# Annex 2

## Publications acknowledging ICGNMD

1. ***INTRODUCTORY ALL-ICGNMD PUBLICATION***: Neuromuscular disease genetics in underrepresented populations: increasing data diversity. **Wilson LA, Macken WL, Perry LD, Record CJ, Schon KR, Frezatti RSS, Raga S, Naidu K, Köken ÖY, Polat I, Kapapa MM, Dominik N, Efthymiou S, Morsy H, Nel M, Fassad MR, Gao F, Patel K, Schoonen M, Bisschoff M, Vorster A, Jonvik H, Human R, Lubbe E, Nonyane M, Vengalil S, Nashi S, Srivastava K, Lemmers RJLF, Reyaz A, Mishra R, Töpf A, Trainor CI, Steyn EC, Mahungu AC, van der Vliet PJ, Ceylan AC, Hiz AS, Çavdarlı B, Semerci Gündüz CN, Ceylan GG, Nagappa M, Tallapaka KB, Govindaraj P, van der Maarel SM, Narayanappa G, Nandeesh BN, Wa Somwe S, Bearden DR, Kvalsund MP, Ramdharry GM, Oktay Y, Yiş U, Topaloğlu H, Sarkozy A, Bugiardini E, Henning F, Wilmshurst JM, Heckmann JM, McFarland R, Taylor RW, Smuts I, van der Westhuizen FH, Sobreira CFDR, Tomaselli PJ, Marques W, Bhatia R, Dalal A, Srivastava MVP, Yareeda S, Nalini A, Vishnu VY, Thangaraj K, Straub V, Horvath R, Chinnery PF, Pitceathly RDS, Muntoni F, Houlden H, Vandrovcova J, Reilly MM, Hanna MG.** Brain. 2023 Jul 30:awad254. doi: 10.1093/brain/awad254. Online ahead of print. PMID: 37516995
2. Safety and efficacy of arimoclomol for inclusion body myositis: a multicentre, randomised, double-blind, placebo-controlled trial. Pedro M Machado, Michael P McDermott, Thomas Blaettler, Claus Sundgreen, Anthony A Amato, Emma Ciafaloni, Miriam Freimer, Summer B Gibson, Sarah M Jones, Todd D Levine, Thomas E Lloyd, Tahseen Mozaffar, Aziz I Shaibani, Matthew Wicklund, Anders Rosholm, Tim Dehli Carstensen, Karen Bonefeld, Anders Nørkær Jørgensen, Karina Phonekeo, Andrew J Heim, Laura Herbelin, Richard J Barohn†, Michael G Hanna†, Mazen M Dimachkie†, on behalf of the Arimoclomol in IBM Investigator Team of the Neuromuscular Study Group* Lancet Neurol 2023; 22: 900–11
3. Identification of genetic risk loci and causal insights associated with Parkinson's disease in African and African admixed populations: a genome-wide association study. Rizig M, Bandres-Ciga S, Makarious MB, Ojo OO, Crea PW, Abiodun OV, Levine KS, Abubakar SA, Achoru CO, Vitale D, Adeniji OA, Agabi OP, Koretsky MJ, Agulanna U, Hall DA, Akinyemi RO, Xie T, Ali MW, Shamim EA, Ani-Osheku I, Padmanaban M, Arigbodi OM, Standaert DG, Bello AH, Dean MN, Erameh CO, Elsayed I, Farombi TH, Okunoye O, Fawale MB, Billingsley KJ, Imarhiagbe FA, Jerez PA, Iwuozo EU, Baker B, Komolafe MA, Malik L, Nwani PO, Daida K, Nwazor EO, Miano-Burkhardt A, Nyandaiti YW, Fang ZH, Obiabo YO, Kluss JH, Odeniyi OA, Hernandez DG, Odiase FE, Tayebi N, Ojini FI, Sidranksy E, Onwuegbuzie GA, D'Souza AM, Osaigbovo GO, Berhe B, Osemwegie N, Reed X, Oshinaike OO, Leonard HL, Otubogun FM, Alvarado CX, Oyakhire SI, Ozomma SI, Samuel SC, Taiwo FT, Wahab KW, Zubair YA, Iwaki H, Kim JJ, Morris HR, Hardy J, Nalls MA, Heilbron K, Norcliffe-Kaufmann L; Nigeria Parkinson Disease Research Network; International Parkinson's Disease Genomics Consortium Africa; Black and African American Connections to Parkinson's Disease Study Group; 23andMe Research Team; Blauwendraat C, Houlden H, Singleton A, Okubadejo NU; Global Parkinson's Genetics Program. Lancet Neurol. 2023 Aug 23:S1474-4422(23)00283-1. doi: 10.1016/S1474-4422(23)00283-1. Epub ahead of print. PMID: 37633302.
4. The mutational profile in a South African cohort with inherited neuropathies and spastic paraplegia. Mahungu AC, Steyn E, Floudiotis N, Wilson LA, Vandrovcova J, Reilly MM, Record CJ, Benatar M, Wu G, Raga S, Wilmshurst JM, Naidu K, Hanna M, Nel M, Heckmann JM. Front Neurol. 2023 Aug 29;14:1239725. doi: 10.3389/fneur.2023.1239725. PMID: 37712079; PMCID: PMC10497947.

5. Genetic Neuromuscular Disorders in Zambia: Health Services Access, Utilization, and Needs. Pediatric Neurology, Kapapa M, Bearden D, Kvalsund M. ***Advance online in Pediatric Neurology***
6. Muscle MRI in periodic paralysis shows myopathy is common and correlates with intramuscular fat accumulation. **Vivekanandam V,** Seutterlin K, Matthews E, Thornton J, Jayaseelan D, Shah S, **Morrow JM**, Yousry T, **Hanna MG**. Muscle Nerve. 2023 Jul 28. doi: 10.1002/mus.27947. Online ahead of print. PMID: 37515374
7. Development of a diagnostic framework for vestibular causes of dizziness and unsteadiness in patients with multisensory neurological disease: a Delphi consensus. Male AJ, Holmes SL, **Hanna MG, Pitceathly RDS, Ramdharry GM,** Kaski D. J Neurol. 2023 Jun;270(6):3252-3257. doi: 10.1007/s00415-023-11640-2. Epub 2023 Feb 26. PMID: 36842099
8. Unexpected frequency of the pathogenic AR CAG repeat expansion in the general population. Zanovello M, Ibáñez K, Brown AL, Sivakumar P, Bombaci A, Santos L, van Vugt JJFA, Narzisi G, Karra R, Scholz SW, Ding J, Gibbs JR, Chiò A, Dalgard C, Weisburd B; American Genome Center (TAGC) consortium, Genomics England Research Consortium, Project MinE ALS Sequencing Consortium, The NYGC ALS Consortium; **Hanna MG,** Greensmith L, Phatnani H, Veldink JH, Traynor BJ, Polke J, **Houlden H,** Fratta P, Tucci A. Brain. 2023 Jul 3;146(7):2723-2729. doi: 10.1093/brain/awad050. PMID: 36797998
9. Prevalence of genetically confirmed skeletal muscle channelopathies in the era of next generation sequencing. **Vivekanandam V**, Jaibaji R, Sud R, Ellmers R, Skorupinska I, Germaine L, James N, Holmes S, Mannikko R, Jayaseelan D, **Hanna MG**. Neuromuscul Disord. 2023 Mar;33(3):270-273. doi: 10.1016/j.nmd.2023.01.007. Epub 2023 Jan 28. PMID: 36796140
10. Identification of Novel Associations and Localization of Signals in Idiopathic Inflammatory Myopathies Using Genome-Wide Imputation. Rothwell S, Amos CI, Miller FW, Rider LG, Lundberg IE, Gregersen PK, Vencovsky J, McHugh N, Limaye V, Selva-O'Callaghan A, **Hanna MG,** Machado PM, Pachman LM, Reed AM, Molberg Ø, Benveniste O, Mathiesen P, Radstake T, Doria A, De Bleecker JL, De Paepe B, Maurer B, Ollier WE, Padyukov L, O'Hanlon TP, Lee A, Wedderburn LR, Chinoy H, Lamb JA; Myositis Genetics Consortium. Arthritis Rheumatol. 2023 Jun;75(6):1021-1027. doi: 10.1002/art.42434. Epub 2023 Mar 20. PMID: 36580032
11. Anti-HMGCR myopathy: barriers to prompt recognition. Barp A, Merve A, Shah S, Desikan M, **Hanna MG, Bugiardini E**. Pract Neurol. 2023 Jun;23(3):239-242. doi: 10.1136/pn-2022-003589. Epub 2022 Dec 23. PMID: 36564213
12. Advances in methods to analyse cardiolipin and their clinical applications. Bautista JS, Falabella M, Flannery PJ, **Hanna MG,** Heales SJR, Pope SAS, **Pitceathly RDS.** Trends Analyt Chem. 2022 Dec;157:116808. doi: 10.1016/j.trac.2022.116808. PMID: 36751553
13. Discussion of off-target and tentative genomic findings may sometimes be necessary to allow evaluation of their clinical significance. Horton RH, **Macken WL, Pitceathly RDS**, Lucassen AM. J Med Ethics. 2023 Jun 20:jme-2023-109108. doi: 10.1136/jme-2023-109108. Online ahead of print. PMID: 37339848
14. Muscle magnetic resonance imaging involvement patterns in nemaline myopathies. **Perry L**, Stimpson G, Singh L, **Morrow JM,** Shah S, Baranello G, **Muntoni F, Sarkozy A.** Ann Clin Transl Neurol. 2023 Jul;10(7):1219-1229. doi: 10.1002/acn3.51816. Epub 2023 Jun 2. PMID: 37265148
15. Astrocytic pathology in Alpers' syndrome. Smith LA, Chen C, Lax NZ, **Taylor RW,** Erskine D, **McFarland R.** Acta Neuropathol Commun. 2023 May 31;11(1):86. doi: 10.1186/s40478-023-01579-w. PMID: 37259148

16. Mitochondrial Mutations Can Alter Neuromuscular Transmission in Congenital Myasthenic Syndrome and Mitochondrial Disease. O'Connor K, Spendiff S, Lochmüller H, **Horvath R.** Int J Mol Sci. 2023 May 9;24(10):8505. doi: 10.3390/ijms24108505. PMID: 37239850
17. FBXL4 suppresses mitophagy by restricting the accumulation of NIX and BNIP3 mitophagy receptors. Nguyen-Dien GT, Kozul KL, Cui Y, Townsend B, Kulkarni PG, Ooi SS, Marzio A, Carrodus N, Zuryn S, Pagano M, Parton RG, Lazarou M, Millard SS, **Taylor RW**, Collins BM, Jones MJ, Pagan JK. EMBO J. 2023 Jul 3;42(13):e112767. doi: 10.15252/embj.2022112767. Epub 2023 May 10. PMID: 37161784
18. Opinion: more mouse models and more translation needed for ALS. Fisher EMC, Greensmith L, Malaspina A, Fratta P, **Hanna MG,** Schiavo G, Isaacs AM, Orrell RW, Cunningham TJ, Arozena AA. Mol Neurodegener. 2023 May 4;18(1):30. doi: 10.1186/s13024-023-00619-2. PMID: 37143081
19. Normal Outcome With Prenatal Intervention for Riboflavin Transporter Defect. Elks N, **Wilmshurst JM, Raga SV.** Pediatr Neurol. 2023 Jul;144:16-18. doi: 10.1016/j.pediatrneurol.2023.04.004. Epub 2023 Apr 7. PMID: 37116404
20. Correlation Between the SARA and A-T NEST Clinical Severity Scores in Adults with Ataxia-Telangiectasia. Major T, Tiet MY, **Horvath R,** Hensiek AE. Cerebellum. 2023 Apr 10. doi: 10.1007/s12311-023-01528-2. Online ahead of print. PMID: 37036622
21. Pathological variants in TOP3A cause distinct disorders of mitochondrial and nuclear genome stability. Erdinc D, Rodríguez-Luis A, Fassad MR, Mackenzie S, Watson CM, Valenzuela S, Xie X, Menger KE, Sergeant K, Craig K, Hopton S, Falkous G; Genomics England Research Consortium; Poulton J, Garcia-Moreno H, Giunti P, de Moura Aschoff CA, Morales Saute JA, Kirby AJ, Toro C, Wolfe L, Novacic D, Greenbaum L, Eliyahu A, Barel O, Anikster Y, **McFarland R,** Gorman GS, Schaefer AM, Gustafsson CM, **Taylor RW,** Falkenberg M, Nicholls TJ. EMBO Mol Med. 2023 May 8;15(5):e16775. doi: 10.15252/emmm.202216775. Epub 2023 Apr 4. PMID: 37013609
22. Neuromuscular junction involvement in inherited motor neuropathies: genetic heterogeneity and effect of oral salbutamol treatment. McMacken G, Whittaker RG, Wake R, Lochmuller H, **Horvath R.** J Neurol. 2023 Jun;270(6):3112-3119. doi: 10.1007/s00415-023-11643-z. Epub 2023 Mar 4. PMID: 36869887
23. Cell lineage-specific mitochondrial resilience during mammalian organogenesis. Burr SP, Klimm F, Glynos A, Prater M, Sendon P, Nash P, Powell CA, Simard ML, Bonekamp NA, Charl J, Diaz H, Bozhilova LV, Nie Y, Zhang H, Frison M, Falkenberg M, Jones N, Minczuk M, Stewart JB, **Chinnery PF.** Cell. 2023 Mar 16;186(6):1212-1229.e21. doi: 10.1016/j.cell.2023.01.034. Epub 2023 Feb 23. PMID: 36827974
24. Incidence and risk factors for patellofemoral dislocation in adults with Charcot-Marie-Tooth disease: An observational study. Leone E, Davenport S, Robertson C, **Laurà M, Skorupinska M, Reilly MM, Ramdharry G.** Physiother Res Int. 2023 Feb 19:e1996. doi: 10.1002/pri.1996. Online ahead of print. PMID: 36807482
25. Not to Miss: Intronic Variants, Treatment, and Review of the Phenotypic Spectrum in *VPS13D*-Related Disorder. Pauly MG, Brüggemann N, **Efthymiou S,** Grözinger A, Diaw SH, Chelban V, Turchetti V, Vona B, Tadic V, **Houlden H,** Münchau A, Lohmann K. Int J Mol Sci. 2023 Jan 18;24(3):1874. doi: 10.3390/ijms24031874. PMID: 36768210
26. Mitochondrial signalling and homeostasis: from cell biology to neurological disease. Collier JJ, Oláhová M, McWilliams TG, **Taylor RW.** Trends Neurosci. 2023 Feb;46(2):137-152. doi: 10.1016/j.tins.2022.12.001. Epub 2023 Jan 10. PMID: 36635110
27. A role for BCL2L13 and autophagy in germline purifying selection of mtDNA. Kremer LS, Bozhilova LV, Rubalcava-Gracia D, Filograna R, Upadhyay M, Koolmeister C, **Chinnery PF,** Larsson NG. PLoS

Genet. 2023 Jan 6;19(1):e1010573. doi: 10.1371/journal.pgen.1010573. eCollection 2023 Jan. PMID: 36608143

28. Consolidating the association of biallelic *MAPKAPK5* pathogenic variants with a distinct syndromic neurodevelopmental disorder. Maroofian R, **Efthymiou S,** Suri M, Rahman F, Zaki MS, Maqbool S, Anwa N, Ruiz-Pérez VL, Yanovsky-Dagan S, Elpeleg O, Sudhakar S, Mankad K, Harel T, **Houlden H.** J Med Genet. 2023 Aug;60(8):791-796. doi: 10.1136/jmg-2022-108566. Epub 2022 Dec 29. PMID: 36581449
29. High diagnostic yield of targeted next-generation sequencing panel as a first-tier molecular test for the patients with myopathy or muscular dystrophy. **Çavdarlı B, Köken ÖY**, Satılmış SBA, Bilen Ş, **Ardıçlı D, Ceylan AC, Gündüz CNS, Topaloğlu H.** Ann Hum Genet. 2023 May;87(3):104-114. doi: 10.1111/ahg.12492. Epub 2022 Dec 27. PMID: 36575883
30. Biallelic variants in OGDH encoding oxoglutarate dehydrogenase lead to a neurodevelopmental disorder characterized by global developmental delay, movement disorder, and metabolic abnormalities. Whittle EF, Chilian M, Karimiani EG, Progri H, Buhas D, Kose M, Ganetzky RD, Toosi MB, Torbati PN, Badv RS, Shelihan I, Yang H, Elloumi HZ, Lee S, Jamshidi Y, Pittman AM, **Houlden H,** Ignatius E, Rahman S, Maroofian R, Yoon WH, Carroll CJ. Genet Med. 2023 Feb;25(2):100332. doi: 10.1016/j.gim.2022.11.001. Epub 2022 Dec 15. PMID: 36520152

