitor activity; Atypical hemolytic-uremic syndrome; C3 glomerulopathy; Barraquer-Simons syndrome; Lawence syndrome; several immunodeficiencies: C1q, C2, C3, C5, C6, C7, C8 , factor H, factor I.	1-600 patients/RD	ADN, ARN, serum and plasma

Node	RD	Number of samples/patients	Sample type
14	Hypoacusis, retinal dystrophies, Usher syndrome, Huntington disease, Charcot-Marie-Tooth disease, Duchenne's muscle distrophy, Steinert myotonic dystrophy, adult onset cerebellar ataxia, Friedreich ataxia, achondroplasia, albinism, spinal muscular atrophy	>500 samples	Tissue, cells and cell lines.
15	Bone disorders (Osteogenesis imperfecta, Ellis-van Creveld syndrom and bone dysplasias)	500 samples aprox.	DNA and fibroblast cell lines
16	Neuromuscular diseases	>1,500 samples	Tissue (muscle, nerve and skin), DNA from blood.
17	Hematological and Immunological diseases	20	DNA
18	Hemolytic anemia due to glucophosphate isomerase deficiency; Hemolytic anemia due to red cell pyruvate kinase deficiency; Triose phosphate-isomerase deficiency; Hereditary elliptocytosis; Hereditary spherocytosis; and other membrane pathologies without definitive diagnosis.	1-50 patients/RD	Leukocyte pellets