POSTER	POSTER SESSION	POSTER TITLE	Poster_Disease	Poster_Disease Group	Poster_MACRO-category
1	3	P2X7 RECEPTOR ANTAGONIST REDUCES FIBROSIS AND INFLAMMATION IN A MOUSE MODEL OF ALPHA-SARCOGLYCAN MUSCULAR DYSTROPHY	Alpha-sarcoglycanopathy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
2	1	3D MODELLING OF RARE MUSCULAR DISEASES, A POWERFUL PLATFORM FOR BASIC STUDIES AND DRUG VALIDATION	Alpha-sarcoglycanopathy; Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
3	1	NATURAL HISTORY OF BECKER MUSCULAR DYSTROPHY: TOWARD TRIAL READINESS	Becker muscular dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
4	1	IDENTIFICATION OF NEW BIOMARKERS MONITORING DMD PATHOLOGY AND RESPONSE TO TREATMENT	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
5	1	Duchenne Muscular Dystrophy: phenotype-genotype correlations	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
6	1	CHARACTERIZING PHENOTHYPES IN NON AMBULANT DUCHENNE MUSCULAR DYSTROPHY	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
7	1	CHARACTERIZATION OF THE PHENOTYPIC DIVERSITY IN DUPEX2 DUCHENNE MUSCULAR DYSTROPHY AND IDENTIFICATION OF PREDICTIVE/PROGNOSTIC MARKERS	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
8	1	INHIBITION OF COMPLEMENT C1 AMELIORATES THE DYSTROPHIC MUSCLE PHENOTYPE OBSERVED IN A MOUSE MODEL OF DUCHENNE MUSCULAR DYSTROPHY	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
9	1	INSTRUCTING ER-PHAGY TO COUNTERACT MUSCLE DISEASES	Duchenne Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
10	1	A MITOCHONDRIAL THERAPY FOR MUSCULAR DYSTROPHIES	Duchenne Muscular Dystrophy; Ullrich Congenital Muscular Dystrophy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
11	1	AT THE ORIGIN OF CONGENITAL MUSCULAR DYSTROPHY: SHEDDING LIGHT ON THE TDARK PROTEINS DPM2 AND DPM3	Muscular Dystrophies, congenital	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
12	3	SPERMIDINE AS NEW CANDIDATE FOR THE TREATMENT OF COL6 MYOPATHIES	Ullrich Congenital Muscular Dystrophy; Bethlem Myopathy	Genetic muscular disease\Muscular dystrophies	Neuromuscular disorders
13	3	CHROMATIN DYSFUNCTION IN EMERY DREIFUSS MUSCULAR DYSTROPHY	Emery-Dreifuss muscular dystrophy	Genetic muscular disease\Myopathies and cardiomyopathies	Neuromuscular disorders

14	1	TOWARDS PRECISION MEDICINE WITH HUMAN INDUCED PLURIPOTENT STEM CELLS FOR DYSTROPHIN ASSOCIATED CARDIOMYOPATHY	Muscular Dystrophies, cardiomyopathy	Genetic muscular disease\Myopathies and cardiomyopathies	Neuromuscular disorders
15	3	NEW PHARMACOLOGICAL TREATMENT FOR TUBULAR AGGREGATE MYOPATHIES	Tubular Aggregate Myopathy	Genetic muscular disease\Myopathies and cardiomyopathies	Neuromuscular disorders
16	1	ROLE OF STORE-OPERATED CA2+ ENTRY (SOCE) IN TUBULAR AGGREGATE MYOPATHY.	Tubular Aggregate myopathy	Genetic muscular disease\Myopathies and cardiomyopathies	Neuromuscular disorders
17	1	GENE EDITING IN MYOTONIC DYSTROPHY TYPE 1: ASSESSMENT OF EFFICIENCY, SAFETY AND THERAPEUTIC EFFECT OF CTG-REPEAT DELETION IN A MOUSE MODEL OF DISEASE	Myotonic Dystrophy type 1	Genetic muscular disease\Myotonic disorders	Neuromuscular disorders
18	1	TRIAL READINESS AND ENDPOINT ASSESSMENT IN CONGENITAL AND CHILDHOOD MYOTONIC DYSTROPHY: OUTCOME MEASURES AND ENDPOINT ASSESSMENTS (GUP19002)	Myotonic Dystrophy type 1	Genetic muscular disease\Myotonic disorders	Neuromuscular disorders
19	3	ROLE OF POLYAMINES-EIF5A-AUTOPHAGY AXIS IN THE PATHOGENESIS OF MYOTONIC DYSTROPHY TYPE 2	Myotonic Dystrophy Type 2	Genetic muscular disease\Myotonic disorders	Neuromuscular disorders
20	2	MITOCHONDRIAL MYOPATHY ASSOCIATED TO FDX2 MUTATIONS: A CROSSROADS OF FES PROTEIN BIOGENESIS AND COENZYMEE Q BIOSINTHESYS	MEOAL	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
21	1	APPLICATION OF THE ESCHERICHIA COLI MODEL SYSTEM TO STUDY THE HUMAN POLYRIBONUCLEOTIDE PHOSPHORYLASE	Mitochondrial Diseases; Hereditary hearing loss; Leigh Syndrome	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
22	3	DEORPHANIZING AND FUNCTIONALIZING THE MITOCHONDRIAL PROTEIN TMEM65	Mitochondrial encephalomyopathy	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
23	1	NOVEL STRATEGIES TO BLOCK TOXICITY OF THE MUTANT ANDROGEN RECEPTOR IN SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA)	Spinal and Bulbar Muscular Atrophy	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
24	1	INVESTIGATION OF TRANSLATIONAL DEFECTS IN MULTIPLE MODELS OF SMA	Spinal Muscular Atrophy	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
25	1	SMN CIRCULAR RNAS AS POTENTIAL NEW TARGETS AND BIOMARKERS FOR THE THERAPEUTIC RESPONSE IN SPINAL MUSCULAR ATROPHY	Spinal Muscular Atrophy	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
26	1	PEROXISOMAL-MITOCHONDRIAL INTERACTION IMPINGING ON MUSCLE FUNCTION	Zellweger Spectrum Disorders	Genetic neurological disorder\Neuromuscular diseases	Neuromuscular disorders
27	1	FINDING NEW TARGETS TO COUNTERACT BRAIN PROGENITOR CELLS DYSREGULATION IN AGC1 DEFICIENCY HYPOMYELINATION: A MULTIDISCIPLINARY APPROACH.	AGC1 deficiency	Genetic neurological disorder	Neurological disorders

