MASTER PEDIATRIC ENDOCRINOLOGY AND DIABETES

Matteo Della Monica list of publications

Lonardo F, Lonardo MS, Acquaviva F, **Della Monica M**, Scarano F, Scarano G. Say-Barber-Biesecker-Young-Simpson syndrome and Genitopatellar syndrome: lumping or splitting? Clin Genet. 2017 Aug 30. doi: 10.1111/cge.13127. [ABSTRACT]

Bargiacchi S, **Della Monica M**, Biagiotti R, Andreucci E, Ciabattoni S, Poggi P, Di Maurizio M, Defilippi C, Cariati E, Giglio S. Metatropic dysplasia in third trimester of pregnancy and a novel causative variant in the TRPV4 gene. Eur J Med Genet. 2017 Jul;60(7):365-368. doi: 10.1016/j.ejmg.2017.04.007. [ABSTRACT]

Acquaviva F, Sana ME, **Della Monica M**, Pinelli M, Postorivo D, Fontana P, Falco MT, Nardone AM, Lonardo F, Iascone M, Scarano G. First evidence of Smith-Magenis syndrome in mother and daughter due to a novel RAI mutation. Am J Med Genet A. 2017 Jan;173(1):231-238. doi: 10.1002/ajmg.a.37989. [ABSTRACT] [FULL TEXT] [PDF]

Zollino M, Marangi G, Ponzi E, Orteschi D, Ricciardi S, Lattante S, Murdolo M, Battaglia D, Contaldo I, Mercuri E, Stefanini MC, Caumes R, Edery P, Rossi M, Piccione M, Corsello G, **Della Monica M**, Scarano F, Priolo M, Gentile M, Zampino G, Vijzelaar R, Abdulrahman O, Rauch A, Oneda B, Deardorff MA, Saitta SC, Falk MJ, Dubbs H, Zackai E. Intragenic KANSL1 mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotype-phenotype correlations in a large cohort of patients. J Med Genet. 2015 Dec;52(12):804-14. doi:10.1136/jmedgenet-2015-103184. [ABSTRACT]

Wenger TL, Harr M, Ricciardi S, Bhoj E, Santani A, Adam MP, Barnett SS, Ganetzky R, McDonald-McGinn DM, Battaglia D, Bigoni S, Selicorni A, Sorge G, **Monica MD**, Mari F, Andreucci E, Romano S, Cocchi G, Savasta S, Malbora B, Marangi G, Garavelli L, Zollino M, Zackai EH. "CHARGE-like presentation, craniosynostosis and mild Mowat-Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases" American Journal of Medical Genetics Part A. 164:2557-2566, 2014. Am J Med Genet A. 2015 Jul;167(7):1682-3. doi: 10.1002/ajmg.a.36860. [ABSTRACT]

Negri G, Milani D, Colapietro P, Forzano F, **Della Monica M**, Rusconi D, Consonni L, Caffi LG, Finelli P, Scarano G, Magnani C, Selicorni A, Spena S, Larizza L, Gervasini C. Clinical and molecular characterization of Rubinstein-Taybi syndrome patients carrying distinct novel mutations of the EP300 gene. Clin Genet. 2015 Feb;87(2):148-54. doi: 10.1111/cge.12348. [ABSTRACT]

Ritelli M, Chiarelli N, Dordoni C, Reffo E, Venturini M, Quinzani S, **Monica MD**, Scarano G, Santoro G, Russo MG, Calzavara-Pinton P, Milanesi O, Colombi M. Arterial Tortuosity Syndrome: homozygosity for two novel and one recurrent SLC2A10 missense mutations in three families with severe cardiopulmonary complications in infancy and a literature review. BMC Med Genet. 2014 Nov 6;15:122. doi: 10.1186/s12881-014-0122-5. [ABSTRACT] [FULL TEXT] [PDF]

Wenger TL, Harr M, Ricciardi S, Bhoj E, Santani A, Adam MP, Barnett SS, Ganetzky R, McDonald-McGinn DM, Battaglia D, Bigoni S, Selicorni A, Sorge G, **Monica MD**, Mari F, Andreucci E, Romano S, Cocchi G, Savasta S, Malbora B, Marangi G, Garavelli L, Zollino M, Zackai EH. CHARGE-like presentation, craniosynostosis and mild Mowat-Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. Am J Med Genet A. 2014 Oct;164A(10):2557-66. doi: 10.1002/ajmg.a.36696. [ABSTRACT]

Micale L, Augello B, Maffeo C, Selicorni A, Zucchetti F, Fusco C, De Nittis P, Pellico MT, Mandriani B, Fischetto R, Boccone L, Silengo M, Biamino E, Perria C, Sotgiu S, Serra G, Lapi E, Neri M, Ferlini A, Cavaliere ML, Chiurazzi P, **Monica MD**, Scarano G, Faravelli F, Ferrari P, Mazzanti L, Pilotta A, Patricelli MG, Bedeschi MF, Benedicenti F, Prontera P, Toschi B, Salviati L, Melis D, Di Battista E, Vancini A, Garavelli L, Zelante L, Merla G. Molecular analysis, pathogenic mechanisms, and readthrough therapy on a large cohort of Kabuki syndrome patients. Hum Mutat. 2014 Jul;35(7):841-50. doi: 10.1002/humu.22547. [ABSTRACT] [FULL TEXT] [PDF]

