Curriculum Vitae

Personal information

Mireia Del Toro Riera

Work experience

1. Employer: Hospital Universitari Vall d'Hebron
   - Start date: 011999
   - End date:
   - Position: Pediatric Neurology Consultant
   - Activities:
   - Country: Spain
2. Employer: Hospital Universitari Vall d'Hebron
   - Start date: 012012
   - End date:
   - Position: Metabolic Unit Coordinator
   - Activities:
   - Country: Spain
3. Employer: Hospital Universitari Vall d'Hebron
   - Start date: 102020
   - End date:
   - Position: Rare Diseases Coordinator
   - Activities:
   - Country: Spain
4. Employer: Servei Català de la Salut
   - Start date: 022017
   - End date: 022020
   - Position: Member of Hospital Medication Advisory Comittee
   - Activities:
   - Country: Spain
5. Employer: Servei Català de la Salut
   - Start date: 022016
   - End date:
   - Position: Member of the Lysosomal treatment Advisory Comittee
   - Activities:
   - Country: Spain
6. Employer: Institut Catalá de la Salut
   - Start date: 022019
   - End date:
   - Position: Coordinator of Rare Diseases Pharmacy Comittee
   - Activities:
   - Country: Spain

Education and training

1. Subject: Universitat Autonoma de Barcelona
   - Start date: 091982
   - End date: 061989
   - Qualification: Medical Degree
   - Organisation:
   - Country: Spain
2. Subject: Hospital Universitari Vall d'Hebron
   - Start date: 091991
   - End date: 091994
   - Qualification: Pediatric Degree
   - Organisation:
   - Country: Spain

Additional information

Publications

- Valayannopoulos V, et al. Carglumic acid enhances rapid ammonia detoxification in classical organic acidurias with a favourable risk_benefit profile: a retrospective observational study. Orphanet J Rare Dis. 2016 Mar 31;11:32
- Pérez_López J, et al. Transition process from paediatric to adult care in patients with inborn errors of metabolism. Consensus statement]. Med Clin (Barc). 2016 Dec 2;147(11):506.e1_506.e7. doi:10.1016/j.medcli.2016.09.018. Spanish. PubMed PMID: 27816186.
- Couce ML, et al. Transition from paediatric care to adult care for patients with mucopolysaccharidosis. Rev Clin Esp. 2018; 218:17_21. doi: 10.1016/j.rce.2017.06.005
- Pérez_López J, et al. Efficacy of Idursulfase therapy in patients with Mucopolysaccharidosis type II who initiated enzyme replacement therapy in adult age. A systematic review of the literature. Mol Genet Metab. 2018 Apr 30. pii:S1096_7192(18)30128_8. doi: 10.1016/j.ymgme.2018.04.013
- Quintero J, et al. The role of liver transplantation in propionic acidemia. Liver Transpl. Dec;24(12):1736_1745 doi: 10.1002/lt.25344 • González_Gutiérrez_Solana et al. Diagnosis and follow_up of patients with Hunter syndrome in Spain: A Delphi consensus. Medicine (Baltimore). 2018 Jul;97(29):e11246. doi: 10.1097/MD.0000000000011246.