28	1	DISSECTING INNATE IMMUNITY AND NUCLEIC ACID SENSING IN GENE THERAPY AND DISEASE	Aicardi-Goutières Syndrome	Genetic neurological disorder	Neurological disorders
29	2	EXPERIMENTAL GENE THERAPY IN MITOCHONDRIAL DISORDERS	Complex I deficiency	Genetic neurological disorder	Neurological disorders
30	2	PATHOLOGICAL MOLECULAR MECHANISMS UNDERLYING APOPT1/COA8 LOSS OF FUNCTION	Complex IV deficiency, nuclear type	Genetic neurological disorder	Neurological disorders
31	2	THE LNCRNA PHOX2B-AS1 IN THE PATHOGENESIS AND AS POTENTIAL DRUG TARGET IN CONGENITAL CENTRAL HYPOVENTILATION SYNDROME (CCHS)	Congenital central hypoventilation syndrome	Genetic neurological disorder	Neurological disorders
32	3	EVALUATION OF IN VITRO NEURONAL CULTURES AS TESTING MODEL FOR PHARMACOLOGICAL TREATMENTS OF A GENETIC FORM OF MIGRAINE	Familial Hemiplegic Migraine type 3	Genetic neurological disorder	Neurological disorders
33	3	CORRECTING FOXG1 ACTIVITY LEVELS BY SMALL RNA-ANALOGS MODULATING THE CORRESPONDING MRNA LEVELS AND THEIR TRANSLATION RATES	FOXG1 syndrome	Genetic neurological disorder	Neurological disorders
34	2	GLUT1 DEFICIENCY: NEW THERAPEUTIC STRATEGIES TO INCREASE GLUCOSE TRANSPORT ACROSS THE BLOOD BRAIN BARRIER (BBB)	GLUT1 Deficiency Syndrome	Genetic neurological disorder	Neurological disorders
35	2	LEADING GLUT1 TOWARDS THE PLASMA MEMBRANE	GLUT1 Deficiency Syndrome	Genetic neurological disorder	Neurological disorders
36	2	THERAPEUTIC EFFICACY OF MIR-181A/B DOWN REGULATION IN LEIGH SYNDROME	Leigh Syndrome	Genetic neurological disorder	Neurological disorders
37	1	A REDOX CYCLER-BASED THERAPEUTIC STRATEGY AGAINST MITOCHONDRIAL RESPIRATORY CHAIN DYSFUNCTION-LINKED DISEASES	Mitochondrial Diseases	Genetic neurological disorder	Neurological disorders
38	2	MITMED: IDENTIFICATION AND CHARACTERIZATION OF NEW DISEASE GENES FOR MITOCHONDRIAL DISORDERS	Mitochondrial Diseases	Genetic neurological disorder	Neurological disorders
39	2	AAV-MEDIATED INHIBITION OF MIR-181A/B AS GENE-INDEPENDENT THERAPEUTIC TOOL FOR MITOCHONDRIAL DISEASES	Mitochondrial Diseases	Genetic neurological disorder	Neurological disorders
40	1	IDENTIFICATION OF DRUGS TARGETING POLG DISORDERS BY YEAST/ZEBRAFISH PRE-SCREENING	POLG mitochondrial disorders	Genetic neurological disorder	Neurological disorders
41	1	REGULATION OF ALTERNATIVE SPLICING OF CA2+ CHANNELS BY CRISPR/CAS9-MEDIATED GENOME EDITING AS AN ALL-PURPOSE GENETIC THERAPY FOR LOSS-OF-FUNCTION CACNA1A MUTATIONS	Episodic ataxia type 2	Genetic neurological disorder\Ataxias	Neurological disorders

42	3	CELL-PENETRATING SIL1 PROTEIN REPLACEMENT THERAPY FOR MARINESCO-SJOGREN SYNDROME	Marinesco-Sjogren syndrome	Genetic neurological disorder\Ataxias	Neurological disorders
43	3	PRECLINICAL EFFICACY STUDY OF PERK SIGNALING INHIBITORS AND TUDCA IN MARINESCO-SJÖGREN SYNDROME	Marinesco-Sjögren syndrome	Genetic neurological disorder\Ataxias	Neurological disorders
44	1	AN UNEXPECTED ROLE OF THE NIJMEGEN BREAKAGE SYNDROME PROTEIN (NBS1) AT THE PRIMARY CILIUM AND IN HEDGEHOG SIGNALING IS IMPORTANT FOR CEREBELLAR DEVELOPMENT AND MEDULLOBLASTOMA	Nijmegen Breakage Syndrome	Genetic neurological disorder\Ataxias	Neurological disorders
45	2	THE ALTERATION OF MITOCHONDRIAL ENERGETIC METABOLISM CONTRIBUTES TO THE PATHOGENESIS OF POSTERIOR COLUMN ATAXIA AND RETINITIS PIGMENTOSA	Posterior Column Ataxia and Retinitis Pigmentosa	Genetic neurological disorder\Ataxias	Neurological disorders
46	2	CORTICOSPINAL TRACT MICROSTRUCTURAL INTEGRITY AND ITS CORRELATION WITH CLINICAL AND MOLECULAR BIOMARKERS: A PROFILOMETRY MRI STUDY TO IDENTIFY IN-VIVO BIOMARKERS OF DISEASE SEVERITY IN ARSACS	Spastic Ataxia of Charlevoix-Saguenay (ARSACS)	Genetic neurological disorder\Ataxias	Neurological disorders
47	2	DANIO RERIO AS A MODEL TO REVEAL NEW INSIGHT OF RETINAL DEFECTS IN ARSACS	Spastic Ataxia of Charlevoix-Saguenay (ARSACS)	Genetic neurological disorder\Ataxias	Neurological disorders
48	2	DEVELOPMENT OF AN ALLELE-SPECIFIC EPIGENETIC SILENCING PLATFORM FOR THE TREATMENT OF SCA2	Spinocerebellar ataxia type 2	Genetic neurological disorder\Ataxias	Neurological disorders
49	2	PPAR GAMMA AGONIST PIOGLITAZONE RESTORES MITOCHONDRIAL QUALITY CONTROL IN FIBROBLASTS OF PITRM1 DEFICIENT PATIENTS	Spinocerebellar ataxia, autosomal recessive	Genetic neurological disorder\Ataxias	Neurological disorders
50	2	EXPLAINABLE ARTIFICIAL INTELLIGENCE AND FRACTAL DIMENSION OF BRAIN MRI IN FRIEDREICH ATAXIA AND SCAS	Spinocerebellar ataxia; Friedreich ataxia	Genetic neurological disorder\Ataxias	Neurological disorders
51	1	ANALYSIS OF INSYN1 FUNCTIONING IN THE REGULATION OF INHIBITORY NEURONAL TRANSMISSION IN A MOUSE MODEL OF CDKL5 DEFICIENCY DISORDER	CDKL5 deficiency disorder	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