**2022**

31. Specialist multidisciplinary input maximises rare disease diagnoses from whole genome sequencing. **Macken WL,** Falabella M, McKittrick C, Pizzamiglio C, Ellmers R, Eggleton K, Woodward CE, Patel Y, Labrum R; Genomics England Research Consortium; Phadke R, **Reilly MM,** DeVile C, Sarkozy A, Footitt E, Davison J, Rahman S, **Houlden H, Bugiardini E,** Quinlivan R, **Hanna MG, Vandrovcova J, Pitceathly RDS**. Nat Commun. 2022 Nov 7;13(1):6324. doi: 10.1038/s41467-022-32908-7. PMID: 36344503
32. Integrin α7 Mutations Are Associated With Adult-Onset Cardiac Dysfunction in Humans and Mice. **Bugiardini E,** Nunes AM, Oliveira-Santos A, Dagda M, Fontelonga TM, Barraza-Flores P, Pittman AM, Morrow JM, Parton M, Houlden H, Elliott PM, Syrris P, Maas RP, Akhtar MM, Küsters B, Raaphorst J, Schouten M, Kamsteeg EJ, van Engelen B, **Hanna MG**, Phadke R, Lopes LR, Matthews E, Burkin DJ. J Am Heart Assoc. 2022 Dec 6;11(23):e026494. doi: 10.1161/JAHA.122.026494. Epub 2022 Nov 29. PMID: 36444867
33. In silico versus functional characterization of genetic variants: lessons from muscle channelopathies. **Vivekanandam V**, Ellmers R, Jayaseelan D, **Houlden H**, Männikkö R, **Hanna MG**. Brain. 2023 Apr 19;146(4):1316-1321. doi: 10.1093/brain/awac431. PMID: 36382348
34. De novo KCNA6 variants with attenuated $K_V$ 1.6 channel deactivation in patients with epilepsy. Salpietro V, Galassi Deforie V, **Efthymiou S,** O'Connor E, Marcé-Grau A, Maroofian R, Striano P, Zara F, Morrow MM; SYNAPS Study Group; Reich A, Blevins A, Sala-Coromina J, Accogli A, Fortuna S, Alesandrini M, Au PYB, Singhal NS, Cogne B, Isidor B, **Hanna MG,** Macaya A, Kullmann DM, **Houlden H**, Männikkö R. Epilepsia. 2023 Feb;64(2):443-455. doi: 10.1111/epi.17455. Epub 2022 Dec 5. PMID: 36318112
35. Gene therapy for primary mitochondrial diseases: experimental advances and clinical challenges. Falabella M, Minczuk M, **Hanna MG,** Viscomi C, **Pitceathly RDS**. Nat Rev Neurol. 2022 Nov;18(11):689-698. doi: 10.1038/s41582-022-00715-9. Epub 2022 Oct 18. PMID: 36257993
36. Designing clinical trials for rare diseases: unique challenges and opportunities. Pizzamiglio C, Vernon HJ**, Hanna MG, Pitceathly RDS**. Nat Rev Methods Primers. 2022 Mar 10;2(1):s43586-022-00100-2. doi: 10.1038/s43586-022-00100-2. eCollection 2022 Dec. PMID: 36254116

37. Evidence of nerve hypertrophy in patients with inclusion body myositis on lower limb MRI. Elmansy M, **Morrow JM**, Shah S, Fischmann A, Wastling S, **Reilly MM, Hanna MG**, Helmy EM, El-Essawy SS, Thornton JS, Yousry TA. Muscle Nerve. 2022 Dec;66(6):744-749. doi: 10.1002/mus.27728. Epub 2022 Oct 7. PMID: 36151728
38. COVID-19 infection and vaccination in patients with skeletal muscle channelopathies. **Vivekanandam V,** Jayaseelan D, **Hanna MG.** Muscle Nerve. 2022 Nov;66(5):617-620. doi: 10.1002/mus.27704. Epub 2022 Sep 2. PMID: 36053900
39. Longitudinal Changes in MRI Muscle Morphometry and Composition in People With Inclusion Body Myositis. Laurent D, Riek J, Sinclair CDJ, Houston P, Roubenoff R, Papanicolaou DA, Nagy A, Pieper S, Yousry TA, **Hanna MG,** Thornton JS, Machado PM. Neurology. 2022 Aug 30;99(9):e865-e876. doi: 10.1212/WNL.0000000000200776. Epub 2022 Jun 3. PMID: 36038279
40. Episodic Ataxia Type 1: Natural History and Effect on Quality of Life. Graves TD, Griggs RC, Bundy BN, Jen JC, Baloh RW, **Hanna MG;** CINCH Investigators. Cerebellum. 2023 Aug;22(4):578-586. doi: 10.1007/s12311-021-01360-6. Epub 2022 Jun 3. PMID: 35655106
41. SHIP164 is a chorein motif lipid transfer protein that controls endosome-Golgi membrane traffic. **Hanna MG,** Suen PH, Wu Y, Reinisch KM, De Camilli P. J Cell Biol. 2022 Jun 6;221(6):e202111018. doi: 10.1083/jcb.202111018. Epub 2022 May 2. PMID: 35499567
42. British Society for Rheumatology guideline on management of paediatric, adolescent and adult patients with idiopathic inflammatory myopathy. Oldroyd AGS, Lilleker JB, Amin T, Aragon O, Bechman K, Cuthbert V, Galloway J, Gordon P, Gregory WJ, Gunawardena H, **Hanna MG,** Isenberg D, Jackman J, Kiely PDW, Livermore P, Machado PM, Maillard S, McHugh N, Murphy R, Pilkington C, Prabu A, Rushe P, Spinty S, Swan J, Tahir H, Tansley SL, Truepenny P, Truepenny Y, Warrier K, Yates M, Papadopoulou C, Martin N, McCann L, Chinoy H; British Society for Rheumatology Standards, Audit and Guidelines Working Group. Rheumatology (Oxford). 2022 May 5;61(5):1760-1768. doi: 10.1093/rheumatology/keac115. PMID: 35355064
43. Diagnostic and prognostic value of anti-cN1A antibodies in inclusion body myositis. Salam S, Dimachkie MM, **Hanna MG,** Machado PM. Clin Exp Rheumatol. 2022 Feb;40(2):384-393. doi: 10.55563/clinexprheumatol/r625rm. Epub 2022 Jan 25. PMID: 35225226
44. Expanding SPTAN1 monoallelic variant associated disorders: From epileptic encephalopathy to pure spastic paraplegia and ataxia. **Morsy H,** Benkirane M, Cali E, Rocca C, Zhelcheska K, Cipriani V, Galanaki E, Maroofian R, **Efthymiou S,** Murphy D, O'Driscoll M, Suri M, Banka S, Clayton-Smith J, Wright T, Redman M, Bassetti JA, Nizon M, Cogne B, Jamra RA, Bartolomaeus T, Heruth M, Krey I, Gburek-Augustat J, Wieczorek D, Gattermann F, Mcentagart M, Goldenberg A, Guyant-Marechal L, Garcia-Moreno H, Giunti P, Chabrol B, Bacrot S, Buissonnière R, Magry V, Gowda VK, Srinivasan VM, Melegh B, Szabó A, Sümegi K, Cossée M, Ziff M, Butterfield R, Hunt D, Bird-Lieberman G, **Hanna M,** Koenig M, Stankewich M, **Vandrovcova J, Houlden H;** Genomics England Research Consortium. Genet Med. 2023 Jan;25(1):76-89. doi: 10.1016/j.gim.2022.09.013. Epub 2022 Nov 4. PMID: 36331550
45. Factors associated with the severity of COVID-19 outcomes in people with neuromuscular diseases: Data from the International Neuromuscular COVID-19 Registry. Pizzamiglio C, **Pitceathly RDS,** Lunn MP, Brady S, De Marchi F, Galan L, **Heckmann JM**, Horga A, Molnar MJ, Oliveira ASB, Pinto WBVR, Primiano G, Santos E, Schoser B, Servidei S, Sgobbi Souza PV, **Venugopalan V, Hanna MG,** Dimachkie MM, Machado PM; Neuromuscular Diseases and COVID-19 Study Group. Eur J Neurol. 2023 Feb;30(2):399-412. doi: 10.1111/ene.15613. Epub 2022 Nov 18. PMID: 36303290
46. Cardiac Outcomes in Adults With Mitochondrial Diseases. Savvatis K, Vissing CR, Klouvi L, Florian A, Rahman M, Béhin A, Fayssoil A, Masingue M, Stojkovic T, Bécane HM, Berber N, Mochel F, Duboc D, Fontaine B, Krett B, Stalens C, Lejeune J, **Pitceathly RDS,** Lopes L, Saadi M, Gossios T,

Procaccio V, Spinazzi M, Tard C, De Groote P, Dhaenens CM, Douillard C, Echaniz-Laguna A, Quinlivan R, **Hanna MG,** Yilmaz A, Vissing J, Laforêt P, Elliott P, Wahbi K. J Am Coll Cardiol. 2022 Oct 11;80(15):1421-1430. doi: 10.1016/j.jacc.2022.08.716. PMID: 36202532

47. COVID-19-Related Outcomes in Primary Mitochondrial Diseases: An International Study. Pizzamiglio C, Machado PM, Thomas RH, Gorman GS, **McFarland R, Hanna MG, Pitceathly RDS**; MitoCOVID-19 Study Group. Neurology. 2022 Apr 5;98(14):576-582. doi: 10.1212/WNL.0000000000200240. Epub 2022 Feb 21. PMID: 35190464
48. Genetic testing for mitochondrial disease: the United Kingdom best practice guidelines. Mavraki E, Labrum R, Sergeant K, Alston CL, Woodward C, Smith C, Knowles CVY, Patel Y, Hodsdon P, Baines JP, Blakely EL, Polke J, **Taylor RW,** Fratter C. Eur J Hum Genet. 2023 Feb;31(2):148-163. doi: 10.1038/s41431-022-01249-w. Epub 2022 Dec 13. PMID: 36513735
49. A case for genomic medicine in South African paediatric patients with neuromuscular disease. **Raga SV, Wilmshurst JM, Smuts I, Meldau S, Bardien S, Schoonen M, van der Westhuizen FH.** Front Pediatr. 2022 Nov 17;10:1033299. doi: 10.3389/fped.2022.1033299. eCollection 2022. PMID: 36467485
50. Inclusion body myositis: from genetics to clinical trials. Nagy S, Khan A, Machado PM, **Houlden H.** J Neurol. 2023 Mar;270(3):1787-1797. doi: 10.1007/s00415-022-11459-3. Epub 2022 Nov 18. PMID: 36399165
51. Beware next-generation sequencing gene panels as the first-line genetic test in Charcot-Marie-Tooth disease. **Record CJ,** Pipis M, Poh R, Polke JM, **Reilly MM.** J Neurol Neurosurg Psychiatry. 2023 Apr;94(4):327-328. doi: 10.1136/jnnp-2022-330223. Epub 2022 Nov 14. PMID: 36376020
52. Brain monoamine vesicular transport disease caused by homozygous SLC18A2 variants: A study in 42 affected individuals. Saida K, **Maroofian R,** Sengoku T, Mitani T, Pagnamenta AT, Marafi D, Zaki MS, O'Brien TJ, Karimiani EG, Kaiyrzhanov R, Takizawa M, Ohori S, Leong HY, Akay G, Galehdari H, Zamani M, Romy R, Carroll CJ, Toosi MB, Ashrafzadeh F, Imannezhad S, Malek H, Ahangari N, Tomoum H, Gowda VK, Srinivasan VM, Murphy D, Dominik N, Elbendary HM, Rafat K, Yilmaz S, Kanmaz S, Serin M, Krishnakumar D, Gardham A, Maw A, Rao TS, Alsubhi S, Srour M, Buhas D, Jewett T, Goldberg RE, Shamseldin H, Frengen E, Misceo D, Strømme P, Magliocco Ceroni JR, Kim CA, Yesil G, Sengenc E, Guler S, Hull M, Parnes M, Aktas D, Anlar B, Bayram Y, Pehlivan D, Posey JE, Alavi S, Madani Manshadi SA, Alzaidan H, Al-Owain M, Alabdi L, Abdulwahab F, Sekiguchi F, Hamanaka K, Fujita A, Uchiyama Y, Mizuguchi T, Miyatake S, Miyake N, Elshafie RM, Salayev K, Guliyeva U, Alkuraya FS, Gleeson JG, Monaghan KG, Langley KG, Yang H, Motavaf M, Safari S, Alipour M, Ogata K, Brown AEX, Lupski JR, **Houlden H,** Matsumoto N. Genet Med. 2023 Jan;25(1):90-102. doi: 10.1016/j.gim.2022.09.010. Epub 2022 Oct 31. PMID: 36318270
53. Genotypic and phenotypic spectrum of infantile liver failure due to pathogenic TRMU variants. Vogel GF, Mozer-Glassberg Y, Landau YE, Schlieben LD, Prokisch H, Feichtinger RG, Mayr JA, Brennenstuhl H, Schröter J, Pechlaner A, Alkuraya FS, Baker JJ, Barcia G, Baric I, Braverman N, Burnyte B, Christodoulou J, Ciara E, Coman D, Das AM, Darin N, Della Marina A, Distelmaier F, Eklund EA, Ersoy M, Fang W, Gaignard P, Ganetzky RD, Gonzales E, Howard C, Hughes J, Konstantopoulou V, Kose M, Kerr M, Khan A, Lenz D, **McFarland R,** Margolis MG, Morrison K, Müller T, Murayama K, Nicastro E, Pennisi A, Peters H, Piekutowska-Abramczuk D, Rötig A, Santer R, Scaglia F, Schiff M, Shagrani M, Sharrard M, Soler-Alfonso C, Staufner C, Storey I, Stormon M, **Taylor RW,** Thorburn DR, Teles EL, Wang JS, Weghuber D, Wortmann S. Genet Med. 2023 Jun;25(6):100314. doi: 10.1016/j.gim.2022.09.015. Epub 2022 Oct 29. PMID: 36305855
54. Phenotypic continuum of NFU1-related disorders. Kaiyrzhanov R, Zaki MS, Lau T, Sen S, Azizimalamiri R, Zamani M, Sayin GY, Hilander T, Efthymiou S, Chelban V, Brown R, Thompson K, Scarano MI, Ganesh J, Koneev K, Gülaçar IM, Person R, Sadykova D, Maidyrov Y, Seifi T, Zadagali

A, Bernard G, Allis K, Elloumi HZ, Lindy A, Taghiabadi E, Verma S, Logan R, Kirmse B, Bai R, Khalaf SM, Abdel-Hamid MS, Sedaghat A, Shariati G, Issa M, Zeighami J, Elbendary HM, Brown G, Taylor RW, Galehdari H, Gleeson JJ, Carroll CJ, Cowan JA, Moreno-De-Luca A, **Houlden H,** Maroofian R. Ann Clin Transl Neurol. 2022 Dec;9(12):2025-2035. doi: 10.1002/acn3.51679. Epub 2022 Oct 18. PMID: 36256512