- Chakrapani A, et al. Effect of carglumic acid with or without ammonia scavengers on hyperammonaemia in acute decompensation episodes of organic acidurias. Orphanet J Rare Dis. 2018; 20:97. doi: 10.1186/s13023_018_0840_4.
- Pintos_Morell G, et al. Elosulfase alfa formucopolysaccharidosis type IVA: Real_world experience in 7 patients from theSpanish Morquio_A early access program. Mol Genet Metab Rep. 2018 5;15:116_120. doi: 10.1016/j.ymgmr.2018.03.009
- Wijburg FA, et al. Intrathecalheparan_N_sulfatase in patients with Sanfilippo syndrome type A: A phase IIb randomized trial. Mol Genet Metab. 2019;126):121_130. doi:10.1016/j.ymgme.2018.10.006
- Baide_Mairena H, et al. Mutations in the mitochondrial complex I assembly factor NDUFAF6 cause isolated bilateral striatal necrosis and progressive dystonia in childhood.Mol Genet Metab. 2019; 126:250_258. doi: 10.1016/j
- Kurolap A, et al. Gaucher disease type 3c: New patients with unique presentations and review of the literature. Mol Genet Metab. 2019 Jun;127(2):138_146. doi: 10.1016/j.ymgme.2019.05.011.
- Andrade F, et al. Quantification of urinary derivatives of Phenylbutyric and Benzoic acids by LC_MS/MS as treatment compliance biomarkers in Urea Cycle disorders. J Pharm Biomed Anal. 2019 Aug1;176:112798. doi: 10.1016/j.jpba.2019.112798
- Pajares S, et al. An incidental finding in newborn screening leading to the diagnosis of a patient with ECHS1 mutations. Mol Genet Metab Rep. 2020 Jan 2;22:100553. doi: 10.1016/j.ymgmr.2019.100553. PMID: 31908952; PMCID: PMC6940607.
- Lampe C, et al. Transition of patients with mucopolysaccharidosis from paediatric to adult care. Mol Genet Metab Rep. 2019 Oct 21;21:100508. doi: 10.1016/j.ymgmr.2019.100508. PMID: 31687335; PMCID: PMC6819742.
- Marti_Sanchez L, et al. Delineating the neurological phenotype in children with defects in the ECHS1 or HIBCH gene. J Inherit Metab Dis. 2020 Jul 17. doi: 10.1002/jimd.12288. Epub ahead of print. PMID: 32677093.
- Reichert SC, et al. HNRNPH1_related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clin Genet. 2020 Jul;98(1):91_98. doi: 10.1111/cge.13765. Epub 2020 May 15. PMID: 32335897
- Mengel E, et al. Clinical disease progression and biomarkers in Niemann_Pick disease type C: a prospective cohort study. Orphanet J Rare Dis. 2020 Nov 23;15(1):328. doi: 10.1186/s13023_020_01616_0. PMID: 33228797; PMCID: PMC7684888
- Sala_Coromina J, et al. Leigh syndrome associated with TRMU gene mutations. Mol Genet Metab Rep. 2020 Dec 15;26:100690. doi: 10.1016/j.ymgmr.2020.100690
- Mengel E, et al. Clinical disease progression and biomarkers in Niemann_Pick disease type C: a prospective cohort study. Orphanet J Rare Dis. 2020 Nov 23;15(1):328. doi: 10.1186/s13023_020_01616_0
- Stepien KM, et al. Challenges in Transition From Childhood to Adulthood Care in Rare Metabolic Diseases: Results From the First Multi_Center European Survey. Front Med (Lausanne). 2021 Feb 25;8:652358. doi: 10.3389/fmed.2021.652358. PMID: 33738294; PMCID: PMC7962750
- Pajares S, et al. Implementation of second_tier tests in newborn screening for the detection of vitamin B12 related acquired and genetic disorders: results on 258,637 newborns. Orphanet J Rare Dis. 2021 Apr 30;16(1):195. doi: 10.1186/s13023_021_01784_7. PMID: 33931066; PMCID: PMC8086297.
- Mazurkiewicz_Bełdzińska M, et al. Managing CLN2 disease: a treatable neurodegenerative condition among other treatable early childhood epilepsies. Expert Rev Neurother. 2021 Mar 4:1_8. doi: 10.1080/14737175.2021.1885374. PMID: 33538188.
- Wijburg FA, et al. A multicenter open_label extension study of intrathecal heparan_N_sulfatase in patients with Sanfilippo syndrome type A. Mol Genet Metab. 2021 Jul 7:S1096_7192(21)00743_5. doi: 10.1016/j.ymgme.2021.07.001. Epub ahead of print. PMID: 34247932.
- Mengel E, et al, Efficacy and safety of arimoclomol in Niemann_Pick disease type C: Results from a double_blind, randomised, placebo_controlled, multinational phase 2/3 trial of a novel treatment. J Inherit Metab Dis. 2021 Aug 21. doi: 10.1002/jimd.12428. PMID: 34418116.
- Szot JO, et al. New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Hum Mutat. 2021 Jul;42(7):862_876. doi: 10.1002/humu.24211. Epub 2021 May 16. PMID: 33942433; PMCID: PMC8238843.
- de Castro MJ et al. Gene Therapy for Neuronopathic Mucopolysaccharidoses: State of the Art. Int J Mol Sci. 2021 Aug 25;22(17):9200. doi: 10.3390/ijms22179200. PMID: 34502108; PMCID: PMC8430935.
- Bellusci M, et al. The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes (Basel). 2021 Oct 9;12(10):1590. doi: 10.3390/genes12101590. PMID: 34680984; PMCID: PMC8535857.
- Schlüter A, et al; GWMD working group. Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. Neurology. 2022 Mar 1;98(9):e912-e923. doi: 10.1212/WNL13278. PMID: 35012964; PMCID: PMC8901178.
- Martín-Hernández E, et al. . Switching to Glycerol Phenylbutyrate in 48 Patients with Urea Cycle Disorders: Clinical Experience in Spain. J Clin Med. 2022 Aug 28;11(17):5045. doi: 10.3390/jcm11175045. PMID: 36078975; PMCID: PMC9457033.
- Rodrigues D, de Castro MJ, Crujeiras P, Duat-Rodriguez A, Marco AV, Del Toro M, Couce ML, Colón C. The LINCE Project: A Pathway for Diagnosing NCL2 Disease. Front Pediatr. 2022 Mar 29;10:876688. doi: 10.3389/fped.2022.876688. PMID: 35425725; PMCID: PMC9002010.
- Carnicer-Cáceres C, et als. Biomarkers in Fabry Disease. Implications for Clinical Diagnosis and Follow-up. J Clin Med. 2021 Apr 13;10(8):1664. doi: 10.3390/jcm10081664. PMID: 33924567; PMCID: PMC8068937.
- Tumienė B, Del Toro Riera M, et al. Multidisciplinary Care of Patients with Inherited Metabolic Diseases and Epilepsy: Current Perspectives. J Multidiscip Healthc. 2022 Mar 25;15:553-566. doi: 10.2147/JMDH.S251863. PMID: 35387391; PMCID: PMC8977775.
- Schlüter A, et al. ClinPrior: an algorithm for diagnosis and novel gene discovery by network-based prioritization. Genome Med. 2023 Sep 7;15(1):68. doi: 10.1186/s13073-023-01214-2. PMID: 37679823; PMCID: PMC10486091.
- Moreno-Galdó A, et al. Implementation of programmes for the transition of adolescents to adult care. An Pediatr (Engl Ed). 2023 Dec;99(6):422-430. doi: 10.1016/j.anpede.2023.09.014. Epub 2023 Nov 27. PMID: 38016858.