52	2	IN VIVO CROSS-CORRECTION ENHANCES THE EFFICACY OF GENE THERAPY IN A MOUSE MODEL OF CDKL5 DEFICIENCY DISORDER	CDKL5 deficiency disorder	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
53	1	UNVEILING THE FUNCTIONAL ROLE OF CDKL5 AT THE INHIBITORY SYNAPSE THROUGH ITS INTERACTION WITH THE CYTOPLASMATIC COLLYBISTIN-GEPHYRIN COMPLEX	CDKL5 deficiency disorder	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
54	2	CHARACTERIZATION OF THE GUT MICROBIOTA IN CDKL5 DEFICIENCY DISORDER TO REVEAL NOVEL BIOMARKERS AND THERAPEUTIC STRATEGIES	CDKL5 deficiency disorder	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
55	2	INTEGRATED COMPUTATIONAL AND EXPERIMENTAL APPROACHES TO DRUG REPOSITIONING FOR RARE GENETIC DISORDERS	CDKL5 deficiency disorder	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders

56	**2**	PCDH19-RELATED NEURODEVELOPMENTAL SYNDROME: UNRAVELING THE PLAYERS OF NEURONAL HYPEREXCITABILITY IN SEARCH OF NEW THERAPEUTIC TARGETS	Developmental and Epileptic Encephalopathy 9	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
57	**2**	TEMPORAL MANIPULATION OF SCN1A GENE EXPRESSION IN DRAVET SYNDROME	Dravet syndrome	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
58	**1**	NANOBODY-MEDIATED MODULATION OF HCN1 CHANNELS IN EPILEPTIC DISORDERS	Early infantile epileptic encephalopathy, undetermined	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
59	**2**	NOVEL INSIGHTS ON CHLORIDE REGULATIONS: IMPLICATION FOR DISEASE ETIOLOGY AND TREATMENT	Epilepsy, Cognitive Deficits	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
60	**2**	RNA-BASED RESCUE OF INHIBITION AS POTENTIAL TREATMENT FOR GENETIC GABRA1-DEPENDENT EPILEPSY	Epilepsy, GABRA1-dependent	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
61	**3**	CURE MERRF: FROM FIBROBLASTS TO ORGANOIDS SPEEDING BASIC SCIENCE INTO CLINICAL TRIALS FOR MITOCHONDRIAL DISEASES	MERFF	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
62	**2**	INTERACTION OF PRRT2 WITH NA+ CHANNELS: PATHOGENETIC BASIS AND NEW TARGETS FOR THE CURE OF PRRT2-ASSOCIATED PAROXYSMAL DISORDERS	Paroxysmal kinesigenic dyskinesia	Genetic neurological disorder\Epilepsy and Seizures	Neurological disorders
63	**2**	PERINATAL OXYTOCIN AMELIORATES BEHAVIORAL AND IMMUNOLOGICAL TRAJECTORIES IN 22Q11.2 DELETION SYNDROME MICE CLOSING BRAIN BARRIERS	22q11.2 deletion syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
64	**2**	BOOSTING MITOCHONDRIAL BIOGENESIS DURING POSTNATAL DEVELOPMENT TO PREVENT COGNITIVE DEFICITS IN 22Q11 DELETION SYNDROME.	22q11.2 deletion syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
65	**1**	MECHANISMS OF SYNAPTIC DYSFUNCTION IN THE ANGELMAN SYNDROME	Angelman Syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
66	**1**	THE TETRASPANIN TSPAN5 REGULATES AMPARS EXOCYTOSIS BY INTERACTING WITH THE AP-4 COMPLEX	AP4 deficiency syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
67	**2**	MECHANISMS AND DISEASE MODELS OF NEURODEVELOPMENTAL DISORDERS INVOLVING CLC ANION TRANSPORTERS	CLCN3-7 related neurodevelopmental disorders	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
68	**1**	CREATINE DEFICIENCY SYNDROME: NOVEL INSIGHT INTO BRAIN FUNCTION AND THERAPEUTIC STRATEGIES	Creatine Transporter Deficiency	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
69	**2**	MODELING FMR1 EXPRESSION DYNAMIC DURING FIRST PHASES OF NEURODEVELOPMENT USING FXS IPSC-DERIVED 3D CORTICAL BRAIN ORGANOIDS	Fragile X Syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders

70	**2**	NEW THERAPEUTIC STRATEGIES FOR THE FRAGILE X SYNDROME	Fragile X Syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
71	**2**	RAC GTPASE IN INTELLECTUAL DISABILITY: PRECLINICAL OPPORTUNITIES FROM INTERFERING WITH A RAC1 SPECIFIC PROTEIN::PROTEIN INTERACTION	Intellectual Disability, non-syndromic X-linked	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
72	**2**	DETAILING AND MODELING DENDRITIC SPINE PRUNING PATHWAYS AND COGNITION IN RAB39B XLID MOUSE MODEL	Intellectual Disability, non-syndromic X-linked	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
73	**3**	EXPLORING THE EPIGENETIC REWIRING ASSOCIATED TO RLF MUTATIONS AS A DRIVER OF INTELLECTUAL DISABILITY	Intellectual disability-hypotonia-facial dysmorphism syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
74	**3**	DISSECTING THE PATHOMOLECULAR MECHANISMS OF PRR12 GENE INACTIVATION LEADING TO NEURODEVELOPMENTAL AND EYE ABNORMALITIES.	Neuroocular syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
75	**3**	TARGETING OLIGODENDROGLIAL CELL DYSFUNCTIONS TO TREAT COGNITIVE DEFECTS AND EPILEPSY IN PRIMARY AUTOSOMAL RECESSIVE MICROCEPHALY-17 (MCPH17) MODELS	Primary microcephaly 18, autosomal recessive	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
76	**2**	NEW VECTOR DESIGNING TO INCREASE EFFICACY AND SAFETY OF GENE-BASED THERAPIES FOR RETT SYNDROME	Rett syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
77	**2**	TARGETING RETT SYNDROME HYPEREXCITABILITY THROUGH ENHANCING GLUTAMATERGIC HOMEOSTASIS	Rett syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