55. Whole-genome sequencing for mitochondrial disorders identifies unexpected mimics. **Schon KR, Chinnery PF.** Pract Neurol. 2023 Feb;23(1):2-3. doi: 10.1136/pn-2022-003570. Epub 2022 Oct 17. PMID: 36253087
56. A Novel Homozygous Founder Variant of *RTN4IP1* in Two Consanguineous Saudi Families. Aldosary M, Alsagob M, AlQudairy H, González-Álvarez AC, Arold ST, Dababo MA, Alharbi OA, Almass R, AlBakheet A, AlSarar D, Qari A, Al-Ansari MM, Oláhová M, Al-Shahrani SA, AlSayed M, Colak D, **Taylor RW,** AlOwain M, Kaya N. Cells. 2022 Oct 7;11(19):3154. doi: 10.3390/cells11193154. PMID: 36231115
57. Conduction block and temporal dispersion in a SIGMAR1-related neuropathy. **Frezatti RSS, Tomaselli PJ**, Figueiredo FB, Zuchner S, **Reilly MM, Marques W Jr.** J Peripher Nerv Syst. 2022 Dec;27(4):316-319. doi: 10.1111/jns.12517. Epub 2022 Nov 1. PMID: 36222432
58. Nuclear-embedded mitochondrial DNA sequences in 66,083 human genomes. Wei W, **Schon KR,** Elgar G, Orioli A, Tanguy M, Giess A, Tischkowitz M, Caulfield MJ, **Chinnery PF**. Nature. 2022 Nov;611(7934):105-114. doi: 10.1038/s41586-022-05288-7. Epub 2022 Oct 5. PMID: 36198798
59. Severe distinct dysautonomia in RFC1-related disease associated with Parkinsonism. **Record CJ,** Alsukhni RA, Curro R, Kaski D, Rubin JS, Morris HR, Cortese A, Iodice V, **Reilly MM.** J Peripher Nerv Syst. 2022 Dec;27(4):311-315. doi: 10.1111/jns.12515. Epub 2022 Oct 7. PMID: 36177974
60. Modulating mitochondrial DNA mutations: factors shaping heteroplasmy in the germ line and somatic cells. Chiaratti MR, **Chinnery PF.** Pharmacol Res. 2022 Nov;185:106466. doi: 10.1016/j.phrs.2022.106466. Epub 2022 Sep 27. PMID: 36174964
61. Bi-allelic LETM1 variants perturb mitochondrial ion homeostasis leading to a clinical spectrum with predominant nervous system involvement. Kaiyrzhanov R, Mohammed SEM, Maroofian R, Husain RA, Catania A, Torraco A, Alahmad A, Dutra-Clarke M, Grønborg S, Sudarsanam A, Vogt J, Arrigoni F, Baptista J, Haider S, Feichtinger RG, Bernardi P, Zulian A, Gusic M, Efthymiou S, Bai R, Bibi F, Horga A, Martinez-Agosto JA, Lam A, Manole A, Rodriguez DP, Durigon R, Pyle A, Albash B, Dionisi-Vici C, Murphy D, Martinelli D, Bugiardini E, Allis K, Lamperti C, Reipert S, Risom L, Laugwitz L, Di Nottia M, **McFarland R**, Vilarinho L, **Hanna M**, Prokisch H, Mayr JA, Bertini ES, Ghezzi D, Østergaard E, Wortmann SB, Carrozzo R, Haack TB, **Taylor RW**, Spinazzola A, Nowikovsky K, **Houlden H**. Am J Hum Genet. 2022 Sep 1;109(9):1692-1712. doi: 10.1016/j.ajhg.2022.07.007. PMID: 36055214
62. Heterozygous UCHL1 loss-of-function variants cause a neurodegenerative disorder with spasticity, ataxia, neuropathy, and optic atrophy. Park J, Tucci A, Cipriani V, Demidov G, Rocca C, Senderek J, Butryn M, Velic A, Lam T, Galanaki E, Cali E, Vestito L, Maroofian R, Deininger N, Rautenberg M, Admard J, Hahn GA, Bartels C, van Os NJH, **Horvath R, Chinnery PF**, Tiet MY, Hewamadduma C, Hadjivassiliou M, Tofaris GK; Genomics England Research Consortium; Wood NW, Hayer SN, Bender F, Menden B, Cordts I, Klein K, Nguyen HP, Krauss JK, Blahak C, Strom TM, Sturm M, van de Warrenburg B, Lerche H, Maček B, Synofzik M, Ossowski S, Timmann D, Wolf ME, Smedley D, Riess O, Schöls L, **Houlden H,** Haack TB, Hengel H. Genet Med. 2022 Oct;24(10):2079-2090. doi: 10.1016/j.gim.2022.07.006. Epub 2022 Aug 20. PMID: 35986737
63. A recurrent homozygous missense DPM3 variant leads to muscle and brain disease. Nagy S, Lau T, Alavi S, Karimiani EG, Vallian J, Ng BG, Noroozi Asl S, Akhondian J, Bahreini A, Yaghini O, Uapinyoying P, Bonnemann C, Freeze HH, Dissanayake VHW, Sirisena ND, Schmidts M, **Houlden**

**H,** Moreno-De-Luca A, Maroofian R. Clin Genet. 2022 Dec;102(6):530-536. doi: 10.1111/cge.14208. Epub 2022 Aug 19. PMID: 35932216

64. Novel DNML1 variants impair mitochondrial dynamics through divergent mechanisms. Nolden KA, Egner JM, Collier JJ, Russell OM, Alston CL, Harwig MC, Widlansky ME, Sasorith S, Barbosa IA, Douglas AG, Baptista J, Walker M, Donnelly DE, Morris AA, Tan HJ, Kurian MA, Gorman K, Mordekar S, Deshpande C, Samanta R, **McFarland R,** Hill RB, **Taylor RW,** Oláhová M. Life Sci Alliance. 2022 Aug 1;5(12):e202101284. doi: 10.26508/lsa.202101284. PMID: 35914810
65. GGPS1-associated muscular dystrophy with and without hearing loss. Kaiyrzhanov R, **Perry L,** Rocca C, Zaki MS, Hosny H, Araujo Martins Moreno C, Phadke R, Zaharieva I, Camelo Gontijo C, Beetz C, Pini V, Movahedinia M, Zanoteli E, DiTroia S, Vuillaumier-Barrot S, Isapof A, Mehrjardi MYV, Ghasemi N, **Sarkozy A, Muntoni F,** Whalen S, Vona B, **Houlden H,** Maroofian R. Ann Clin Transl Neurol. 2022 Sep;9(9):1465-1474. doi: 10.1002/acn3.51633. Epub 2022 Jul 23. PMID: 35869884
66. Exploratory analysis of lower limb muscle MRI in a case series of patients with SORD neuropathy. O'Donnell LF, Cortese A, **Rossor AM, Laura M,** Blake J, Skorupinska M, Lunn MP, Thornton JS, Currò R, **Morrow JM, Reilly MM.** J Neurol Neurosurg Psychiatry. 2022 Jul 22:jnnp-2022-329432. doi: 10.1136/jnnp-2022-329432. Online ahead of print. PMID: 35868853
67. Delineating selective vulnerability of inhibitory interneurons in Alpers' syndrome. Smith LA, Erskine D, Blain A, **Taylor RW, McFarland R**, Lax NZ. Neuropathol Appl Neurobiol. 2022 Oct;48(6):e12833. doi: 10.1111/nan.12833. Epub 2022 Jul 19. PMID: 35790454
68. Biallelic loss of EMC10 leads to mild to severe intellectual disability. Kaiyrzhanov R, Rocca C, Suri M, Gulieva S, Zaki MS, Henig NZ, Siquier K, Guliyeva U, Mounir SM, Marom D, Allahverdiyeva A, Megahed H, van Bokhoven H, Cantagrel V, Rad A, Pourkeramti A, Dehghani B, Shao DD, Markus-Bustani K, Sofrin-Drucker E, Orenstein N, Salayev K, Arrigoni F, **Houlden H,** Maroofian R. Ann Clin Transl Neurol. 2022 Jul;9(7):1080-1089. doi: 10.1002/acn3.51602. Epub 2022 Jun 9. PMID: 35684946
69. RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Shintaku J, Pernice WM, Eyaid W, Gc JB, Brown ZP, Juanola-Falgarona M, Torres-Torronteras J, Sommerville EW, Hellebrekers DM, Blakely EL, Donaldson A, van de Laar I, Leu CS, Marti R, Frank J, Tanji K, Koolen DA, Rodenburg RJ, **Chinnery PF**, Smeets HJM, Gorman GS, Bonnen PE, **Taylor RW,** Hirano M. J Clin Invest. 2022 Jul 1;132(13):e145660. doi: 10.1172/JCI145660. PMID: 35617047
70. Novel phosphopantothenoylcysteine synthetase (PPCS) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of PPCS-related disorders. Lok A, Fernandez-Garcia MA, **Taylor RW,** French C, **MacFarland R,** Bodi I, Champion M, Josifova D, Raymond FL, Iuso A, Jungbluth H, Milan A, Singh RR. Am J Med Genet A. 2022 Sep;188(9):2783-2789. doi: 10.1002/ajmg.a.62848. Epub 2022 May 26. PMID: 35616428
71. Biallelic variants in ZNF142 lead to a syndromic neurodevelopmental disorder. Christensen MB, Levy AM, Mohammadi NA, Niceta M, Kaiyrzhanov R, Dentici ML, Al Alam C, Alesi V, Benoit V, Bhatia KP, Bierhals T, Boßelmann CM, Buratti J, Callewaert B, Ceulemans B, Charles P, De Wachter M, Dehghani M, D'haenens E, Doco-Fenzy M, Geßner M, Gobert C, Guliyeva U, Haack TB, Hammer TB, Heinrich T, Hempel M, Herget T, Hoffmann U, Horvath J, **Houlden H,** Keren B, Kresge C, Kumps C, Lederer D, Lermine A, Magrinelli F, Maroofian R, Vahidi Mehrjardi MY, Moudi M, Müller AJ, Oostra AJ, Pletcher BA, Ros-Pardo D, Samarasekera S, Tartaglia M, Van Schil K, Vogt J, Wassmer E, Winkelmann J, Zaki MS, Zech M, Lerche H, Radio FC, Gomez-Puertas P, Møller RS, Tümer Z. Clin Genet. 2022 Aug;102(2):98-109. doi: 10.1111/cge.14165. Epub 2022 Jun 8. PMID: 35616059

72. Defining mitochondrial protein functions through deep multiomic profiling. Rensvold JW, Shishkova E, Sverchkov Y, Miller IJ, Cetinkaya A, Pyle A, Manicki M, Brademan DR, Alanay Y, Raiman J, Jochem A, Hutchins PD, Peters SR, Linke V, Overmyer KA, Salome AZ, Hebert AS, Vincent CE, Kwiecien NW, Rush MJP, Westphall MS, Craven M, Akarsu NA, **Taylor RW,** Coon JJ, Pagliarini DJ. Nature. 2022 Jun;606(7913):382-388. doi: 10.1038/s41586-022-04765-3. Epub 2022 May 25. PMID: 35614220
73. A translatable RNAi-driven silences PMP22/Pmp22 genes and improves neuropathy in CMT1A mice. Stavrou M, Kagiava A, Choudury SG, Jennings MJ, Wallace LM, Fowler AM, Heslegrave A, Richter J, Tryfonos C, Christodoulou C, Zetterberg H, **Horvath R,** Harper SQ, Kleopa KA. J Clin Invest. 2022 Jul 1;132(13):e159814. doi: 10.1172/JCI159814. PMID: 35579942
74. Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Alix JJP, Plesia M, Lloyd GR, Dudgeon AP, Kendall CA, Hewamadduma C, Hadjivassiliou M, McDermott CJ, Gorman GS, **Taylor RW,** Shaw PJ, Day JCC. Analyst. 2022 May 30;147(11):2533-2540. doi: 10.1039/d1an01932e. PMID: 35545877
75. Molecular and neurological features of MELAS syndrome in paediatric patients: A case series and review of the literature. Seed LM, Dean A, Krishnakumar D, Phyu P, Horvath R, Harijan PD. Mol Genet Genomic Med. 2022 Jul;10(7):e1955. doi: 10.1002/mgg3.1955. Epub 2022 Apr 26. PMID: 35474314
76. WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. Majander A, Jurkute N, Burté F, Brock K, João C, Huang H, Neveu MM, Chan CM, Duncan HJ, Kelly S, Burkitt-Wright E, Khoyratty F, Lai YT, Subash M, **Chinnery PF,** Bitner-Glindzicz M, Arno G, Webster AR, Moore AT, Michaelides M, Stockman A, Robson AG, Yu-Wai-Man P. Am J Ophthalmol. 2022 Sep;241:9-27. doi: 10.1016/j.ajo.2022.04.003. Epub 2022 Apr 22. PMID: 35469785
77. Unusual upper limb features in SORD neuropathy. **Record CJ,** Pipis M, Blake J, Curro R, Lunn MP, **Rossor AM, Laura M,** Cortese A, **Reilly MM.** J Peripher Nerv Syst. 2022 Jun;27(2):175-177. doi: 10.1111/jns.12492. Epub 2022 Apr 13. PMID: 35419909
78. Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Yépez VA, Gusic M, Kopajtich R, Mertes C, Smith NH, Alston CL, Ban R, Beblo S, Berutti R, Blessing H, Ciara E, Distelmaier F, Freisinger P, Häberle J, Hayflick SJ, Hempel M, Itkis YS, Kishita Y, Klopstock T, Krylova TD, Lamperti C, Lenz D, Makowski C, Mosegaard S, Müller MF, Muñoz-Pujol G, Nadel A, Ohtake A, Okazaki Y, Procopio E, Schwarzmayr T, Smet J, Staufner C, Stenton SL, Strom TM, Terrile C, Tort F, Van Coster R, Vanlander A, Wagner M, Xu M, Fang F, Ghezzi D, Mayr JA, Piekutowska-Abramczuk D, Ribes A, Rötig A, **Taylor RW**, Wortmann SB, Murayama K, Meitinger T, Gagneur J, Prokisch H. Genome Med. 2022 Apr 5;14(1):38. doi: 10.1186/s13073-022-01019-9. PMID: 35379322
79. Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. Calabrese C, Pyle A, Griffin H, Coxhead J, Hussain R, Braund PS, Li L, Burgess A, Munroe PB, Little L, Warren HR, Cabrera C, Hall A, Caulfield MJ, Rothwell PM, Samani NJ, Hudson G, **Chinnery PF.** PLoS Genet. 2022 Apr 1;18(4):e1010068. doi: 10.1371/journal.pgen.1010068. eCollection 2022 Apr. PMID: 35363781
80. Reply to: Juvenile PLA2G6-parkinsonism due to Indian 'Asian' p.R741Q mutation, and response to STN DBS. Magrinelli F, Rajapaksha I, Kobylecki C, Latorre A, Mulroy E, Estevez-Fraga C, **Houlden H**, Tinazzi M, Bhatia KP. Mov Disord. 2022 Mar;37(3):658-662. doi: 10.1002/mds.28955. Epub 2022 Feb 13. PMID: 35152491
81. Biallelic Loss-of-Function *NDUFA12* Variants Cause a Wide Phenotypic Spectrum from Leigh/Leigh-Like Syndrome to Isolated Optic Atrophy. Magrinelli F, Cali E, Braga VL, Yis U, Tomoum H, Shamseldin H, Raiman J, Kernstock C, Rezende Filho FM, Barsottini OGP, **Taylor RW,** Østergaard E, Tamim A, Schäferhoff K, Sallum JMF, Zaki MS, Kok F, Bhatia KP, Wissinger B, Sergeant K, Haack

TB, **Horvath R, Hiz S,** Alkuraya FS, **Houlden H,** Pedroso JL, Maroofian R. Mov Disord Clin Pract. 2022 Jan 3;9(2):218-228. doi: 10.1002/mdc3.13398. eCollection 2022 Feb. PMID: 35141356

82. Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Scala M, Wortmann SB, Kaya N, Stellingwerff MD, Pistorio A, Glamuzina E, van Karnebeek CD, Skrypnyk C, Iwanicka-Pronicka K, Piekutowska-Abramczuk D, Ciara E, Tort F, Sheidley B, Poduri A, Jayakar P, Jayakar A, Upadia J, Walano N, Haack TB, Prokisch H, Aldhalaan H, Karimiani EG, Yildiz Y, Ceylan AC, Santiago-Sim T, Dameron A, Yang H, Toosi MB, Ashrafzadeh F, Akhondian J, Imannezhad S, Mirzadeh HS, Maqbool S, Farid A, Al-Muhaizea MA, Alshwameen MO, Aldowsari L, Alsagob M, Alyousef A, AlMass R, AlHargan A, Alwadei AH, AlRasheed MM, Colak D, Alqudairy H, Khan S, Lines MA, García Cazorla MÁ, Ribes A, Morava E, Bibi F, Haider S, Ferla MP, Taylor JC, Alsaif HS, Firdous A, Hashem M, Shashkin C, Koneev K, Kaiyrzhanov R, Efthymiou S, Genomics QS, Schmitt-Mechelke T, Ziegler A, Issa MY, Elbendary HM, Striano P, Alkuraya FS, Zaki MS, Gleeson JG, Barakat TS, Bierau J, van der Knaap MS, Maroofian R, **Houlden H**. Hum Mutat. 2022 Mar;43(3):403-419. doi: 10.1002/humu.24326. Epub 2022 Jan 12. PMID: 34989426
83. Identification and characterization of novel MPC1 gene variants causing mitochondrial pyruvate carrier deficiency. Jiang H, Alahmad A, Fu S, Fu X, Liu Z, Han X, Li L, Song T, Xu M, Liu S, Wang J, Albash B, Alaqeel A, Catalina V, Prokisch H, **Taylor RW, McFarland R,** Fang F. J Inherit Metab Dis. 2022 Mar;45(2):264-277. doi: 10.1002/jimd.12462. Epub 2022 Jan 8. PMID: 34873722

**2021**

84. Mitochondrial Strokes: Diagnostic Challenges and Chameleons. Pizzamiglio C, **Bugiardini E, Macken WL**, Woodward CE, **Hanna MG, Pitceathly RDS**. Genes (Basel). 2021 Oct 19;12(10):1643. doi: 10.3390/genes12101643. PMID: 34681037
85. Ageing contributes to phenotype transition in a mouse model of periodic paralysis. Suetterlin KJ, Tan SV, Mannikko R, Phadke R, Orford M, Eaton S, Sayer AA, Grounds MD, Matthews E, Greensmith L, **Hanna MG**. JCSM Rapid Commun. 2021 Jul-Dec;4(2):245-259. doi: 10.1002/rco2.41. Epub 2021 May 5. PMID: 35174322
86. Self-reported postural symptoms predict vestibular dysfunction and falls in patients with multi-sensory impairment. Bennett E, Holmes S, Koohi N, Islam S, Bancroft M, Male A, **Hanna MG, Pitceathly RDS**, Kaski D. J Neurol. 2022 May;269(5):2788-2791. doi: 10.1007/s00415-021-10921-y. Epub 2022 Jan 5. PMID: 34984513
87. Forecasting stroke-like episodes and outcomes in mitochondrial disease. Ng YS, Lax NZ, Blain AP, Erskine D, Baker MR, Polvikoski T, Thomas RH, Morris CM, Lai M, Whittaker RG, Gebbels A, Winder A, Hall J, Feeney C, Farrugia ME, Hirst C, Roberts M, Lawthom C, Chrysostomou A, Murphy K, Baird T, Maddison P, Duncan C, Poulton J, Nesbitt V, **Hanna MG, Pitceathly RDS, Taylor RW**, Blakely EL, Schaefer AM, T**urnbull DM, McFarland R,** Gorman GS. Brain. 2022 Apr 18;145(2):542-554. doi: 10.1093/brain/awab353. PMID: 34927673
88. Andersen-Tawil syndrome: deep phenotyping reveals significant cardiac and neuromuscular morbidity. **Vivekanandam V**, Männikkö R, Skorupinska I, Germain L, Gray B, Wedderburn S, Kozyra D, Sud R, James N, Holmes S, Savvatis K, Fialho D, Merve A, Pattni J, Farrugia M, Behr ER, Marini-Bettolo C, **Hanna MG,** Matthews E. Brain. 2022 Jun 30;145(6):2108-2120. doi: 10.1093/brain/awab445. PMID: 3491963[5]
89. 2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Pantic B, Ives D, Mennuni M, Perez-Rodriguez D, Fernandez-Pelayo U, Lopez de Arbina A, Muñoz-Oreja M, Villar-Fernandez M, Dang TJ, Vergani L, Johnston IG, **Pitceathly RDS, McFarland R, Hanna MG, Taylor RW**, Holt IJ,

Spinazzola A. Nat Commun. 2021 Dec 6;12(1):6997. doi: 10.1038/s41467-021-26829-0. PMID: 34873176

90. Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. Ghosh SG, Becker K, Huang H, Salazar TD, Chai G, Salpietro V, Al-Gazali L, Waisfisz Q, Wang H, Vaux KK, Stanley V, Manole A, Akpulat U, Weiss MM, **Efthymiou S, Hanna MG**, Minetti C, Striano P, Pisciotta L, De Grandis E, Altmüller J, Weixler L, Nürnberg P, Thiele H, Yis U, Okur TD, Polat AI, Amiri N, Doosti M, Karimani EG, Toosi MB, Haddad G, Karakaya M, Wirth B, van Hagen JM, Wolf NI, Maroofian R, **Houlden H,** Cirak S, Gleeson JG. Am J Hum Genet. 2021 Dec 2;108(12):2385. doi: 10.1016/j.ajhg.2021.11.013. PMID: 34861176
91. Long-term Safety and Efficacy of Mexiletine in Myotonic Dystrophy Types 1 and 2. Mousele C, Matthews E, **Pitceathly RDS, Hanna MG**, MacDonald S, Savvatis K, Carr A, Turner C. Neurol Clin Pract. 2021 Oct;11(5):e682-e685. doi: 10.1212/CPJ.0000000000001073. PMID: 34840883
92. The long exercise test as a functional marker of periodic paralysis. Ribeiro A, Suetterlin KJ, Skorupinska I, Tan SV, **Morrow JM,** Matthews E, **Hanna MG,** Fialho D. Muscle Nerve. 2022 May;65(5):581-585. doi: 10.1002/mus.27465. Epub 2021 Dec 6. PMID: 34817893
93. Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. **Schon KR, Horvath R,** Wei W, Calabrese C, Tucci A, Ibañez K, Ratnaike T, **Pitceathly RDS, Bugiardini E**, Quinlivan R, **Hanna MG**, Clement E, Ashton E, Sayer JA, Brennan P, Josifova D, Izatt L, Fratter C, Nesbitt V, Barrett T, McMullen DJ, Smith A, Deshpande C, Smithson SF, Festenstein R, Canham N, Caulfield M, **Houlden H**, Rahman S, **Chinnery PF;** Genomics England Research Consortium. BMJ. 2021 Nov 3;375:e066288. doi: 10.1136/bmj-2021-066288. PMID: 34732400 Clinical Trial.
94. A cross-sectional study of memory and executive functions in patients with sporadic inclusion body myositis. Lu K, Yong KXX, Skorupinska I, Deriziotis S, Collins JD, Henley SMD, **Hanna MG**, Rossor MN, Ridha BH, Machado PM. Muscle Nerve. 2022 Jan;65(1):105-109. doi: 10.1002/mus.27426. Epub 2021 Oct 21. PMID: 34605039
95. Cardiopulmonary exercise performance and factors associated with aerobic capacity in neuromuscular diseases. **Ramdharry GM**, Wallace A, Hennis P, Dewar E, Dudziec M, Jones K, Pietrusz A, **Reilly MM, Hanna MG**. Muscle Nerve. 2021 Dec;64(6):683-690. doi: 10.1002/mus.27423. Epub 2021 Oct 6. PMID: 34550609
96. Translating genetic and functional data into clinical practice: a series of 223 families with myotonia. Suetterlin K, Matthews E, Sud R, McCall S, Fialho D, Burge J, Jayaseelan D, Haworth A, Sweeney MG, Kullmann DM, Schorge S, **Hanna MG**, Männikkö R. Brain. 2022 Apr 18;145(2):607-620. doi: 10.1093/brain/awab344. PMID: 34529042
97. Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Lin SJ, Vona B, Barbalho PG, Kaiyrzhanov R, Maroofian R, Petree C, Severino M, Stanley V, Varshney P, Bahena P, Alzahrani F, Alhashem A, Pagnamenta AT, Aubertin G, Estrada-Veras JI, Hernández HAD, Mazaheri N, Oza A, Thies J, Renaud DL, Dugad S, McEvoy J, Sultan T, Pais LS, Tabarki B, Villalobos-Ramirez D, Rad A; Genomics England Research Consortium; Galehdari H, Ashrafzadeh F, Sahebzamani A, Saeidi K, Torti E, Elloumi HZ, Mora S, Palculict TB, Yang H, Wren JD, Ben Fowler, Joshi M, Behra M, Burgess SM, Nath SK, **Hanna MG**, Kenna M, Merritt JL 2nd, **Houlden H**, Karimiani EG, Zaki MS, Haaf T, Alkuraya FS, Gleeson JG, Varshney GK. Genet Med. 2021 Oct;23(10):1933-1943. doi: 10.1038/s41436-021-01239-1. Epub 2021 Jun 25. PMID: 34172899
98. Correction: VPS13D bridges the ER to mitochondria and peroxisomes via Miro. Guillén-Samander A, Leonzino M, **Hanna MG**, Tang N, Shen H, De Camilli P. J Cell Biol. 2021 Aug

2;220(8):e20201000405052021c. doi: 10.1083/jcb.20201000405052021c. Epub 2021 Jun 22. PMID: 34156432

99. Long-term efficacy and safety of dichlorphenamide for treatment of primary periodic paralysis. Sansone VA, Johnson NE, **Hanna MG**, Ciafaloni E, Statland JM, Shieh PB, Cohen F, **Griggs RC**. Muscle Nerve. 2021 Sep;64(3):342-346. doi: 10.1002/mus.27354. Epub 2021 Jul 9. PMID: 34129236 Clinical Trial.

100. A form of muscular dystrophy associated with pathogenic variants in JAG2. Coppens S, Barnard AM, Puusepp S, Pajusalu S, Õunap K, Vargas-Franco D, Bruels CC, Donkervoort S, Pais L, Chao KR, Goodrich JK, England EM, Weisburd B, Ganesh VS, Gudmundsson S, O'Donnell-Luria A, Nigul M, Ilves P, Mohassel P, Siddique T, Milone M, Nicolau S, Maroofian R, **Houlden H, Hanna MG**, Quinlivan R, Toosi MB, Karimiani EG, Costagliola S, Deconinck N, Kadhim H, Macke E, Lanpher BC, Klee EW, Łusakowska A, Kostera-Pruszczyk A, Hahn A, Schrank B, Nishino I, Ogasawara M, El Sherif R, Stojkovic T, Nelson I, Bonne G, Cohen E, Boland-Augé A, Deleuze JF, Meng Y, Töpf A, Vilain C, Pacak CA, Rivera-Zengotita ML, Bönnemann CG, Straub V, Handford PA, Draper I, Walter GA, Kang PB. Am J Hum Genet. 2021 Jun 3;108(6):1164. doi: 10.1016/j.ajhg.2021.04.018. PMID: 34087166

101. Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. Matalonga L, Hernández-Ferrer C, Piscia D; Solve-RD SNV-indel working group; Schüle R, Synofzik M, **Töpf A**, Vissers LELM, de Voer R; Solve-RD DITF-GENTURIS; Solve-RD DITF-ITHACA; Solve-RD DITF-euroNMD; Solve-RD DITF-RND; Tonda R, Laurie S, Fernandez-Callejo M, Picó D, Garcia-Linares C, Papakonstantinou A, Corvó A, Joshi R, Diez H, Gut I, Hoischen A, Graessner H, Beltran S; Solve-RD Consortia. Eur J Hum Genet. 2021 Sep;29(9):1337-1347. doi: 10.1038/s41431-021-00852-7. Epub 2021 Jun 1. PMID: 34075210

102. Annual Renal Ultrasound May Prevent Acute Presentation With Acetazolamide-Associated Urolithiasis. Suetterlin KJ, **Vivekanandam V**, James N, Sud R, Holmes S, Fialho D, **Hanna MG**, Matthews EL. Neurol Clin Pract. 2021 Feb;11(1):e40-e42. doi: 10.1212/CPJ.0000000000000761. PMID: 33968492

103. VPS13D bridges the ER to mitochondria and peroxisomes via Miro. Guillén-Samander A, Leonzino M, **Hanna MG**, Tang N, Shen H, De Camilli P. J Cell Biol. 2021 May 3;220(5):e202010004. doi: 10.1083/jcb.202010004. PMID: 33891013

104. A form of muscular dystrophy associated with pathogenic variants in JAG2. Coppens S, Barnard AM, Puusepp S, Pajusalu S, Õunap K, Vargas-Franco D, Bruels CC, Donkervoort S, Pais L, Chao KR, Goodrich JK, England EM, Weisburd B, Ganesh VS, Gudmundsson S, O'Donnell-Luria A, Nigul M, Ilves P, Mohassel P, Siddique T, Milone M, Nicolau S, Maroofian R, **Houlden H, Hanna MG**, Quinlivan R, Beiraghi Toosi M, Ghayoor Karimiani E, Costagliola S, Deconinck N, Kadhim H, Macke E, Lanpher BC, Klee EW, Łusakowska A, Kostera-Pruszczyk A, Hahn A, Schrank B, Nishino I, Ogasawara M, El Sherif R, Stojkovic T, Nelson I, Bonne G, Cohen E, Boland-Augé A, Deleuze JF, Meng Y, **Töpf A,** Vilain C, Pacak CA, Rivera-Zengotita ML, Bönnemann CG, **Straub V**, Handford PA, Draper I, Walter GA, Kang PB. Am J Hum Genet. 2021 May 6;108(5):840-856. doi: 10.1016/j.ajhg.2021.03.020. Epub 2021 Apr 15. PMID: 33861953

105. Longitudinal observational study investigating outcome measures for clinical trials in inclusion body myositis. Sangha G, Yao B, Lunn D, **Skorupinska I,** Germain L, Kozyra D, Parton M, Miller J, **Hanna MG**, Hilton-Jones D, Freebody J, Machado PM. J Neurol Neurosurg Psychiatry. 2021 Apr 13:jnnp-2020-325141. doi: 10.1136/jnnp-2020-325141. Online ahead of print. PMID: 33849999

106. Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Horga A, Manole A, Mitchell AL, **Bugiardini E,** Hargreaves IP, Mowafi W, Bettencourt C, Blakely EL, He L, Polke JM, Woodward CE, Dalla Rosa I, Shah S, Pittman AM,

Quinlivan R, **Reilly MM, Taylor RW**, Holt IJ, **Hanna MG, Pitceathly RDS**, Spinazzola A, **Houlden H.** Mol Biol Rep. 2021 Mar;48(3):2093-2104. doi: 10.1007/s11033-021-06188-1. Epub 2021 Mar 19. PMID: 33742325

107. Cardiolipin, Mitochondria, and Neurological Disease. Falabella M, Vernon HJ, **Hanna MG**, Claypool SM, **Pitceathly RDS.** Trends Endocrinol Metab. 2021 Apr;32(4):224-237. doi: 10.1016/j.tem.2021.01.006. Epub 2021 Feb 24. PMID: 33640250

108. Applying genomic and transcriptomic advances to mitochondrial medicine. **Macken WL, Vandrovcova J**, **Hanna MG, Pitceathly RDS**. Nat Rev Neurol. 2021 Apr;17(4):215-230. doi: 10.1038/s41582-021-00455-2. Epub 2021 Feb 23. PMID: 33623159

109. Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis: Long-term Extension of RESILIENT. Amato AA, **Hanna MG,** Machado PM, Badrising UA, Chinoy H, Benveniste O, Karanam AK, Wu M, Tankó LB, Schubert-Tennigkeit AA, Papanicolaou DA, Lloyd TE, Needham M, Liang C, Reardon KA, de Visser M, Ascherman DP, Barohn RJ, Dimachkie MM, Miller JAL, Kissel JT, Oskarsson B, Joyce NC, Van den Bergh P, Baets J, De Bleecker JL, Karam C, David WS, Mirabella M, Nations SP, Jung HH, Pegoraro E, Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, Yamashita S, Suzuki N, Aoki M, Katsuno M, Morihata H, Murata K, Nodera H, Nishino I, Romano CD, Williams VSL, Vissing J, Zhang Auberson L; RESILIENT Study Extension Group. Neurology. 2021 Mar 23;96(12):e1595-e1607. doi: 10.1212/WNL.0000000000011626. Epub 2021 Feb 17. PMID: 33597289 Clinical Trial.

110. Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. Hathazi D, Griffin H, Jennings MJ, Giunta M, Powell C, Pearce SF, Munro B, Wei W, Boczonadi V, Poulton J, Pyle A, Calabrese C, Gomez-Duran A, Schara U, **Pitceathly RDS, Hanna MG**, Joost K, Cotta A, Paim JF, Navarro MM, Duff J, Mattman A, Chapman K, Servidei S, Della Marina A, Uusimaa J, Roos A, Mootha V, Hirano M, Tulinius M, Giri M, Hoffmann EP, Lochmüller H, DiMauro S, Minczuk M, **Chinnery PF,** Müller JS**, Horvath R.** EMBO J. 2020 Dec 1;39(23):e105364. doi: 10.15252/embj.2020105364. Epub 2020 Oct 31. PMID: 33128823

111. Andersen-Tawil Syndrome Presenting with Complete Heart Block. Suetterlin K, Männikkö R, Flossmann E, Sud R, Fialho D, **Vivekanandam V,** James N, Gossios TD, **Hanna MG**, Savvatis K, Matthews E. J Neuromuscul Dis. 2021;8(1):151-154. doi: 10.3233/JND-200572. PMID: 33074188

112. Musclesense: a Trained, Artificial Neural Network for the Anatomical Segmentation of Lower Limb Magnetic Resonance Images in Neuromuscular Diseases. Kanber B, **Morrow JM**, Klickovic U, Wastling S, Shah S, Fratta P, McDowell AR, Hall MG, Clark CA, **Muntoni F, Reilly MM, Hanna MG,** Alexander DC, Yousry T, Thornton JS. Neuroinformatics. 2021 Apr;19(2):379-383. doi: 10.1007/s12021-020-09485-5. PMID: 32892313

113. 2-Deoxy-D-glucose couples mitochondrial DNA replication with mitochondrial fitness and promotes the selection of wild-type over mutant mitochondrial DNA. Pantic B, Ives D, Mennuni M, Perez-Rodriguez D, Fernandez-Pelayo U, Lopez de Arbina A, Muñoz-Oreja M, Villar-Fernandez M, Dang TJ, Vergani L, Johnston IG, **Pitceathly RDS, McFarland R, Hanna MG, Taylor RW,** Holt IJ, Spinazzola A. Nat Commun. 2021 Dec 6;12(1):6997. doi: 10.1038/s41467-021-26829-0. PMID: 34873176

114. Iterative Reanalysis of Hypertrophic Cardiomyopathy Exome Data Reveals Causative Pathogenic Mitochondrial DNA Variants. Lopes LR, Murphy D, **Bugiardini E,** Salem R, Jager J, Futema M, Majid Akhtar M, Savvatis K, Woodward C, Pittman AM, **Hanna MG,** Syrris P, **Pitceathly RDS**, Elliott PM. Circ Genom Precis Med. 2021 Jun;14(3):e003388. doi: 10.1161/CIRCGEN.121.003388. Epub 2021 May 10. PMID: 33970670

115. Mitochondrial DNA Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. Poole OV, Pizzamiglio C, Murphy D, Falabella M, **Macken WL, Bugiardini**

**E,** Woodward CE, Labrum R, **Efthymiou S**, Salpietro V, Chelban V, Kaiyrzhanov R, Maroofian R; SYNaPS Study Group; Amato AA, Gregory A, Hayflick SJ; Queen Square Genomics; Jonvik H, Wood N, **Houlden H, Vandrovcova J, Hanna MG**, Pittman A, **Pitceathly RDS**. Ann Neurol. 2021 Jun;89(6):1240-1247. doi: 10.1002/ana.26063. Epub 2021 Apr 1. PMID: 33704825

116. Mitochondrial DNA variants in genomic data: diagnostic uplifts and predictive implications. **Macken WL**, Lucassen AM, **Hanna MG, Pitceathly RDS**. Nat Rev Genet. 2021 Sep;22(9):547-548. doi: 10.1038/s41576-021-00381-5. PMID: 34050335

117. Single-molecule mitochondrial DNA sequencing shows no evidence of CpG methylation in human cells and tissues. Bicci I, Calabrese C, Golder ZJ, Gomez-Duran A, **Chinnery PF.** Nucleic Acids Res. 2021 Dec 16;49(22):12757-12768. doi: 10.1093/nar/gkab1179. PMID: 34850165

118. Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. Yap ZY, **Efthymiou S,** Seiffert S, Vargas Parra K, Lee S, Nasca A, Maroofian R, Schrauwen I, Pendziwiat M, Jung S, Bhoj E, Striano P, Mankad K, Vona B, Cuddapah S, Wagner A, Alvi JR, Davoudi-Dehaghani E, Fallah MS, Gannavarapu S, Lamperti C, Legati A, Murtaza BN, Nadeem MS, Rehman MU, Saeidi K, Salpietro V, von Spiczak S, Sandoval A, Zeinali S, Zeviani M, Reich A; SYNaPS Study Group; University of Washington Center for Mendelian Genomics; Jang C, Helbig I, Barakat TS, Ghezzi D, Leal SM, Weber Y, **Houlden H,** Yoon WH. Am J Hum Genet. 2021 Dec 2;108(12):2368-2384. doi: 10.1016/j.ajhg.2021.11.003. Epub 2021 Nov 19. PMID: 34800363

119. The Effect of $tRNA^{[Ser]Sec}$ Isopentenylation on Selenoprotein Expression. Fradejas-Villar N, Bohleber S, Zhao W, Reuter U, Kotter A, Helm M, Knoll R, **McFarland R, Taylor RW,** Mo Y, Miyauchi K, Sakaguchi Y, Suzuki T, Schweizer U. Int J Mol Sci. 2021 Oct 23;22(21):11454. doi: 10.3390/ijms222111454. PMID: 34768885

120. 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. 100,000 Genomes Project Pilot Investigators; Smedley D, Smith KR, Martin A, Thomas EA, McDonagh EM, Cipriani V, Ellingford JM, Arno G, Tucci A, **Vandrovcova J,** Chan G, Williams HJ, Ratnaike T, Wei W, Stirrups K, Ibanez K, Moutsianas L, Wielscher M, Need A, Barnes MR, Vestito L, Buchanan J, Wordsworth S, Ashford S, Rehmström K, Li E, Fuller G, Twiss P, Spasic-Boskovic O, Halsall S, Floto RA, Poole K, Wagner A, Mehta SG, Gurnell M, Burrows N, James R, Penkett C, Dewhurst E, Gräf S, Mapeta R, Kasanicki M, Haworth A, Savage H, Babcock M, Reese MG, Bale M, Baple E, Boustred C, Brittain H, de Burca A, Bleda M, Devereau A, Halai D, Haraldsdottir E, Hyder Z, Kasperaviciute D, Patch C, Polychronopoulos D, Matchan A, Sultana R, Ryten M, Tavares ALT, Tregidgo C, Turnbull C, Welland M, Wood S, Snow C, Williams E, Leigh S, Foulger RE, Daugherty LC, Niblock O, Leong IUS, Wright CF, Davies J, Crichton C, Welch J, Woods K, Abulhoul L, Aurora P, Bockenhauer D, Broomfield A, Cleary MA, Lam T, Dattani M, Footitt E, Ganesan V, Grunewald S, Compeyrot-Lacassagne S, Muntoni F, Pilkington C, Quinlivan R, Thapar N, Wallis C, Wedderburn LR, Worth A, Bueser T, Compton C, Deshpande C, Fassihi H, Haque E, Izatt L, Josifova D, Mohammed S, Robert L, Rose S, Ruddy D, Sarkany R, Say G, Shaw AC, Wolejko A, Habib B, Burns G, Hunter S, Grocock RJ, Humphray SJ, Robinson PN, Haendel M, Simpson MA, Banka S, Clayton-Smith J, Douzgou S, Hall G, Thomas HB, O'Keefe RT, Michaelides M, Moore AT, Malka S, Pontikos N, Browning AC, Straub V, Gorman GS, **Horvath R,** Quinton R, Schaefer AM, Yu-Wai-Man P, **Turnbull DM, McFarland R, Taylor RW,** O'Connor E, Yip J, Newland K, Morris HR, Polke J, Wood NW, Campbell C, Camps C, Gibson K, Koelling N, Lester T, Németh AH, Palles C, Patel S, Roy NBA, Sen A, Taylor J, Cacheiro P, Jacobsen JO, Seaby EG, Davison V, Chitty L, Douglas A, Naresh K, McMullan D, Ellard S, Temple IK, Mumford AD, Wilson G, Beales P, Bitner-Glindzicz M, Black G, Bradley JR, Brennan P, Burn J, **Chinnery PF**, Elliott P, Flinter F, **Houlden H,** Irving M, Newman W, Rahman S, Sayer JA, Taylor JC, Webster AR, Wilkie AOM, Ouwehand WH, Raymond FL, Chisholm

J, Hill S, Bentley D, Scott RH, Fowler T, Rendon A, Caulfield M. N Engl J Med. 2021 Nov 11;385(20):1868-1880. doi: 10.1056/NEJMoa2035790. PMID: 34758253

121. Characterising a homozygous two-exon deletion in UQCRH: comparing human and mouse phenotypes. Vidali S, Gerlini R, Thompson K, Urquhart JE, Meisterknecht J, Aguilar-Pimentel JA, Amarie OV, Becker L, Breen C, Calzada-Wack J, Chhabra NF, Cho YL, da Silva-Buttkus P, Feichtinger RG, Gampe K, Garrett L, Hoefig KP, Hölter SM, Jameson E, Klein-Rodewald T, Leuchtenberger S, Marschall S, Mayer-Kuckuk P, Miller G, Oestereicher MA, Pfannes K, Rathkolb B, Rozman J, Sanders C, Spielmann N, Stoeger C, Szibor M, Treise I, Walter JH, Wurst W, Mayr JA, Fuchs H, Gärtner U, Wittig I, **Taylor RW,** Newman WG, Prokisch H, Gailus-Durner V, Hrabě de Angelis M. EMBO Mol Med. 2021 Dec 7;13(12):e14397. doi: 10.15252/emmm.202114397. Epub 2021 Nov 8. PMID: 34750991

122. Constitutive activation of the PI3K-Akt-mTORC1 pathway sustains the m.3243 A > G mtDNA mutation. Chung CY, Singh K, Kotiadis VN, Valdebenito GE, Ahn JH, Topley E, Tan J, Andrews WD, Bilanges B, **Pitceathly RDS,** Szabadkai G, Yuneva M, Duchen MR. Nat Commun. 2021 Nov 4;12(1):6409. doi: 10.1038/s41467-021-26746-2. PMID: 34737295

123. Primary mitochondrial myopathies in childhood. Olimpio C, Tiet MY, **Horvath R.** Neuromuscul Disord. 2021 Oct;31(10):978-987. doi: 10.1016/j.nmd.2021.08.005. PMID: 34736635

124. Emerging roles of ATG7 in human health and disease. Collier JJ, Suomi F, Oláhová M, McWilliams TG, **Taylor RW.** EMBO Mol Med. 2021 Dec 7;13(12):e14824. doi: 10.15252/emmm.202114824. Epub 2021 Nov 2. PMID: 34725936

125. Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Lim AZ, Ng YS, Blain A, Jiminez-Moreno C, Alston CL, Nesbitt V, Simmons L, Santra S, Wassmer E, Blakely EL, **Turnbull DM, Taylor RW,** Gorman GS, **McFarland R.** Ann Neurol. 2022 Jan;91(1):117-130. doi: 10.1002/ana.26260. Epub 2021 Nov 12. PMID: 34716721

126. A Novel Homozygous *ADCY5* Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. Kaiyrzhanov R, Zaki MS, Maroofian R, **Dominik N**, Rad A, Vona B, **Houlden H**. Mov Disord Clin Pract. 2021 Jul 31;8(7):1140-1143. doi: 10.1002/mdc3.13310. eCollection 2021 Oct. PMID: 34631954

127. Dissecting the Phenotype and Genotype of PLA2G6-Related Parkinsonism. Magrinelli F, Mehta S, Di Lazzaro G, Latorre A, Edwards MJ, Balint B, Basu P, Kobylecki C, Groppa S, Hegde A, Mulroy E, Estevez-Fraga C, Arora A, Kumar H, Schneider SA, Lewis PA, Jaunmuktane Z, Revesz T, Gandhi S, Wood NW, Hardy JA, Tinazzi M, Lal V, **Houlden H,** Bhatia KP. Mov Disord. 2022 Jan;37(1):148-161. doi: 10.1002/mds.28807. Epub 2021 Oct 8. PMID: 34622992

128. Homozygous missense *WIPI2* variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Maroofian R, Gubas A, Kaiyrzhanov R, Scala M, Hundallah K, Severino M, Abdel-Hamid MS, Rosenfeld JA, Ebrahimi-Fakhari D, Ali Z, Rahim F, **Houlden H**, Tooze SA, Alsaleh NS, Zaki MS. Brain Commun. 2021 Sep 3;3(3):fcab183. doi: 10.1093/braincomms/fcab183. eCollection 2021. PMID: 34557665

129. α-Synuclein (SNCA) A30G Mutation as a Cause of a Complex Phenotype Without Parkinsonism. Sokratous M, Breza M, Senkevich K, Gan-Or Z, Kalampokini S, Spanaki C, Provatas A, Zaunmuktane Z, Valotassiou V, Georgoulias P, **Efthymiou S**, Hadjigeorgiou GM, **Houlden H,** Xiromerisiou G. Mov Disord. 2021 Sep;36(9):2209-2212. doi: 10.1002/mds.28735. PMID: 34543462

130. Charcot-Marie-Tooth disease type 2CC due to *NEFH* variants causes a progressive, non-length-dependent, motor-predominant phenotype. Pipis M, Cortese A, Polke JM, Poh R, **Vandrovcova J, Laura M**, Skorupinska M, Jacquier A, Juntas-Morales R, Latour P, Petiot P, Sole G,

Fromes Y, Shah S, Blake J, Choi BO, Chung KW, Stojkovic T, **Rossor AM, Reilly MM**. J Neurol Neurosurg Psychiatry. 2022 Jan;93(1):48-56. doi: 10.1136/jnnp-2021-327186. Epub 2021 Sep 13. PMID: 34518334

131. GM1-Gangliosidosis Type III Associated Parkinsonism. Kaiyrzhanov R, Guliyeva U, Gulieva S, Salayev K, Mursalova A, Allahyarova P, Ferla MP**, Houlden H.** Mov Disord Clin Pract. 2021 Sep 3;8(Suppl 1):S21-S23. doi: 10.1002/mdc3.13289. eCollection 2021 Aug. PMID: 34514040

132. MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Ratnaike TE, Greene D, Wei W, Sanchis-Juan A, **Schon KR,** van den Ameele J, Raymond L, **Horvath R,** Turro E, **Chinnery PF**. Nucleic Acids Res. 2021 Sep 27;49(17):9686-9695. doi: 10.1093/nar/gkab726. PMID: 34428295

133. Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Weerts MJA, Lanko K, Guzmán-Vega FJ, Jackson A, Ramakrishnan R, Cardona-Londoño KJ, Peña-Guerra KA, van Bever Y, van Paassen BW, Kievit A, van Slegtenhorst M, Allen NM, Kehoe CM, Robinson HK, Pang L, Banu SH, Zaman M, **Efthymiou S, Houlden H,** Järvelä I, Lauronen L, Määttä T, Schrauwen I, Leal SM, Ruivenkamp CAL, Barge-Schaapveld DQCM, Peeters-Scholte CMPCD, Galehdari H, Mazaheri N, Sisodiya SM, Harrison V, Sun A, Thies J, Pedroza LA, Lara-Taranchenko Y, Chinn IK, Lupski JR, Garza-Flores A, McGlothlin J, Yang L, Huang S, Wang X, Jewett T, Rosso G, Lin X, Mohammed S, Merritt JL 2nd, Mirzaa GM, Timms AE, Scheck J, Elting MW, Polstra AM, Schenck L, Ruzhnikov MRZ, Vetro A, Montomoli M, Guerrini R, Koboldt DC, Mosher TM, Pastore MT, McBride KL, Peng J, Pan Z, Willemsen M, Koning S, Turnpenny PD, de Vries BBA, Gilissen C, Pfundt R, Lees M, Braddock SR, Klemp KC, Vansenne F, van Gijn ME, Quindipan C, Deardorff MA, Hamm JA, Putnam AM, Baud R, Walsh L, Lynch SA, Baptista J, Person RE, Monaghan KG, Crunk A, Keller-Ramey J, Reich A, Elloumi HZ, Alders M, Kerkhof J, McConkey H, Haghshenas S; Genomics England Research Consortium; Maroofian R, Sadikovic B, Banka S, Arold ST, Barakat TS. Genet Med. 2021 Nov;23(11):2122-2137. doi: 10.1038/s41436-021-01246-2. Epub 2021 Aug 3. PMID: 34345025