Projects

- 2001_ongoing: HOS Hunter Outcome Survey A Global, Multi_Center, Long_Term, Observational Registry of Patients with Hunter Syndrome (Mucopolysaccharidosis Type II, MPS II)
- 2010_2015: Carbaglu® Retrospective Observational Study Of Hyperammonaemia In Organic Acidaemia Decompensation Episodes Treated With Control Arm (ammonia scavengers)
- 2011_2018: SOBI NTBC: A non_interventional Post Authorization Safety Study (PASS) to evaluate long_term safety of Orfadin treatment in hypertyrosinemia type 1 (HT_1) patients in standard clinical care.
- 2013_2016: HGT_SAN_093 A Randomized, Controlled, Open_label, Multicenter, Phase IIb Safety and Efficacy Study of HGT_1410 (Recombinant Human Heparan N Sulfatase) Administration via an Intrathecal Drug Delivery Device in Pediatric Patients with Early Stage Mucopolysaccharidosis Type IIIA Disease
- 2014_ongoing: GEN_FAB_2013_01 Fabry registry
- 2014_ongoing: GEN_GAU_2013_01 Gaucher registry
- 2015_2016: SHP_610_201 _ A Open_Label Extension of Study HGT_SAN_093 Evaluating the Safety and Efficacy Study of HGT_1410 (Recombinant Human Heparan N Sulfatase) Administration via an Intrathecal Drug Delivery Device in Pediatric Patients with Mucopolysaccharidosis Type IIIA Disease
- 2015_2016: CT_ORZY_NPC_001: A prospective non_therapeutic study in patients diagnosed with Niemann_Pick disease type C. Orion Clinical Services.
- 2016_2020: CT_ORZY_NPC_002: Arimoclomol prospective double blind, randomised, placebo_controlled study in patients diagnosed with Niemann_Pick disease type C.

- 2016_2020: VTS_ 301: A Phase 2b/3 Prospective, Randomized, Double_Blind, Sham_Controlled Trial of VTS_270 (2_hydroxypropyl_β_cyclodextrin) in Subjects with Neurologic manifestations of Niemann_Pick Type C1 (NPC1) Disease
- 2019_ongoing: SHP611601 A Global, Multicenter, Open_label, Matched Historical Control Study of Intrathecal SHP611 in Subjects with Late Infantile Metachromatic Leukodystrophy
- 2021_ongoing: Understanding the Long_Term Management of Organic Acidemia Patients with CARBAGLU®: A Mixed Methods Approach • 2021_ongoing: ABT001 Gene therapy for patients with MPS IIIA.
- 2021-: Phase I/II gene transfer clinical trial of scAAV9.U1a.hSGSH for Mucopolysaccharidosis (MPS) IIIA NCT02716246
- 2022- A Phase 3 Randomized, Double-Blind, Placebo-Controlled Study of Adeno-Associated Virus (AAV) Serotype 8 (AAV8)-Mediated Gene Transfer of Human Ornithine Transcarbamylase (OTC) in Patients with Late-Onset OTC Deficiency NCT05345171
- 2022- A Phase 2/3, Multicenter, Double-Blind, Randomized Study to Determine the Efficacy and Safety of DNL310 vs Idursulfase in Pediatric Participants with Neuronopathic or Non-Neuronopathic Mucopolysaccharidosis Type II. NCT05371613

Memberships

- Metab ERN Medical Executive Board Member Metab ERN
- Metab ERN WP2 Coordinator
- Coordinator of Lysosomal Working group in the Spanish Association for IEM (AECOM)
- Teacher in Máster en Neurología Pediátrica de la Universidad Autónoma de Barcelona
- Teacher in Máster Universitario en Enfermedades Lisosomales de la Universidad de Alcalá de Henares
- Coordinator and teacher in Master en Enfermedades Metabólicas Congénitas de la Universidad de Santiago de Compostela
- Periodical conferences on MPS in the Meetings of the Spanish Association for the Study of Inborn Errors of Metabolism
- Periodical conferences in the Annual Meetings of several patients Associations (MPS, NPC)
- Periodical conferences on MPS Diagnosis and Treatment in several Hospitals of Catalonia and Spain

Other Relevant Information