78	**2**	THE INTERPLAY BETWEEN HPCAL4 AND MECP2: IDENTIFICATION AND CHARACTERIZATION OF A NOVEL PUTATIVE TARGET FOR RETT SYNDROME THERAPY	Rett syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
79	**2**	FUNCTIONAL STUDY ON A NEW PHARMACOLOGICAL APPROACH IN THE RETT SYNDROME	Rett syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
80	**2**	BASE AND PRIME EDITING OF DNA AS NEW PERSONALIZED TREATMENT FOR RETT DISEASE	Rett syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
81	**1**	IDENTIFICATION OF POSSIBLE THERAPEUTIC TARGETS TO RESCUE NEURONAL AND SYNAPTIC DYSFUNCTIONS CAUSED BY DELETIONS AND MUTATIONS OF THE TCF20 INTELLECTUAL DISABILITY GENE	TCF20-associated neurodevelopmental disorders	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
82	**3**	FUNCTIONAL DISSECTION OF PRC2-DEPENDENT DYSREGULATION IN WEAVER SYNDROME THROUGH CORTICAL BRAIN ORGANOIDS AND CRISPR/CAS9 GENOME EDITING SYSTEM	Weaver syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders
83	**2**	SINGLE-CELL MULTIOMIC DISSECTION OF ELECTROPHYSIOLOGICAL CORRELATES OF WILLIAMS-BEUREN- AND 7Q11.23 MICRODUPLICATION- SYNDROMES	Williams Beuren Syndrome	Genetic neurological disorder\Intellectual Disabilities	Neurological disorders

84	3	ROLE OF MYELOPEROXIDASE-MEDIATED NEUROINFLAMMATION IN ACERULOPLASMINEMIA	Aceruloplasminemia	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
85	1	TARGETING MITOCHONDRIAL METABOLISM TO PROMOTE NEURONAL MATURATION IN AHDS: DEVELOPING NEW THERAPEUTIC APPROACHES IN 3D MOUSE BRAIN MODELS	Allan-Herndon-Dudley Syndrome (MCT8 deficiency)	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
86	1	REPURPOSING CFTR CORRECTORS IN ALLAN HERNDON DUDLEY SYNDROME	Allan-Herndon-Dudley Syndrome (MCT8 deficiency)	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
87	3	MENINGES AS AN OVERLOOKED PHARMACOLOGICAL TARGET FOR GLOBOID CELL LEUKODYSTROPHY	Globoid Cell Leukodystrophy (Krabbe disease)	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
88	3	IN VITRO VALIDATION OF LENTIVIRAL VECTORS ENCODING FOR CHIMERIC MURINE AND HUMAN GALC ENZYMES TO IMPROVE THE EFFICACY OF GENE THERAPY APPROACHES FOR GLOBOID CELL LEUKODYSTROPHY	Globoid Cell Leukodystrophy (Krabbe disease)	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
89	2	ALTERATION OF LIPID METABOLISM IN THE PATHOGENESIS OF HEREDITARY SPASTIC PARAPLEGIA: UNRAVELLING THE MECHANISMS TO RECOVER CELL FUNCTION.	Hereditary Spastic Paraplegia	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
90	1	TARGETING SPASTIN PROTEIN DEGRADATION FOR HEREDITARY SPASTIC PARAPLEGIA (HSP) TREATMENT	Hereditary Spastic Paraplegia	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
91	2	REGULATING THE MITOCHONDRIAL PERMEABILITY TRANSITION PORE FOR TREATING HEREDITARY SPASTIC PARAPLEGIA TYPE 7 (SPG7)	Hereditary Spastic Paraplegia, tipo 7	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
92	2	DELVING INTO THE MECHANISMS UNDERLYING HPDL-RELATED DISORDERS WITH A MULTI-MODEL APPROACH	Hereditary Spastic Paraplegia, type 83	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
93	2	MODULATION OF PRE- AND POST-SYNAPTIC ADAM10 AND ITS CONTRIBUTION IN HUNTINGTON'S DISEASE CORTICO-STRIATAL DYSFUNCTION	Huntington's Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
94	1	METABOLISM OF POLYSIALIC ACID: NEW INSIGHT INTO PATHOLOGICAL MECHANISMS AND POTENTIAL TREATMENTS FOR HUNTINGTON'S DISEASE	Huntington's Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
95	1	A GENOME-WIDE SCREENING IN PLURIPOTENT CELLS IDENTIFIES MTF1 AS A NOVEL SUPPRESSOR OF MUTANT HUNTINGTIN TOXICITY	Huntington's Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
96	2	DEVELOPMENT OF AN EPIGENETIC EDITING STRATEGY FOR THE TREATMENT OF HUNTINGTON’S DISEASE.	Huntington's Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
97	1	NEUROPATHOLOGICAL FEATURES OF PARKIN R275W MOUSE MODEL	Juvenile Parkinsonism, autosomal recessive	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders

98	**2**	THE ROLE OF MICROGLIA IN LAFORA DISEASE: CHARACTERISATION OF MICROGLIAL SIGNATURES AND SCREENING OF ANTI-INFLAMMATORY MOLECULES IN A NOVEL ZEBRAFISH MODEL.	Lafora Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
99	**1**	DISSECTING THE MECHANISMS OF MYELOID-TO-NEURAL ENZYMATIC CROSS-CORRECTION IN THE CONTEXT OF HEMATOPOIETIC STEM CELL GENE THERAPY FOR METACHROMATIC LEUKODYSTROPHY	Metachromatic Leukodystrophy	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
100	**2**	DEVELOPMENTAL LACK OF TREM2 CAUSES DEFECTIVE SYNAPSE STRENGTHENING IN YOUNG ADULT MICE	Nasu-Hakola Disease	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
101	**2**	INSIGHT CLN5: APPROACHING THERAPIES IN THE NEURONAL CEROID LIPOFUSCINOSIS, USING ZEBRAFISH AS A TOOL	Neuronal ceroid lipofuscinosis 5	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
102	**3**	NPC INTRACELLULAR CHOLESTEROL TRANSPORTER 1 MEDIATES SARS-COV2 INFECTION	Niemann Pick type C1; COVID-19	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