134. A novel MT-CO2 variant causing cerebellar ataxia and neuropathy: The role of muscle biopsy in diagnosis and defining pathogenicity. Baty K, Farrugia ME, Hopton S, Falkous G, Schaefer AM, Stewart W, Willison HJ, **Reilly MM,** Blakely EL, **Taylor RW,** Ng YS. Neuromuscul Disord. 2021 Nov;31(11):1186-1193. doi: 10.1016/j.nmd.2021.05.014. Epub 2021 Jun 4. PMID: 34325999

135. ATG7 safeguards human neural integrity. Collier JJ, Oláhová M, McWilliams TG, **Taylor RW.** Autophagy. 2021 Sep;17(9):2651-2653. doi: 10.1080/15548627.2021.1953267. Epub 2021 Jul 27. PMID: 34313536

136. Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Iqbal M, Maroofian R, **Çavdarlı B,** Riccardi F, Field M, Banka S, Bubshait DK, Li Y, Hertecant J, Baig SM, Dyment D, **Efthymiou S**, Abdullah U, Makhdoom EUH, Ali Z, Scherf de Almeida T, Molinari F, Mignon-Ravix C, Chabrol B, Antony J, Ades L, Pagnamenta AT, Jackson A, Douzgou S; Genomics England Research Consortium; Beetz C, Karageorgou V, Vona B, Rad A, Baig JM, Sultan T, Alvi JR, Maqbool S, Rahman F, Toosi MB, Ashrafzadeh F, Imannezhad S, Karimiani EG, Sarwar Y, Khan S, Jameel M, Noegel AA, Budde B, Altmüller J, Motameny S, Höhne W, **Houlden H,** Nürnberg P, Wollnik B, Villard L, Alkuraya FS, Osmond M, Hussain MS, Yigit G. Genet Med. 2021 Nov;23(11):2138-2149. doi: 10.1038/s41436-021-01260-4. Epub 2021 Jul 9. PMID: 34244665

137. Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. Zaki MS, Accogli A, Mirzaa G, Rahman F, Mohammed H, Porras-Hurtado GL, **Efthymiou S,** Maqbool S, Shukla A, Vincent JB, Hussain A, Mir A, Beetz C, Leubauer A, **Houlden H**, Gleeson JG, Maroofian R. Eur J Hum Genet. 2021 Aug;29(8):1226-1234. doi: 10.1038/s41431-021-00910-0. Epub 2021 Jun 24. PMID: 34163010

138. Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. Collier JJ, Guissart C, Oláhová M, Sasorith S, Piron-Prunier F, Suomi F, Zhang D, Martinez-Lopez N, Leboucq N, Bahr A, Azzarello-Burri S, Reich S, Schöls L, Polvikoski TM, Meyer P, Larrieu L, Schaefer AM, Alsaif HS, Alyamani S, Zuchner S, Barbosa IA, Deshpande C, Pyle A, Rauch A, Synofzik M, Alkuraya FS, Rivier F, Ryten M, **McFarland R**, Delahodde A, McWilliams TG, Koenig M, **Taylor RW.** N Engl J Med. 2021 Jun 24;384(25):2406-2417. doi: 10.1056/NEJMoa1915722. PMID: 34161705

139. Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). **Töpf A**, Pyle A, Griffin H, Matalonga L, **Schon K;** Solve-RD SNV-indel working group; Solve-RD DITF-euroNMD; Sickmann A, Schara-Schmidt U, Hentschel A, **Chinnery PF**, Kölbel H, Roos A, **Horvath R**. Eur J Hum Genet. 2021 Sep;29(9):1348-1353. doi: 10.1038/s41431-021-00851-8. Epub 2021 Jun 1. PMID: 34075209

140. Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Dworschak GC, Punetha J, Kalanithy JC, Mingardo E, Erdem HB, Akdemir ZC, Karaca E, Mitani T, Marafi D, Fatih JM, Jhangiani SN, Hunter JV, Dakal TC, Dhabhai B, Dabbagh O, Alsaif HS, Alkuraya FS, Maroofian R, **Houlden H, Efthymiou S, Dominik N,** Salpietro V, Sultan T, Haider S, Bibi F, Thiele H, Hoefele J, Riedhammer KM, Wagner M, Guella I, Demos M, Keren B, Buratti J, Charles P, Nava C, Héron D, Heide S, Valkanas E, Waddell LB, Jones KJ, Oates EC, Cooper ST, MacArthur D, Syrbe S, Ziegler A, Platzer K, Okur V, Chung WK, O'Shea SA, Alcalay R, Fahn S, Mark PR, Guerrini R, Vetro A, Hudson B, Schnur RE, Hoganson GE, Burton JE, McEntagart M, Lindenberg T, Yilmaz Ö, Odermatt B, Pehlivan D, Posey JE, Lupski JR, Reutter H. Genet Med. 2021 Sep;23(9):1715-1725. doi: 10.1038/s41436-021-01196-9. Epub 2021 May 30. PMID: 34054129

141. Machine learning algorithms reveal the secrets of mitochondrial dynamics. Collier JJ, **Taylor RW**. EMBO Mol Med. 2021 Jun 7;13(6):e14316. doi: 10.15252/emmm.202114316. Epub 2021 May 27. PMID: 34043876

142. Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. Pottie L, Adamo CS, Beyens A, Lütke S, Tapaneeyaphan P, De Clercq A, Salmon PL, De Rycke R, Gezdirici A, Gulec EY, Khan N, Urquhart JE, Newman WG, Metcalfe K, **Efthymiou S,** Maroofian R, Anwar N, Maqbool S, Rahman F, Altweijri I, Alsaleh M, Abdullah SM, Al-Owain M, Hashem M, Houlden H, Alkuraya FS, Sips P, Sengle G, Callewaert B. Am J Hum Genet. 2021 Jun 3;108(6):1095-1114. doi: 10.1016/j.ajhg.2021.04.016. Epub 2021 May 14. PMID: 33991472

143. Use of Twitter in Neurology: Boon or Bane? Mishra B, Saini M, Doherty CM, **Pitceathly RDS,** Rajan R, Siddiqi OK, **Ramdharry G,** Asranna A, **Tomaselli PJ,** Kermode AG, Bajwa JA, Garg D, **Vishnu VY.** J Med Internet Res. 2021 May 14;23(5):e25229. doi: 10.2196/25229. PMID: 33988522

144. Two novel bi-allelic KDELR2 missense variants cause osteogenesis imperfecta with neurodevelopmental features. **Efthymiou S,** Herman I, Rahman F, Anwar N, Maroofian R, Yip J, Mitani T, Calame DG, Hunter JV, Sutton VR, Yilmaz Gulec E, Duan R, Fatih JM, Marafi D, Pehlivan D, Jhangiani SN, Gibbs RA, Posey JE; SYNAPS Study Group; Maqbool S, Lupski JR, **Houlden H.** Am J Med Genet A. 2021 Jul;185(7):2241-2249. doi: 10.1002/ajmg.a.62221. Epub 2021 May 8. PMID: 33964184

145. Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltage-gated sodium channel function. Scala M, **Efthymiou S,** Sultan T, De Waele J, Panciroli M, Salpietro V, Maroofian R, Striano P, Van Petegem F, **Houlden H**, Bosmans F. Epilepsia. 2021 Jun;62(6):e82-e87. doi: 10.1111/epi.16913. Epub 2021 Apr 26. PMID: 33901312

146. Muscle fat replacement and modified ragged red fibers in two patients with reversible infantile respiratory chain deficiency. Cotta A, Carvalho E, da-Cunha-Junior A, Navarro MM, Paim JF, Valicek J, Baptista-Junior S, da Silveira EB, Lima MI, Carellos EVM, de-La-Rocque-Ferreira A,

Takata RI, **Horvath R.** Neuromuscul Disord. 2021 Jun;31(6):551-557. doi: 10.1016/j.nmd.2021.02.017. Epub 2021 Feb 21. PMID: 33832841

147. Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. Bourinaris T, Athanasiou A, **Efthymiou S,** Wiethoff S, Salpietro V, **Houlden H.** Eur J Hum Genet. 2021 Jun;29(6):1027-1031. doi: 10.1038/s41431-021-00866-1. Epub 2021 Apr 6. PMID: 33824468

148. A glimpse of the genetics of young-onset Parkinson's disease in Central Asia. Kaiyrzhanov R, Aitkulova A, **Vandrovcova J,** Murphy D, Zharkinbekova N, Shashkin C, Akhmetzhanov V, Kaishibayeva G, Karimova A, Myrzayev Z, Murray M, Khaibullin T, Hardy J, **Houlden H**. Mol Genet Genomic Med. 2021 Jun;9(6):e1671. doi: 10.1002/mgg3.1671. Epub 2021 Apr 5. PMID: 33818904

149. Miyoshi myopathy and limb girdle muscular dystrophy R2 are the same disease. Moore U, Gordish H, Diaz-Manera J, James MK, Mayhew AG, Guglieri M, Fernandez-Torron R, Rufibach LE, Feng J, Blamire AM, Carlier PG, Spuler S, Day JW, Jones KJ, Bharucha-Goebel DX, Salort-Campana E, Pestronk A, Walter MC, Paradas C, Stojkovic T, Mori-Yoshimura M, Bravver E, Pegoraro E, Lowes LP, Mendell JR, Bushby K, **Straub V;** Jain COS Consortium. Neuromuscul Disord. 2021 Apr;31(4):265-280. doi: 10.1016/j.nmd.2021.01.009. Epub 2021 Jan 21. PMID: 33610434

150. Assessing Dysferlinopathy Patients Over Three Years With a New Motor Scale. Jacobs MB, James MK, Lowes LP, Alfano LN, Eagle M, Muni Lofra R, Moore U, Feng J, Rufibach LE, Rose K, Duong T, Bello L, Pedrosa-Hernández I, Holsten S, Sakamoto C, Canal A, Sanchez-Aguilera Práxedes N, Thiele S, Siener C, Vandevelde B, DeWolf B, Maron E, Guglieri M, Hogrel JY, Blamire AM, Carlier PG, Spuler S, Day JW, Jones KJ, Bharucha-Goebel DX, Salort-Campana E, Pestronk A, Walter MC, Paradas C, Stojkovic T, Mori-Yoshimura M, Bravver E, Díaz-Manera J, Pegoraro E, Mendell JR; Jain COS Consortium; Mayhew AG, **Straub V**. Ann Neurol. 2021 May;89(5):967-978. doi: 10.1002/ana.26044. Epub 2021 Feb 26. PMID: 33576057

151. Expanding the Spectrum of AP5Z1-Related Hereditary Spastic Paraplegia (HSP-SPG48): A Multicenter Study on a Rare Disease. Breza M, Hirst J, Chelban V, Banneau G, Tissier L, Kol B, Bourinaris T, Said SA, Péréon Y, Heinzmann A, Debs R, Juntas-Morales R, Martinez VG, Camdessanche JP, Scherer-Gagou C, Zola JM, Athanasiou-Fragkouli A, **Efthymiou S**, Vavougios G, Velonakis G, Stamelou M, Tzartos J, Potagas C, Zambelis T, Mariotti C, Blackstone C, Vandrovcova J, Mavridis T, Kartanou C, Stefanis L, Wood N, Karadima G, LeGuern E, Koutsis G, **Houlden H,** Stevanin G. Mov Disord. 2021 Apr;36(4):1034-1038. doi: 10.1002/mds.28487. Epub 2021 Feb 5. PMID: 33543803

152. Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Kaiyrzhanov R, Wortmann S, Reid T, Dehghani M, Vahidi Mehrjardi MY, Alhaddad B, Wagner M, Deschauer M, Cordts I, Fernandez-Murray JP, Treffer V, Metanat Z, Pitman A, **Houlden H,** Meitinger T, Carroll C, McMaster CR, Maroofian R. Brain. 2021 Apr 12;144(3):e30. doi: 10.1093/brain/awaa442. PMID: 33454747

**2020**

153. Differential Diagnoses of Inclusion Body Myositis. **Vivekanandam V, Bugiardini E**, Merve A, Parton M, **Morrow JM, Hanna MG,** Machado PM. Neurol Clin. 2020 Aug;38(3):697-710. doi: 10.1016/j.ncl.2020.03.014. PMID: 32703477

154. Skeletal Muscle Channelopathies. **Vivekanandam V,** Munot P, **Hanna MG,** Matthews E. Neurol Clin. 2020 Aug;38(3):481-491. doi: 10.1016/j.ncl.2020.04.003. PMID: 32703462

155. Improving genetic diagnostics of skeletal muscle channelopathies. **Vivekanandam V,** Männikkö R, Matthews E**, Hanna MG**. Expert Rev Mol Diagn. 2020 Jul;20(7):725-736. doi: 10.1080/14737159.2020.1782195. Epub 2020 Jul 12. PMID: 32657178

156. Managing pregnancy and anaesthetics in patients with skeletal muscle channelopathies. Raja Rayan DL, **Hanna MG**. Neuromuscul Disord. 2020 Jul;30(7):539-545. doi: 10.1016/j.nmd.2020.05.007. Epub 2020 May 28. PMID: 32622512

157. Concurrent sodium channelopathies and amyotrophic lateral sclerosis supports shared pathogenesis. Franklin JP, Cooper-Knock J, Baheerathan A, Moll T, Männikkö R, Heverin M, Hardiman O, Shaw PJ, **Hanna MG.** Amyotroph Lateral Scler Frontotemporal Degener. 2020 Nov;21(7-8):627-630. doi: 10.1080/21678421.2020.1786128. Epub 2020 Jul 3. PMID: 32619119

158. Primary mitochondrial diseases increase susceptibility to bipolar affective disorder. Colasanti A, **Bugiardini E**, Amawi S, Poole OV, Skorupinska I, Skorupinska M, Germain L, Kozyra D, Holmes S, James N, Woodward CE, Quinlivan R, Young AH, **Hanna MG, Pitceathly RDS**. J Neurol Neurosurg Psychiatry. 2020 Aug;91(8):892-894. doi: 10.1136/jnnp-2020-323632. Epub 2020 Jun 11. PMID: 32527838

159. Sodium channel myotonia may be associated with high-risk brief resolved unexplained events. Cea G, Andreu D, Fletcher E, Ramdas S, Sud R, **Hanna MG,** Matthews E. Wellcome Open Res. 2020 May 12;5:57. doi: 10.12688/wellcomeopenres.15798.2. eCollection 2020. PMID: 32509969

160. Homozygous C-terminal loss-of-function $Na_V1.4$ variant in a patient with congenital myasthenic syndrome. Echaniz-Laguna A, Biancalana V, Nadaj-Pakleza A, Fournier E, Matthews E, **Hanna MG,** Männikkö R. J Neurol Neurosurg Psychiatry. 2020 Aug;91(8):898-900. doi: 10.1136/jnnp-2020-323173. Epub 2020 Jun 2. PMID: 32487525

161. Multisystem mitochondrial disease caused by a rare m.10038G>A mitochondrial $tRNA^{Gly}$ (*MT-TG*) variant. Poole OV, Horga A, Hardy SA, **Bugiardini E,** Woodward CE, Hargreaves IP, Merve A, Quinlivan R, **Taylor RW, Hanna MG, Pitceathly RDS**. Neurol Genet. 2020 Mar 18;6(2):e413. doi: 10.1212/NXG.0000000000000413. eCollection 2020 Apr. PMID: 32337339

162. Chronic pain is common in mitochondrial disease. van den Ameele J, Fuge J, **Pitceathly RDS,** Berry S, McIntyre Z, **Hanna MG,** Lee M, **Chinnery PF**. Neuromuscul Disord. 2020 May;30(5):413-419. doi: 10.1016/j.nmd.2020.02.017. Epub 2020 Feb 29. PMID: 32334903