103	**1**	IPS-DERIVED IRON-BURDEN ASTROCYTE AS MODELS TO APPROACH THE THERAPY FOR PKAN AND COPAN.	PKAN and CoPAN	Genetic neurological disorder\Neurodegenerative diseases	Neurological disorders
104	**2**	MECHANISMS OF AXONAL DEGENERATION IN LATE ONSET CMT1B NEUROPATHIES: MOLECULAR PATHWAYS AND THERAPEUTIC APPROACHES	Charcot-Marie-Tooth neuropathy	Genetic neurological disorder\Polyneuropathies	Neurological disorders
105	**1**	PHARMACOLOGICAL MODULATION OF MYELIN SYNTHESIS AND CYTOSKELETAL REMODELLING AS A THERAPEUTIC STRATEGY FOR CMT4B NEUROPATHIES WITH ABERRANT MYELIN	Charcot-Marie-Tooth neuropathy	Genetic neurological disorder\Polyneuropathies	Neurological disorders
106	**2**	BOOSTING HSPB3 TO PREVENT NEUROMUSCULAR DEGENERATION IN PERIPHERAL NEUROPATHIES	Charcot-Marie-Tooth type 2; Distal hereditary motor neuropathy type 2	Genetic neurological disorder\Polyneuropathies	Neurological disorders
107	**1**	KNOCKDOWN AND REPLACEMENT OF MFN2: A GENE THERAPY TO TREAT DOMINANTLY INHERITED PERIPHERAL NEUROPATHY CMT2A	Charcot-Marie-Tooth type 2A	Genetic neurological disorder\Polyneuropathies	Neurological disorders
108	**2**	PHARMACOLOGICAL DEGRADERS FOR THE CELLULAR PRION PROTEIN	Prion Diseases	Genetic neurological disorder\Prion diseases	Neurological disorders
109	**3**	PHENOTYPE OF THE FIRST MOUSE MODEL OF COLE CARPENTER SYNDROME.	Cole Carpenter Syndrome	Genetic bone disease	Other genetic disorders
110	**2**	“SEARCHING NEW MOLECULAR TARGETS IN FIBRODYSPLASIA OSSIFICANS PROGRESSIVA (FOP): IS THE AUTOPHAGY SIGNALLING A GOOD CANDIDATE?”.	Fibrodysplasia Ossificans Progressiva	Genetic bone disease	Other genetic disorders
111	**2**	HOW LACK OF TRIMERIC INTRACELLULAR CATION CHANNEL B AFFECTS BONE	Osteogenesis Imperfecta	Genetic bone disease	Other genetic disorders

112	**2**	CHARACTERIZING THE MOLECULAR FUNCTIONS OF TENT5/FAM46 PROTEINS	Osteogenesis imperfecta; Systemic Lupus Erythematosus; Charcot-Marie-Tooth disease	Genetic bone disease	Other genetic disorders
113	**3**	AUTOSOMAL DOMINANT OSTEOPETROSIS TYPE 2 (ADO2): CLOSE TO THE CURE. WHAT TO WE MISS?	Osteopetrosis, autosomal dominant type 2	Genetic bone disease	Other genetic disorders
114	**2**	EX VIVO EXPANSION OF HEMATOPOIETIC STEM AND PROGENITOR CELLS (HSPC) FOR GENE THERAPY	Osteopetrosis, malignant autosomal recessive	Genetic bone disease	Other genetic disorders
115	**3**	ELUCIDATING THE SIGNIFICANCE OF OSTEOPETROTIC BONE MARROW NICHE IN HEMATOPOIETIC STEM AND PROGENITOR CELLS, AND ITS IMPLICATIONS FOR STEM CELL THERAPY	Osteopetrosis, malignant autosomal recessive	Genetic bone disease	Other genetic disorders
116	**3**	STRUCTURAL-FUNCTIONAL ANALYSIS OF CLC PROTEIN FAMILY	Osteopetrosis; Bartter syndrome	Genetic bone disease	Other genetic disorders
117	**2**	DEVELOPMENT OF SUBTYPE-SPECIFIC CARDIOMYOCYTE MODELS TO UNRAVEL DISTINCT CELLULAR MECHANISMS OF LMNA-CARDIOMYOPATHY	Cardiomyopathy Dilated 1A	Genetic cardiac disease	Other genetic disorders
118	**1**	STUDY OF THE AMYLOIDOGENIC CONVERSION OF S52P AND V122I TRANSTHYRETIN VARIANTS BY NUCLEAR MAGNETIC RESONANCE: ELUCIDATION OF THE MOLECULAR MECHANISMS LEADING TO ATTR AMYLOIDOSIS.	Hereditary transthyretin amyloidosis	Genetic cardiac disease	Other genetic disorders
119	**2**	HERG POTASSIUM CHANNEL ENHANCERS AS A NOVEL THERAPEUTIC APPROACH FOR LONG QT SYNDROME	Long QT Syndrome	Genetic cardiac disease	Other genetic disorders
120	**3**	MODULATION OF NBAS-RELATED FUNCTIONS IN THE EARLY RESPONSE TO SARS-COV-2 INFECTION	Acrofrontofacionasal dysostosis; COVID-19	Genetic developmental defect during embryogenesis	Other genetic disorders
121	**3**	ZEBRAFISH AS MODEL FOR CATEL-MANZKE SYNDROME: IDENTIFICATION AND CHARACTERIZATION OF DANIO RERIO TGDS	Catel-Manzke Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
122	**3**	CONNECTING CRANIOFACIAL MALFORMATIONS WITH NEURAL CREST SPLICING DEFECTS BY UNCOVERING THE HIDDEN ROLE OF NATURAL KILLER CELL TRIGGERING RECEPTOR GENE (NKTR).	Developmental delay with craniofacial and genital anomalies	Genetic developmental defect during embryogenesis	Other genetic disorders
123	**2**	JOUBERT SYNDROME: BEYOND CONVENTIONAL MENDELIAN GENETICS	Joubert syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
124	**1**	A NOVEL NEURODEVELOPMENT SYNDROME CAUSED BY RECESSIVE VARIANTS IN THE FSD1L GENE	Neurodevelopmental disorders	Genetic developmental defect during embryogenesis	Other genetic disorders
125	**1**	CRISPR-CAS9-BASED FUNCTIONAL INVESTIGATION OF THE “DARK GENOME” IN SEARCH OF PUTATIVE DOWNSTREAM EFFECTORS OF SOX2 IN NEURODEVELOPMENTAL DISEASE	Neurodevelopmental disorders	Genetic developmental defect during embryogenesis	Other genetic disorders

126	**2**	GENERATION OF PATIENT-DERIVED IPSCS FOR UNDERSTANDING THE PATHOGENIC MECHANISMS UNDERLYING ALTERED NEURONAL FUNCTION ASSOCIATED WITH CAMK2B GENE MUTATIONS	Neurodevelopmental syndrome due to CAMK2b mutation	Genetic developmental defect during embryogenesis	Other genetic disorders
127	**2**	ALTERED CORTICAL SENSORY PROCESSING AND FUNCTIONAL CONNECTIVITY IN SHANK3B+/- MICE	Phelan-McDermid Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
128	**2**	MODELING PITT-HOPKINS SYNDROME AND NEW PATHOGENETIC VARIANTS OF TCF4 BY GENE EDITING: A STEP FORWARD TOWARD PRECISION MEDICINE (HOPEFOR)	Pitt-Hopkins Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
129	**2**	THE ROLE OF ANCIENT GENE VARIANTS IN PRADER-WILLI SYNDROME PATHOPHYSIOLOGY	Prader-Willi Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
130	**1**	INVESTIGATING THE RELATIONSHIP BETWEEN TRANSCRIPTIONAL AND REPRESSIVE CONDENSATES IN A STEM CELL-BASED KABUKI SYNDROME MODEL	Kabuki Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
131	**1**	ROLE OF CHROMATIN CONDENSATES IN TUNING NUCLEAR MECHANO-SENSING IN KABUKI SYNDROME	Kabuki Syndrome	Genetic developmental defect during embryogenesis	Other genetic disorders
132	**1**	XQ26.3 DUPLICATIONS IN X-LINKED ACROGIGANTISM DISRUPT A TOPOLOGICALLY ASSOCIATING DOMAIN (TAD) AND REWIRE GPR101-ENHANCER INTERACTIONS	X-linked acrogigantism	Genetic endocrine disease	Other genetic disorders
133	**2**	MIR-181A/B DOWNREGULATION: A MUTATION-INDEPENDENT THERAPEUTIC APPROACH FOR INHERITED RETINAL DISEASES	Inherited Retinal Diseases	Genetic eye disease	Other genetic disorders
134	**2**	IDENTIFICATION OF DRUGS FOR AUTOSOMAL DOMINANT OPTIC ATROPHY (ADOA): FROM ADOA RCGS MODELS TO MICROPARTICLE-BASED DRUG DELIVERY IN AN ADOA MOUSE MODEL	Optic Atrophy, autosomal dominant	Genetic eye disease	Other genetic disorders
135	**2**	PIGMENT EPITHELIUM-DERIVED FACTOR (PEDF) AND DERIVED PEPTIDES AS THERAPEUTIC AGENTS FOR INHERITED RETINAL DEGENERATION	Retinitis Pigmentosa	Genetic eye disease	Other genetic disorders
136	**2**	MODULATING AUTOPHAGY: A NOVEL GENE-INDEPENDENT THERAPEUTIC TREATMENT FOR ADRP	Retinitis Pigmentosa, autosomal dominant	Genetic eye disease	Other genetic disorders
137	**2**	THERAPEUTIC HOMOLOGY-INDEPENDENT TARGETED INTEGRATION IN RETINA AND LIVER	Retinitis Pigmentosa; Mucopolysaccharidosis type VI	Genetic eye disease	Other genetic disorders
138	**1**	UBIAD1 AND FERROPTOSIS: EXPLORING A CURE FOR SCHNYDER CORNEAL DYSTROPHY (SCD)	Schnyder corneal dystrophy	Genetic eye disease	Other genetic disorders
139	**2**	MUTATION-INDEPENDENT GENOME EDITING APPROACHES FOR TREATMENT OF STARGARDT DISEASE	Stargardt disease	Genetic eye disease	Other genetic disorders

140	**2**	AN IN VIVO MODEL OF INTRACTABLE R257C-ACTG2 VISCERAL MYOPATHY TO STUDY PATHOGENESIS AND TO IDENTIFY NEW DISEASE TARGETS	Chronic intestinal pseudoobstruction; Familial visceral myopathy	Genetic gastroenterological disease	Other genetic disorders
141	**2**	THE BIOMOLECULAR CASCADE UNDERGOING CELL CONTRACTION IN PRESENCE OF VSCM CAUSATIVE MUTANTS	Chronic intestinal pseudoobstruction; Familial visceral myopathy	Genetic gastroenterological disease	Other genetic disorders
142	**2**	CELLULAR AND PROTEOMIC APPROACHES TO STUDY THE ROLE OF ACTG2 MUTATION-MEDIATED MISFOLDING AND PROTEIN AGGREGATION AS DRUGGABLE TARGETS IN VISCERAL MYOPATHY	Chronic intestinal pseudoobstruction; Familial visceral myopathy	Genetic gastroenterological disease	Other genetic disorders
143	**1**	AL AMYLOIDOSIS: GENE RESTRICTION REVEALS THE HIDDEN MOLECULAR BASIS OF AMYLOID TRANSFORMATION OF IMMUNOGLOBULIN LIGHT CHAINS	AL amyloidosis	Genetic hematologic disease	Other genetic disorders
144	**2**	THE HUMAN DELTA-GLOBIN GENE AS A THERAPEUTIC TOOL FOR BETA-HEMOGLOBINOPATHIES. POST GWAS TARGET VALIDATION AND EVALUATION OF MOLECULES IN PRECLINICAL MODELS.	Beta-thalassemia; Sickle Cell Disease	Genetic hematologic disease	Other genetic disorders
145	**2**	CELL-BASED THERAPY FOR CONGENITAL THROMBOTIC THROMBOCYTOPENIC PURPURA	Congenital thrombotic thrombocytopenic purpura	Genetic hematologic disease	Other genetic disorders
146	**1**	ADENOSINE DEAMINASE 2 DEFICIENCY: FROM THE UNDERLYING DISEASE MECHANISMS TO GENE THERAPY	Deficiency of adenosine deaminase 2	Genetic hematologic disease	Other genetic disorders
147	**2**	CHARACTERIZATION OF ENDOTHELIAL FUNCTION AND ANGIOGENESIS IN GLANZMANN THROMBASTHENIA: POSSIBLE ROLE IN GASTROINTESTINAL ANGIODYSPLASIA	Glanzmann Thrombasthenia	Genetic hematologic disease	Other genetic disorders
148	**3**	ANALYSIS OF THE PGE2-MEF2A AXIS IN THE BONE MARROW MICROENVIRONMENT	Hematologic genetic diseases	Genetic hematologic disease	Other genetic disorders
149	**1**	FVIII REGULATES ENDOTHELIAL CELL FUNCTIONALITY	Hemophilia A	Genetic hematologic disease	Other genetic disorders
150	**2**	LONG TERM EFFECTIVENESS OF REPLACEMENT THERAPIES IN HEMOPHILIA: A MATTER OF SPECIFIC DENDRITIC CELL SUBSETS?	Hemophilia A	Genetic hematologic disease	Other genetic disorders
151	**3**	EXPLORING THE ROLE OF MEDIATOR COMPLEX SUBUNIT 12-LIKE (MED12L) IN RARE MYELOID NEOPLASMS	Myeloproliferative neoplasm	Genetic hematologic disease	Other genetic disorders