163. Guidelines on clinical presentation and management of nondystrophic myotonias. Stunnenberg BC, LoRusso S, Arnold WD, Barohn RJ, Cannon SC, Fontaine B, Griggs RC, **Hanna MG**, Matthews E, Meola G, Sansone VA, Trivedi JR, van Engelen BGM, Vicart S, Statland JM. Muscle Nerve. 2020 Oct;62(4):430-444. doi: 10.1002/mus.26887. Epub 2020 May 27. PMID: 32270509

164. Deterioration of muscle force and contractile characteristics are early pathological events in spinal and bulbar muscular atrophy mice. Gray AL, Annan L, Dick JRT, La Spada AR, **Hanna MG,** Greensmith L, Malik B. Dis Model Mech. 2020 May 26;13(5):dmm042424. doi: 10.1242/dmm.042424. PMID: 32152060

165. Muscle and brain sodium channelopathies: genetic causes, clinical phenotypes, and management approaches. Matthews E, Balestrini S, Sisodiya SM, **Hanna MG.** Lancet Child Adolesc Health. 2020 Jul;4(7):536-547. doi: 10.1016/S2352-4642(19)30425-0. Epub 2020 Mar 3. PMID: 32142633

166. In vivo assessment of interictal sarcolemmal membrane properties in hypokalaemic and hyperkalaemic periodic paralysis. Tan SV, Suetterlin K, Männikkö R, Matthews E, **Hanna MG,** Bostock H. Clin Neurophysiol. 2020 Apr;131(4):816-827. doi: 10.1016/j.clinph.2019.12.414. Epub 2020 Jan 27. PMID: 32066100

167. Expanding the molecular and phenotypic spectrum of truncating *MT-ATP6* mutations. **Bugiardini E,** Bottani E, Marchet S, Poole OV, Beninca C, Horga A, Woodward C, Lam A, Hargreaves I, Chalasani A, Valerio A, Lamantea E, Venner K, Holton JL, Zeviani M, Houlden H, Quinlivan R,

Lamperti C, **Hanna MG**, **Pitceathly RDS**. Neurol Genet. 2020 Jan 7;6(1):e381. doi: 10.1212/NXG.0000000000000381. eCollection 2020 Feb. PMID: 32042910

168. Clinical, morphological and genetic characterization of Brody disease: an international study of 40 patients. Molenaar JP, Verhoeven JI, Rodenburg RJ, Kamsteeg EJ, Erasmus CE, Vicart S, Behin A, Bassez G, Magot A, Péréon Y, Brandom BW, Guglielmi V, Vattemi G, Chevessier F, Mathieu J, Franques J, Suetterlin K, **Hanna MG,** Guyant-Marechal L, Snoeck MM, Roberts ME, Kuntzer T, Fernandez-Torron R, Martínez-Arroyo A, Seeger J, Kusters B, Treves S, van Engelen BG, Eymard B, Voermans NC, Sternberg D. Brain. 2020 Feb 1;143(2):452-466. doi: 10.1093/brain/awz410. PMID: 32040565

169. Improving specialised care for neuromuscular patients reduces the frequency of preventable emergency hospital admissions. Scalco RS, Quinlivan RM, Nastasi L, Jaffer F, **Hanna MG.** Neuromuscul Disord. 2020 Feb;30(2):173-179. doi: 10.1016/j.nmd.2019.11.013. Epub 2019 Dec 4. PMID: 32005495

170. Concurrent sodium channelopathies and amyotrophic lateral sclerosis supports shared pathogenesis. Franklin JP, Cooper-Knock J, Baheerathan A, Moll T, Männikkö R, Heverin M, Hardiman O, Shaw PJ**, Hanna MG.** Amyotroph Lateral Scler Frontotemporal Degener. 2020 Nov;21(7-8):627-630. doi: 10.1080/21678421.2020.1786128. Epub 2020 Jul 3. PMID: 32619119

171. Deterioration of muscle force and contractile characteristics are early pathological events in spinal and bulbar muscular atrophy mice. Gray AL, Annan L, Dick JRT, La Spada AR, **Hanna MG,** Greensmith L, Malik B. Dis Model Mech. 2020 May 26;13(5):dmm042424. doi: 10.1242/dmm.042424. PMID: 32152060

172. Visualizing, quantifying, and manipulating mitochondrial DNA in vivo. Prole DL, **Chinnery PF,** Jones NS. J Biol Chem. 2020 Dec 18;295(51):17588-17601. doi: 10.1074/jbc.REV120.015101. PMID: 33454000

173. Expanding the phenotype of PIGS-associated early onset epileptic developmental encephalopathy. **Efthymiou S**, Dutra-Clarke M, Maroofian R, Kaiyrzhanov R, Scala M, Reza Alvi J, Sultan T, Christoforou M, Tuyet Mai Nguyen T, Mankad K, Vona B, Rad A, Striano P, Salpietro V, Guillen Sacoto MJ, Zaki MS, Gleeson JG, Campeau PM, Russell BE, **Houlden H.** Epilepsia. 2021 Feb;62(2):e35-e41. doi: 10.1111/epi.16801. Epub 2021 Jan 7. PMID: 33410539

174. Early-onset phenotype of bi-allelic GRN mutations. Neuray C, Sultan T, Alvi JR, Franca MC, Assmann B, Wagner M, Canafoglia L, Franceschetti S, Rossi G, Santana I, Macario MC, Almeida MR, Kamate M, Parikh S, Elloumi HZ, Murphy D, **Efthymiou S,** Maroofian R, **Houlden H.** Brain. 2021 Mar 3;144(2):e22. doi: 10.1093/brain/awaa414. PMID: 33351065

175. Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. De Nittis P, **Efthymiou S,** Sarre A, Guex N, Chrast J, Putoux A, Sultan T, Raza Alvi J, Ur Rahman Z, Zafar F, Rana N, Rahman F, Anwar N, Maqbool S, Zaki MS, Gleeson JG, Murphy D, Galehdari H, Shariati G, Mazaheri N, Sedaghat A; SYNAPS Study Group; Lesca G, Chatron N, Salpietro V, Christoforou M, **Houlden H,** Simonds WF, Pedrazzini T, Maroofian R, Reymond A. J Med Genet. 2021 Dec;58(12):815-831. doi: 10.1136/jmedgenet-2020-107015. Epub 2020 Nov 10. PMID: 33172956

176. PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Tremblay-Laganière C, Kaiyrzhanov R, Maroofian R, Nguyen TTM, Salayev K, Chilton IT, Chung WK, Madden JA, Phornphutkul C, Agrawal PB, **Houlden H,** Campeau PM. Clin Genet. 2021 Feb;99(2):313-317. doi: 10.1111/cge.13877. Epub 2020 Nov 27. PMID: 33156547

177. Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. Bourinaris T, Smedley D, Cipriani V, Sheikh I, Athanasiou-Fragkouli A, **Chinnery P,** Morris H, Real R, Harrison V, Reid E, Wood N; Genomics England Research Consortium;

**Vandrovcova J, Houlden H,** Tucci A. Eur J Hum Genet. 2020 Dec;28(12):1763-1768. doi: 10.1038/s41431-020-00720-w. Epub 2020 Sep 15. PMID: 32934340

178. A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. Ghosh SG, Scala M, Beetz C, Helman G, Stanley V, Yang X, Breuss MW, Mazaheri N, Selim L, Hadipour F, Pais L, Stutterd CA, Karageorgou V, Begtrup A, Crunk A, Juusola J, Willaert R, Flore LA, Kennelly K, Spencer C, Brown M, Trapane P, Hurst ACE, Lane Rutledge S, Goodloe DH, McDonald MT, Shashi V, Schoch K; Undiagnosed Diseases Network; Tomoum H, Zaitoun R, Hadipour Z, Galehdari H, Pagnamenta AT, Mojarrad M, Sedaghat A, Dias P, Quintas S, Eslahi A, Shariati G, Bauer P, Simons C, **Houlden H,** Issa MY, Zaki MS, Maroofian R, Gleeson JG. Eur J Hum Genet. 2021 Feb;29(2):271-279. doi: 10.1038/s41431-020-00717-5. Epub 2020 Sep 8. PMID: 32901138

179. Humans: the ultimate animal models. **Reilly MM, Rossor AM.** J Neurol Neurosurg Psychiatry. 2020 Nov;91(11):1132-1136. doi: 10.1136/jnnp-2020-323016. Epub 2020 Aug 7. PMID: 32769113

180. Biallelic variants in *ADARB1*, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Maroofian R, Sedmík J, Mazaheri N, Scala M, Zaki MS, Keegan LP, Azizimalamiri R, Issa M, Shariati G, Sedaghat A, Beetz C, Bauer P, Galehdari H, O'Connell MA, **Houlden H**. J Med Genet. 2021 Jul;58(7):495-504. doi: 10.1136/jmedgenet-2020-107048. Epub 2020 Jul 27. PMID: 32719099

181. Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Neuray C, Maroofian R, Scala M, Sultan T, Pai GS, Mojarrad M, Khashab HE, deHoll L, Yue W, Alsaif HS, Zanetti MN, Bello O, Person R, Eslahi A, Khazaei Z, Feizabadi MH, Efthymiou S; SYNaPS Study Group; El-Bassyouni HT, Soliman DR, Tekes S, Ozer L, Baltaci V, Khan S, Beetz C, Amr KS, Salpietro V, Jamshidi Y, Alkuraya FS, **Houlden H.** Brain. 2020 Aug 1;143(8):2388-2397. doi: 10.1093/brain/awaa178. PMID: 32705143

182. Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. Scala M, Chua GL, Chin CF, Alsaif HS, Borovikov A, Riazuddin S, Riazuddin S, Chiara Manzini M, Severino M, Kuk A, Fan H, Jamshidi Y, Toosi MB, Doosti M, Karimiani EG, Salpietro V, Dadali E, Baydakova G, Konovalov F, Lozier E, O'Connor E, Sabr Y, Alfaifi A, Ashrafzadeh F, Striano P, Zara F, Alkuraya FS, **Houlden H**, Maroofian R, Silver DL. Eur J Hum Genet. 2020 Nov;28(11):1509-1519. doi: 10.1038/s41431-020-0669-x. Epub 2020 Jun 22. PMID: 32572202

183. A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Breza M, Bourinaris T, **Efthymiou S,** Maroofian R, Athanasiou-Fragkouli A, Tzartos J, Velonakis G, Karavasilis E, Angelopoulou G, Kasselimis D, Potagas C, Stefanis L, Karadima G, Koutsis G, **Houlden H.** Brain. 2020 Jun 1;143(6):e49. doi: 10.1093/brain/awaa120. PMID: 32428220

184. Prevalence of familial cluster headache: a systematic review and meta-analysis. O'Connor E, Simpson BS, **Houlden H, Vandrovcova J,** Matharu M. J Headache Pain. 2020 Apr 25;21(1):37. doi: 10.1186/s10194-020-01101-w. PMID: 32334514

185. Novel likely disease-causing CLN5 variants identified in Pakistani patients with neuronal ceroid lipofuscinosis. Azad B, **Efthymiou S**, Sultan T, Scala M, Alvi JR, Neuray C, **Dominik N**; SYNaPS Study Group; Gul A, **Houlden H**. J Neurol Sci. 2020 Jul 15;414:116826. doi: 10.1016/j.jns.2020.116826. Epub 2020 Apr 7. PMID: 32302805

186. RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Scala M, Mojarrad M, Riazuddin S, Brigatti KW, Ammous Z, Cohen JS, Hosny H, Usmani MA, Shahzad M, Riazuddin S, Stanley V, Eslahi A, Person RE, Elbendary HM, Comi AM, Poskitt L, Salpietro V, Genomics QS, Rosenfeld JA, Williams KB, Marafi D, Xia F, Biderman Waberski M, Zaki

MS, Gleeson J, Puffenberger E, **Houlden H,** Maroofian R. Brain. 2020 Apr 1;143(4):e31. doi: 10.1093/brain/awaa070. PMID: 32227164

187. Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. Schottlaender LV, Abeti R, Jaunmuktane Z, Macmillan C, Chelban V, O'Callaghan B, McKinley J, Maroofian R, **Efthymiou S**, Athanasiou-Fragkouli A, Forbes R, Soutar MPM, Livingston JH, Kalmar B, Swayne O, Hotton G; SYNAPS Study Group; Pittman A, Mendes de Oliveira JR, de Grandis M, Richard-Loendt A, Launchbury F, Althonayan J, McDonnell G, Carr A, Khan S, Beetz C, Bisgin A, Tug Bozdogan S, Begtrup A, Torti E, Greensmith L, Giunti P, Morrison PJ, Brandner S, Aurrand-Lions M, **Houlden H.** Am J Hum Genet. 2020 Mar 5;106(3):412-421. doi: 10.1016/j.ajhg.2020.02.007. PMID: 32142645

188. Rare novel CYP2U1 and ZFYVE26 variants identified in two Pakistani families with spastic paraplegia. Bibi F, **Efthymiou S**, Bourinaris T, Tariq A, Zafar F, Rana N, Salpietro V, **Houlden H,** Raja GK; SYNaPS Study Group; Saeed S, Minhas NM. J Neurol Sci. 2020 Apr 15;411:116669. doi: 10.1016/j.jns.2020.116669. Epub 2020 Jan 11. PMID: 32006740

189. The genetics of intellectual disability: advancing technology and gene editing. Ilyas M, Mir A, **Efthymiou S, Houlden H**. F1000Res. 2020 Jan 16;9:F1000 Faculty Rev-22. doi: 10.12688/f1000research.16315.1. eCollection 2020. PMID: 31984132

190. Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Perenthaler E, Nikoncuk A, Yousefi S, Berdowski WM, Alsagob M, Capo I, van der Linde HC, van den Berg P, Jacobs EH, Putar D, Ghazvini M, Aronica E, van IJcken WFJ, de Valk WG, Medici-van den Herik E, van Slegtenhorst M, Brick L, Kozenko M, Kohler JN, Bernstein JA, Monaghan KG, Begtrup A, Torene R, Al Futaisi A, Al Murshedi F, Mani R, Al Azri F, Kamsteeg EJ, Mojarrad M, Eslahi A, Khazaei Z, Darmiyan FM, Doosti M, Karimiani EG, Vandrovcova J, Zafar F, Rana N, Kandaswamy KK, Hertecant J, Bauer P, AlMuhaizea MA, Salih MA, Aldosary M, Almass R, Al-Quait L, Qubbaj W, Coskun S, Alahmadi KO, Hamad MHA, Alwadaee S, Awartani K, Dababo AM, Almohanna F, Colak D, Dehghani M, Mehrjardi MYV, Gunel M, Ercan-Sencicek AG, Passi GR, Cheema HA, **Efthymiou S, Houlden H,** Bertoli-Avella AM, Brooks AS, Retterer K, Maroofian R, Kaya N, van Ham TJ, Barakat TS. Acta Neuropathol. 2020 Mar;139(3):415-442. doi: 10.1007/s00401-019-02109-6. Epub 2019 Dec 9. PMID: 31820119

**2019**

191. Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre. **Bugiardini E**, Khan AM, Phadke R, Lynch DS, Cortese A, Feng L, Gang Q, Pittman AM, **Morrow JM**, Turner C, Carr AS, Quinlivan R, Rossor AM, Holton JL, Parton M, Blake JC, **Reilly MM, Houlden H**, Matthews E, **Hanna MG**. Neuromuscul Disord. 2019 Oct;29(10):747-757. doi: 10.1016/j.nmd.2019.08.003. Epub 2019 Aug 19. PMID: 31561939

192. Cell-Free Expression of Sodium Channel Domains for Pharmacology Studies. Noncanonical Spider Toxin Binding Site in the Second Voltage-Sensing Domain of Human $Na_v1.4$ Channel. Myshkin MY, Männikkö R, Krumkacheva OA, Kulbatskii DS, Chugunov AO, Berkut AA, Paramonov AS, Shulepko MA, Fedin MV, **Hanna MG,** Kullmann DM, Bagryanskaya EG, Arseniev AS, Kirpichnikov MP, Lyukmanova EN, Vassilevski AA, Shenkarev ZO. Front Pharmacol. 2019 Sep 4;10:953. doi: 10.3389/fphar.2019.00953. eCollection 2019. PMID: 31555136

193. Possible role of *SCN4A* skeletal muscle mutation in apnea during seizure. Türkdoğan D, Matthews E, Usluer S, Gündoğdu A, Uluç K, Mannikko R, **Hanna MG,** Sisodiya SM, Çağlayan HS. Epilepsia Open. 2019 Jul 1;4(3):498-503. doi: 10.1002/epi4.12347. eCollection 2019 Sep. PMID: 31440732

194. Safety and efficacy of intravenous bimagrumab in inclusion body myositis (RESILIENT): a randomised, double-blind, placebo-controlled phase 2b trial. **Hanna MG**, Badrising UA, Benveniste O, Lloyd TE, Needham M, Chinoy H, Aoki M, Machado PM, Liang C, Reardon KA, de Visser M, Ascherman DP, Barohn RJ, Dimachkie MM, Miller JAL, Kissel JT, Oskarsson B, Joyce NC, Van den Bergh P, Baets J, De Bleecker JL, Karam C, David WS, Mirabella M, Nations SP, Jung HH, Pegoraro E, Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, Yamashita S, Suzuki N, Katsuno M, Murata K, Nodera H, Nishino I, Romano CD, Williams VSL, Vissing J, Auberson LZ, Wu M, de Vera A, Papanicolaou DA, Amato AA; RESILIENT Study Group. Lancet Neurol. 2019 Sep;18(9):834-844. doi: 10.1016/S1474-4422(19)30200-5. PMID: 31397289 Clinical Trial.