152	**3**	THE FC RECEPTOR CD32 IS A SPECIFIC CELL-SURFACE MARKER FOR ISOLATING HUMAN HEMOGENIC ENDOTHELIAL CELLS	SCID-X1	Genetic hematologic disease	Other genetic disorders
153	**1**	RIBOSOMAL PATHOLOGIES: MECHANISTIC THERAPY OF SHWACHMAN- DIAMOND SYNDROME AND PREVENTION OF MALIGNANT COMPLICATIONS DUE TO STEM CELL MANIPULATION	Shwachman-Diamond syndrome	Genetic hematologic disease	Other genetic disorders

154	**2**	IDENTIFICATION OF DRUGGABLE PRO-RESOLVING MECHANISMS IN SICKLE CELL DISEASE	Sickle Cell Disease	Genetic hematologic disease	Other genetic disorders
155	**3**	IMPACT OF A NOVEL VARIANT OF THE BETA ISOFORM OF THE TBXA2R GENE ASSOCIATED WITH A HEMORRHAGIC DISORDER ON PLATELET AND ENDOTHELIAL FUNCTION	Thromboxane receptor defect	Genetic hematologic disease	Other genetic disorders
156	**2**	HUMAN HEMATOPOIETIC STEM/PROGENITOR CELL TRAFFICKING AND CLONAL TRACKING	Wiskott-Aldrich Syndrome; ADA-SCID; Metachromatic Leukodystrophy; Mucopoplysaccharidosis I (Hurler syndrome)	Genetic hematologic disease	Other genetic disorders
157	**2**	DECONVOLUTING THE DYNAMICS OF HEMATOPOIETIC RECONSTITUTION IN GENE THERAPY PATIENTS	Wiskott-Aldrich Syndrome; beta-thalassemia; Metachromatic Leukodystrophy	Genetic hematologic disease	Other genetic disorders
158	**1**	AGE OF ADMINISTRATION IMPACTS THE EFFICIENCY OF LENTIVIRAL VECTOR-MEDIATED HEPATOCYTE TRANSDUCTION IN VIVO AND ITS DISTRIBUTION IN THE LIVER LOBULE	Hemophilia B	Genetic hepatic disease	Other genetic disorders
159	**1**	MOLECULAR MECHANISMS COORDINATING MEMBRANE TRAFFICKING AND ION TRANSPORT IN WILSON DISEASE	Wilson disease	Genetic hepatic disease	Other genetic disorders
160	**2**	NUCLEASE-FREE TARGETED INTEGRATION OF A PROMOTERLESS MINI-ATP7B CONFERS PROLIFERATIVE ADVANTAGE TO EDITED HEPATOCYTES AND CORRECTS WILSON DISEASE	Wilson disease	Genetic hepatic disease	Other genetic disorders
161	**1**	MODELING WOLMAN DISEASE USING GENETICALLY ENGINEERED HUMAN LIVER ORGANOIDS	Wolman disease	Genetic hepatic disease	Other genetic disorders
162	**2**	CHARACTERIZATION OF HYPER-IGE CD4+ T LYMPHOCYTES AND THEIR RESPONSES TO OPPORTUNISTIC PATHOGENS.	Hyper-IgE syndrome	Genetic immune disease	Other genetic disorders
163	**1**	GENOME INTEGRITY ASSESSMENT OF EDITED CD4+ LYMPHOCYTES FOR THE TREATMENT OF HYPER-IGM 1	Hyper-IgM 1 syndrome	Genetic immune disease	Other genetic disorders
164	**1**	NUCLEAR STABILITY AND INNATE ACTIVATION IN WASP KO MYELOID CELLS	Wikott-Aldrich Syndrome	Genetic immune disease	Other genetic disorders
165	**3**	A NOVEL THERAPEUTIC STRATEGY FOR RARE GENETIC DISEASES CAUSED BY TELOMERE DYSFUNCTION	Dyskeratosis Congenita	Genetic immune disease	Other genetic disorders
166	**3**	CELLULAR SENESCENCE AND INFLAMMATORY PROGRAMS ARE UNINTENDED CONSEQUENCES OF CRISPR-CAS9 GENE EDITING IN HEMATOPOIETIC STEM AND PROGENITORS CELLS	Dyskeratosis Congenita	Genetic immune disease	Other genetic disorders
167	**2**	DECIPHERING DYSFUNCTIONAL METABOLIC PATHWAYS IN NEPHROPATIC CYSTINOSIS	Cystinosis	Genetic renal disease	Other genetic disorders

168	1	KIDNEY ORGANOIDS UNVEILED A NOVEL ROLE OF OCRL IN LIPID METABOLISM ASSOCIATED WITH THE PROGRESSIVE DECLINE OF KIDNEY FUNCTION IN LOWE SYNDROME	Lowe Syndrome	Genetic renal disease	Other genetic disorders
169	2	DISSECTING THE ROLE OF TFEB IN AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE	Polycystic kidney disease, autosomal dominant	Genetic renal disease	Other genetic disorders
170	2	ROLE OF QUALITY CONTROL IN THE EARLY SECRETORY COMPARTMENT IN AUTOSOMAL DOMINANT TUBULOINTERSTITIAL KIDNEY DISEASE	Tubulointerstitial Kidney Disease, Autosomal Dominant	Genetic renal disease	Other genetic disorders
171	1	UNRAVELLING THE PATHOGENIC MECHANISM OF CEP83 MUTATIONS IN NEPHRONOPHTHISIS	Retinitis Pigmentosa; Nephronophthisis	Genetic renal disease	Other genetic disorders
172	1	RESCUE OF MUTANT CFTR CHLORIDE CHANNELS BY A MIMETIC PEPTIDE TARGETING THE A-KINASE ANCHORING FUNCTION OF PI3KΓ	Cystic fibrosis	Genetic respiratory disease	Other genetic disorders
173	3	PHARMACOLOGICAL MODULATION OF ION TRANSPORT TO TREAT THE BASIC DEFECT IN CYSTIC FIBROSIS	Cystic fibrosis	Genetic respiratory disease	Other genetic disorders
174	2	CRISPR/CAS-MEDIATED BASE EDITING: A PROMISING TOOL FOR DISEASE MODELING AND PERSONALIZED MEDICINE APPROACHES FOR PRIMARY CILIARY DYSKINESIA	Primary Ciliary Diskinesia	Genetic respiratory disease	Other genetic disorders
175	1	NOVEL THERAPEUTIC APPROACHES FOR AEC SYNDROME	AEC Syndrome	Genetic skin disease	Other genetic disorders
176	1	A FUNCTIONAL GENOMICS FRAMEWORK TO INVESTIGATE THE MOLECULAR BASES OF RARE GENETIC DISEASES	AEC Syndrome; EEC Syndrome	Genetic skin disease	Other genetic disorders
177	1	ALLELE-SPECIFIC CRISPR-ENGINEERED CPF1 GENOME EDITING TO TREAT OCULAR SURFACE DISORDER IN ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING (EEC) SYNDROME	EEC syndrome	Genetic skin disease	Other genetic disorders
178	1	ANTIBODY GENE TRANSFER TREATMENT IMPROVES EPIDERMAL PATHOLOGY IN A MOUSE MODEL OF KID SYNDROME	Keratitis-ichthyosis-deafness syndrome	Genetic skin disease	Other genetic disorders
179	2	VASCULAR EHLERS-DANLOS SYNDROME DERMAL FIBROBLASTS' TRANSCRIPTOME: PATHOMECHANISMS AND TARGETABLE MOLECULES	Vascular Ehlers-Danlos syndrome	Genetic skin disease	Other genetic disorders
180	1	MIR22HG EXPRESSION PROFILE IN DIFFERENT CELL POPULATIONS FROM OLIGOARTICULAR JUVENILE IDIOPATHIC ARTHRITIS PATIENTS	Oligoarticular Juvenile Idiopathic Arthritis	Genetic systemic or rheumatologic disease	Other genetic disorders
181	1	LINGLE-CELL TRANSCRIPTOMICS AND LINEAGE TRACING TO ENABLE PRECISION MEDICINE IN LYNCH-DERIVED COLORECTAL NEOPLASIAS	Lynch syndrom	Genetic tumor / neoplasia	Other genetic disorders

182	1	TREATMENT WITH THE CARDIOLIPIN-TARGETED PEPTIDE ELAMIPRETIDE IMPROVES CARDIAC MITOCHONDRIAL DYSFUNCTION IN A MURINE MODEL OF BARTH SYNDROME	Barth Syndrome	Inborn errors of metabolism	Other genetic disorders
183	2	DOWN REGULATION OF MANNOSE-6-PHOSPHATE RECEPTORS IN FABRY DISEASE CARDIOMYOPATHY. POTENTIAL TARGET FOR ENZYME THERAPY ENHANCEMENT	Fabry Disease	Inborn errors of metabolism	Other genetic disorders
184	1	LIVER-DIRECTED PROMOTERLESS GENE TARGETING WITHOUT THE USE OF NUCLEASES AS A POTENTIAL THERAPY FOR FABRY DISEASE	Fabry Disease	Inborn errors of metabolism	Other genetic disorders
185	2	CELL-BASED ASSAYS OF GLA GENETIC VARIANTS OF UNKNOWN SIGNIFICANCE	Fabry Disease	Inborn errors of metabolism	Other genetic disorders
186	1	EXPLOITING REGULATORY T-CELL METABOLIC REPROGRAMMING AND VASCULAR TROPISM AS THERAPEUTIC TOOLS FOR FAMILIAL HYPERCHOLESTEROLAEMIA	Familial Hypercholesterolemia	Inborn errors of metabolism	Other genetic disorders
187	2	DUAL TARGET APPROACH FOR THE TREATMENT OF GAUCHER DISEASE: NEW ANTIOXIDANT PH-SENSITIVE PHARMACOLOGICAL CHAPERONES	Gaucher disease	Inborn errors of metabolism	Other genetic disorders
188	1	FREE CYTOSOLIC-MITOCHONDRIAL DNA TRIGGERS A POTENT TYPE-I INTERFERON RESPONSE IN KEARNS–SAYRE PATIENTS COUNTERACTED BY MOFETIL MYCOPHENOLATE	Kearns-Sayre Syndrome	Inborn errors of metabolism	Other genetic disorders
189	2	EVALUATING THE EFFICACY OF A GENE EDITING STRATEGY FOR PROGRESSIVE FAMILIAR INTRAHEPATIC CHOLESTASIS TYPE 2 (PFIC-2)	Progressive familiar intrahepatic cholestasis	Inborn errors of metabolism	Other genetic disorders
190	1	MICE LACKING TRPML1 PRESENT KIDNEY DISEASE	Mucolipidosis type IV	Inborn errors of metabolism	Other genetic disorders
191	1	BONE DEFECTS AND CROSS-CORRECTION IN MPSIH HSPC-GT	Mucopolysaccharidosis type I (Hurler syndrome)	Inborn errors of metabolism	Other genetic disorders
192	1	AMYLOID AGGREGATION AND LYSOSOMAL MEMBRANE DYNAMICS IN SANFILIPPO DISEASE	Mucopolysaccharidosis type III	Inborn errors of metabolism	Other genetic disorders
193	2	PHARMACOLOGICAL STIMULATION OF AUTOPHAGY TO RESCUE PROTEINOPATHY AND COGNITIVE DECLINE IN MUCOPOLYSACCHARIDOSIS-IIIA	Mucopolysaccharidosis type III A	Inborn errors of metabolism	Other genetic disorders
194	2	EARLY DEVELOPMENT OF MPS-IIIA DOPAMINERGIC NEURONS: AT THE NEXUS OF BEHAVIOR CHANGES AND THERAPY	Mucopolysaccharidosis type III A	Inborn errors of metabolism	Other genetic disorders
195	2	GENERATION, SELECTION AND CHARACTERISATION OF A NOVEL MOUSE MODEL FOR MUCOPOLYSACCHARIDOSIS TYPE IVA	Mucopolysaccharidosis type IV A (Morquio syndrome)	Inborn errors of metabolism	Other genetic disorders

196	**2**	AN INNOVATIVE PLATFORM APPROACH FOR THE DEVELOPMENT OF EX-VIVO GENE THERAPIES FOR THE TREATMENT OF LYSOSOMAL STORAGE DISEASES WITH SKELETAL INVOLVEMENT	Mucopolysaccharidosis type IV; alpha-mannosidosis	Inborn errors of metabolism	Other genetic disorders
197	**1**	A PRO-INFLAMMATORY SIGNATURE IN PATIENTS WITH LYSOSOMAL STORAGE DISORDERS DOES NOT PREVENT THE INDUCTION OF OF TOLEROGENIC CELLS TO PREVENT UNWONTED IMMUNE RESPONSES IN ENZYME REPLACEMENT THERAPY.	Mucopolysaccharidosis; Pompe Disease	Inborn errors of metabolism	Other genetic disorders
198	**1**	INDUCTION OF AUTOPHAGY PATHWAY AS NEW THERAPEUTIC OPTION TO PREVENT THE SYSTEMIC PATHOLOGY IN MULTIPLE SULFATASE DEFICIENCY (MSD)	Multiple sulfatase deficiency	Inborn errors of metabolism	Other genetic disorders
199	**3**	MEMBRANE REARRANGEMENTS DURING SARS-COV-2 INFECTION	COVID-19	Rare genetic diseases in general	Other genetic disorders
200	**3**	MOLECULAR DETERMINANTS OF VIRAL PATHOGENESIS	COVID-19	Rare genetic diseases in general	Other genetic disorders
201	**2**	TELETHON NETWORK OF GENETIC BIOBANKS: A KEY SERVICE FOR DIAGNOSIS AND RESEARCH ON RARE DISEASES	Rare genetic diseases in general	Rare genetic diseases in general	Other genetic disorders
202	**3**	TIGEM SCIENTIFIC OFFICE	Rare genetic diseases in general	Rare genetic diseases in general	Other genetic disorders
203	**1**	TELETHON UNDIAGNOSED DISEASES PROGRAM: THE 2022 MUTATION UPDATE	Undiagnosed rare genetic diseasess	Undiagnosed diseases with proven genetic origin	Other genetic disorders