195. Skeletal muscle MRI differentiates SBMA and ALS and correlates with disease severity. Klickovic U, Zampedri L, Sinclair CDJ, Wastling SJ, Trimmel K, Howard RS, Malaspina A, Sharma N, Sidle K, Emira A, Shah S, Yousry TA, **Hanna MG**, Greensmith L, **Morrow JM,** Thornton JS, Fratta P. Neurology. 2019 Aug 27;93(9):e895-e907. doi: 10.1212/WNL.0000000000008009. Epub 2019 Aug 7. PMID: 31391248

196. AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Salpietro V, Dixon CL, Guo H, Bello OD, **Vandrovcova J, Efthymiou S,** Maroofian R, Heimer G, Burglen L, Valence S, Torti E, Hacke M, Rankin J, Tariq H, Colin E, Procaccio V, Striano P, Mankad K, Lieb A, Chen S, Pisani L, Bettencourt C, Männikkö R, Manole A, Brusco A, Grosso E, Ferrero GB, Armstrong-Moron J, Gueden S, Bar-Yosef O, Tzadok M, Monaghan KG, Santiago-Sim T, Person RE, Cho MT, Willaert R, Yoo Y, Chae JH, Quan Y, Wu H, Wang T, Bernier RA, Xia K, Blesson A, Jain M, Motazacker MM, Jaeger B, Schneider AL, Boysen K, Muir AM, Myers CT, Gavrilova RH, Gunderson L, Schultz-Rogers L, Klee EW, Dyment D, Osmond M, Parellada M, Llorente C, Gonzalez-Peñas J, Carracedo A, Van Haeringen A, Ruivenkamp C, Nava C, Heron D, Nardello R, Iacomino M, Minetti C, Skabar A, Fabretto A; SYNAPS Study Group; Raspall-Chaure M, Chez M, Tsai A, Fassi E, Shinawi M, Constantino JN, De Zorzi R, Fortuna S, Kok F, Keren B, Bonneau D, Choi M, Benzeev B, Zara F, Mefford HC, Scheffer IE, Clayton-Smith J, Macaya A, Rothman JE, Eichler EE, Kullmann DM, **Houlden H**. Nat Commun. 2019 Jul 12;10(1):3094. doi: 10.1038/s41467-019-10910-w. PMID: 31300657

197. MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. **Bugiardini E,** Mitchell AL, Rosa ID, Horning-Do HT, Pitmann AM, Poole OV, Holton JL, Shah S, Woodward C, Hargreaves I, Quinlivan R, Amunts A, Wiesner RJ, **Houlden H,** Holt IJ, **Hanna MG, Pitceathly RDS**, Spinazzola A. Hum Mol Genet. 2019 Aug 15;28(16):2711-2719. doi: 10.1093/hmg/ddz093. PMID: 31039582

198. Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. Salpietro V, Malintan NT, Llano-Rivas I, Spaeth CG, **Efthymiou S,** Striano P, **Vandrovcova J,** Cutrupi MC, Chimenz R, David E, Di Rosa G, Marce-Grau A, Raspall-Chaure M, Martin-Hernandez E, Zara F, Minetti C; Deciphering Developmental Disorders Study; SYNAPS Study Group; Bello OD, De Zorzi R, Fortuna S, Dauber A, Alkhawaja M, Sultan T, Mankad K, Vitobello A, Thomas Q, Mau-Them FT, Faivre L, Martinez-Azorin F, Prada CE, Macaya A, Kullmann DM, Rothman JE, Krishnakumar SS, **Houlden H**. Am J Hum Genet. 2019 Apr 4;104(4):721-730. doi: 10.1016/j.ajhg.2019.02.016. Epub 2019 Mar 28. PMID: 30929742

199. Community exercise is feasible for neuromuscular diseases and can improve aerobic capacity. Wallace A, Pietrusz A, Dewar E, Dudziec M, Jones K, Hennis P, Sterr A, Baio G, Machado PM, **Laurá M**, **Skorupinska I,** Skorupinska M, Butcher K, Trenell M, **Reilly MM, Hanna MG, Ramdharry GM**. Neurology. 2019 Apr 9;92(15):e1773-e1785. doi: 10.1212/WNL.0000000000007265. Epub 2019 Mar 8. PMID: 30850441 Clinical Trial.

200. Vestibular dysfunction: a frequent problem for adults with mitochondrial disease. Holmes S, Male AJ, **Ramdharry G,** Woodward C, James N, **Skorupinska I**, Skorupinska M, Germain L, Kozyra D, **Bugiardini E**, Poole OV, Quinlivan R, **Hanna MG,** Kaski D, **Pitceathly RDS**. J Neurol Neurosurg Psychiatry. 2019 Jul;90(7):838-841. doi: 10.1136/jnnp-2018-319267. Epub 2018 Nov 26. PMID: 30478136

201. Acetazolamide can improve symptoms and signs in ion channel-related congenital myopathy. Matthews E, Hartley L, Sud R, **Hanna MG, Muntoni F,** Munot P. J Neurol Neurosurg Psychiatry. 2019 Feb;90(2):243-245. doi: 10.1136/jnnp-2017-317849. Epub 2018 May 16. PMID: 29769250

202. Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre. **Bugiardini E**, Khan AM, Phadke R, Lynch DS, Cortese A, Feng L, Gang Q, Pittman AM, **Morrow JM,** Turner C, Carr AS, Quinlivan R, **Rossor AM,** Holton JL, Parton M, Blake JC, **Reilly MM, Houlden H,** Matthews E, **Hanna MG**. Neuromuscul Disord. 2019 Oct;29(10):747-757. doi: 10.1016/j.nmd.2019.08.003. Epub 2019 Aug 19. PMID: 31561939

203. Utility of Whole Blood Thiamine Pyrophosphate Evaluation in *TPK1*-Related Diseases. **Bugiardini E**, Pope S, Feichtinger RG, Poole OV, Pittman AM, Woodward CE, Heales S, Quinlivan R, **Houlden H**, Mayr JA, **Hanna MG, Pitceathly RDS**. J Clin Med. 2019 Jul 8;8(7):991. doi: 10.3390/jcm8070991. PMID: 31288420

204. Pathogenic variants in MT-ATP6: A United Kingdom-based mitochondrial disease cohort study. Ng YS, Martikainen MH, Gorman GS, Blain A, **Bugiardini E**, Bunting A, Schaefer AM, Alston CL, Blakely EL, Sharma S, Hughes I, Lim A, de Goede C, McEntagart M, Spinty S, Horrocks I, Roberts M, Woodward CE, **Chinnery PF, Horvath R,** Nesbitt V, Fratter C, Poulton J, **Hanna MG,** Pitceathly RDS, **Taylor RW, Turnbull DM**, **McFarland R.** Ann Neurol. 2019 Aug;86(2):310-315. doi: 10.1002/ana.25525. Epub 2019 Jul 1. PMID: 31187502

205. Drug repurposing in neurological diseases: an integrated approach to reduce trial and error. Clout AE, Della Pasqua O, **Hanna MG,** Orlu M, **Pitceathly RDS**. J Neurol Neurosurg Psychiatry. 2019 Nov;90(11):1270-1275. doi: 10.1136/jnnp-2019-320879. Epub 2019 Jun 6. PMID: 31171583

206. Focused HLA analysis in Caucasians with myositis identifies significant associations with autoantibody subgroups. Rothwell S, Chinoy H, Lamb JA, Miller FW, Rider LG, Wedderburn LR, McHugh NJ, Mammen AL, Betteridge ZE, Tansley SL, Bowes J, Vencovský J, Deakin CT, Dankó K, Vidya L, Selva-O'Callaghan A, Pachman LM, Reed AM, Molberg Ø, Benveniste O, Mathiesen PR, Radstake TRDJ, Doria A, de Bleecker J, Lee AT, **Hanna MG,** Machado PM, Ollier WE, Gregersen PK, Padyukov L, O'Hanlon TP, Cooper RG, Lundberg IE; Myositis Genetics Consortium (MYOGEN). Ann Rheum Dis. 2019 Jul;78(7):996-1002. doi: 10.1136/annrheumdis-2019-215046. Epub 2019 May 28. PMID: 31138531

207. Autosomal dominant optic atrophy and cataract "plus" phenotype including axonal neuropathy. Horga A, **Bugiardini E**, Manole A, Bremner F, Jaunmuktane Z, Dankwa L, Rebelo AP, Woodward CE, Hargreaves IP, Cortese A, Pittman AM, Brandner S, Polke JM, **Pitceathly RDS**, Züchner S**, Hanna MG,** Scherer SS, **Houlden H, Reilly MM**. Neurol Genet. 2019 Apr 1;5(2):e322. doi: 10.1212/NXG.0000000000000322. eCollection 2019 Apr. PMID: 31119193

208. Investigation of the psychometric properties of the inclusion body myositis functional rating scale with rasch analysis. **Ramdharry G, Morrow J,** Hudgens S, **Skorupinska I**, Gwathmey K, Currence M, Herbelin L, Jawdat O, Pasnoor M, Mcvey A, Barohn RJ, Burns TM, Dimachkie MM, Amato AA, **Hanna MG**, Machado PM. Muscle Nerve. 2019 Aug;60(2):161-168. doi: 10.1002/mus.26521. Epub 2019 Jun 7. PMID: 31107564

209. Development of MRC Centre MRI calf muscle fat fraction protocol as a sensitive outcome measure in Hereditary Sensory Neuropathy Type 1. Kugathasan U, Evans MRB, **Morrow JM,** Sinclair CDJ, Thornton JS, Yousry TA, Hornemann T, Suriyanarayanan S, Owusu-Ansah K, Lauria G, Lombardi R, Polke JM, Wilson E, Bennett DLH, **Houlden H, Hanna MG,** Blake JC**, Laura M, Reilly MM**. J Neurol Neurosurg Psychiatry. 2019 Aug;90(8):895-906. doi: 10.1136/jnnp-2018-320198. Epub 2019 Apr 17. PMID: 30995999

210. Gene expression analysis reveals early dysregulation of disease pathways and links Chmp7 to pathogenesis of spinal and bulbar muscular atrophy. Malik B, Devine H, Patani R, La Spada AR, **Hanna MG**, Greensmith L. Sci Rep. 2019 Mar 5;9(1):3539. doi: 10.1038/s41598-019-40118-3. PMID: 30837566

211. Myasthenic congenital myopathy from recessive mutations at a single residue in $Na_V1.4$. Elia N, Palmio J, Castañeda MS, Shieh PB, Quinonez M, Suominen T, **Hanna MG,** Männikkö R, Udd B, Cannon SC. Neurology. 2019 Mar 26;92(13):e1405-e1415. doi: 10.1212/WNL.0000000000007185. Epub 2019 Mar 1. PMID: 30824560

212. Adult-onset Leigh syndrome linked to the novel stop codon mutation m.6579G>A in MT-CO1. Poole OV, Everett CM, Gandhi S, Marino S, **Bugiardini E,** Woodward C, Lam A, Quinlivan R, **Hanna MG, Pitceathly RDS**. Mitochondrion. 2019 Jul;47:294-297. doi: 10.1016/j.mito.2019.02.004. Epub 2019 Feb 8. PMID: 30743023

213. TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. Vandervore LV, Schot R, Milanese C, Smits DJ, Kasteleijn E, Fry AE, Pilz DT, Brock S, Börklü-Yücel E, Post M, Bahi-Buisson N, Sánchez-Soler MJ, van Slegtenhorst M, Keren B, Afenjar A, Coury SA, Tan WH, Oegema R, de Vries LS, Fawcett KA, Nikkels PGJ, Bertoli-Avella A, Al Hashem A, Alwabel AA, Tlili-Graiess K, **Efthymiou S,** Zafar F, Rana N, Bibi F, **Houlden H,** Maroofian R, Person RE, Crunk A, Savatt JM, Turner L, Doosti M, Karimiani EG, Saadi NW, Akhondian J, Lequin MH, Kayserili H, van der Spek PJ, Jansen AC, Kros JM, Verdijk RM, Milošević NJ, Fornerod M, Mastroberardino PG, Mancini GMS. Am J Hum Genet. 2019 Dec 5;105(6):1126-1147. doi: 10.1016/j.ajhg.2019.10.009. Epub 2019 Nov 14. PMID: 31735293

214. Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Due to *MTMR2* Mutations and Implications in Membrane Trafficking. Wang H, Kaçar Bayram A, Sprute R, Ozdemir O, Cooper E, Pergande M, **Efthymiou S,** Nedic I, Mazaheri N, Stumpfe K, Azizi Malamiri R, Shariati G, Zeighami J, Bayram N, Naghibzadeh SK, Tajik M, Yaşar M, Sami Güven A, Bibi F, Sultan T, Salpietro V, **Houlden H**, Per H, Galehdari H, Shalbafan B, Jamshidi Y, Cirak S. Front Neurosci. 2019 Oct 14;13:974. doi: 10.3389/fnins.2019.00974. eCollection 2019. PMID: 31680794

215. Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. Dias CM, Punetha J, Zheng C, Mazaheri N, Rad A, Efthymiou S, Petersen A, Dehghani M, Pehlivan D, Partlow JN, Posey JE, Salpietro V, Gezdirici A, Malamiri RA, Al Menabawy NM, Selim LA, Vahidi Mehrjardi MY, Banu S, Polla DL, Yang E, Rezazadeh Varaghchi J, Mitani T, van Beusekom E, Najafi M, Sedaghat A, Keller-Ramey J, Durham L, Coban-Akdemir Z, Karaca E, Orlova V, Schaeken LLM, Sherafat A, Jhangiani SN, Stanley V, Shariati G, Galehdari H, Gleeson JG, Walsh CA, Lupski JR, Seiradake E, **Houlden H,** van Bokhoven H, Maroofian R. Am J Hum Genet. 2019 Nov 7;105(5):1048-1056. doi: 10.1016/j.ajhg.2019.09.025. Epub 2019 Oct 24. PMID: 31668703

216. Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. **Efthymiou S,** Salpietro V, Malintan N, Poncelet M, Kriouile Y, Fortuna S, De Zorzi R, Payne K, Henderson LB, Cortese A, Maddirevula S, Alhashmi N, Wiethoff S, Ryten M, Botia JA, Provitera V, Schuelke M, **Vandrovcova J;** SYNAPS Study Group; Walsh L, Torti E, Iodice V, Najafi M, Karimiani EG, Maroofian R, Siquier-Pernet K, Boddaert N, De Lonlay P, Cantagrel V, Aguennouz M,

El Khorassani M, Schmidts M, Alkuraya FS, Edvardson S, Nolano M, Devaux J, **Houlden H**. Brain. 2019 Oct 1;142(10):2948-2964. doi: 10.1093/brain/awz248. PMID: 31501903