Lionel AC, Tammimies K, Vaags AK, Rosenfeld JA, Ahn JW, Merico D, Noor A, Runke CK, Pillalamarri VK, Carter MT, Gazzellone MJ, Thiruvahindrapuram B, Fagerberg C, Laulund LW, Pellecchia G, Lamoureux S, Deshpande C, Clayton-Smith J, White AC, Leather S, Trounce J, Melanie Bedford H, Hatchwell E, Eis PS, Yuen RK, Walker S, Uddin M, Geraghty MT, Nikkel SM, Tomiak EM, Fernandez BA, Soreni N, Crosbie J, Arnold PD, Schachar RJ, Roberts W, Paterson AD, So J, Szatmari P, Chrysler C, Woodbury-Smith M, Brian Lowry R, Zwaigenbaum L, Mandyam D, Wei J, Macdonald JR, Howe JL, Nalpathamkalam T, Wang Z, Tolson D, Cobb DS, Wilks TM, Sorensen MJ, Bader PI, An Y, Wu BL, Musumeci SA, Romano C, Postorivo D, Nardone AM, **Monica MD**, Scarano G, Zoccante L, Novara F, Zuffardi O, Ciccone R, Antona V, Carella M, Zelante L, Cavalli P, Poggiani C, Cavallari U, Argiropoulos B, Chernos J, Brasch-Andersen C, Speevak M, Fichera M, Ogilvie CM, Shen Y, Hodge JC, Talkowski ME, Stavropoulos DJ, Marshall CR, Scherer SW. Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Hum Mol Genet. 2014 May 15;23(10):2752-68. doi: 10.1093/hmg/ddt669. [ABSTRACT] [FULL TEXT] [PDF]

Della Monica M, Galzerano D, Di Michele S, Acquaviva F, Gregorio G, Lonardo F, Sguazzo F, Scarano F, Lama D, Scarano G. Science, art, and mistery in the statues and in the anatomical machines of the prince of sansevero: the masterpieces of the "Sansevero Chapel". Am J Med Genet A. 2013 Nov;161A(11):2920-9. doi:10.1002/ajmg.a.36258. [ABSTRACT] [FULL TEXT] [PDF]

Bachetti T, Di Duca M, **Della Monica M**, Grappone L, Scarano G, Ceccherini I. Recurrence of CCHS associated PHOX2B poly-alanine expansion mutation due to maternal mosaicism. Pediatr Pulmonol. 2014 Mar;49(3):E45-7. doi:10.1002/ppul.22790. [ABSTRACT]

Della Monica M, Mauri R, Scarano F, Lonardo F, Scarano G. Genetic Drift: the Salernitan school of medicine: women, men, and children. A syndromological review of the oldest medical school in the western world. Am J Med Genet A. 2013 Apr;161A(4):809-16. doi: 10.1002/ajmg.a.35742. [ABSTRACT] [FULL TEXT] [PDF]

Travaglini L, Brancati F, Silhavy J, Iannicelli M, Nickerson E, Elkhartoufi N, Scott E, Spencer E, Gabriel S, Thomas S, Ben-Zeev B, Bertini E, Boltshauser E, Chaouch M, Cilio MR, de Jong MM, Kayserili H, Ogur G, Poretti A, Signorini S, Uziel G, Zaki MS; International JSRD Study Group, Johnson C, Attié-Bitach T, Gleeson JG, Valente EM. Phenotypic spectrum and prevalence of INPP5E mutations in Joubert syndrome and related disorders. Eur J Hum Genet. 2013 Oct;21(10):1074-8. doi: 10.1038/ejhg.2012.305. [ABSTRACT] [FULL TEXT] [PDF]

Magini P, **Della Monica M**, Uzielli ML, Mongelli P, Scarselli G, Gambineri E, Scarano G, Seri M. Two novel patients with Bohring-Opitz syndrome caused by de novo ASXL1 mutations. Am J Med Genet A. 2012 Apr;158A(4):917-21. doi:10.1002/ajmg.a.35265. [ABSTRACT]

Marangi G, Ricciardi S, Orteschi D, Tenconi R, **Monica MD**, Scarano G, Battaglia D, Lettori D, Vasco G, Zollino M. Proposal of a clinical score for the molecular test for Pitt-Hopkins syndrome. Am J Med Genet A. 2012 Jul;158A(7):1604-11. doi: 10.1002/ajmg.a.35419. [ABSTRACT]

Micale L, Augello B, Fusco C, Selicorni A, Loviglio MN, Silengo MC, Reymond A, Gumiero B, Zucchetti F, D'Addetta EV, Belligni E, Calcagnì A, Digilio MC, Dallapiccola B, Faravelli F, Forzano F, Accadia M, Bonfante A, Clementi M, Daolio C, Douzgou S, Ferrari P, Fischetto R, Garavelli L, Lapi E, Mattina T, Melis D, Patricelli MG, Priolo M, Prontera P, Renieri A, Mencarelli MA, Scarano G, **della Monica M**, Toschi B, Turolla L, Vancini A, Zatterale A, Gabrielli O, Zelante L, Merla G. Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients. Orphanet J Rare Dis. 2011 Jun 9;6:38. doi: 10.1186/1750-1172-6-38. [ABSTRACT] [FULL TEXT] [PDF]

Lonardo F, Perone L, Maioli M, Ciavarella M, Ciccone R, **Monica MD**, Lombardi C, Forino L, Cantalupo G, Masella L, Scarano F. Clinical, cytogenetic and molecular-cytogenetic characterization of a patient with a de novo tandem proximal-intermediate duplication of 16q and review of the literature. Am J Med Genet A. 2011 Apr;155A(4):769-77. doi: 10.1002/ajmg.a.33852. [ABSTRACT]

Piane M, **Della Monica M**, Piatelli G, Lulli P, Lonardo F, Chessa L, Scarano G. Majewski osteodysplastic primordial dwarfism type II (MOPD II) syndrome previously diagnosed as Seckel syndrome: report of a novel mutation of the PCNT gene. Am J Med Genet A. 2009 Nov;149A(11):2452-6. doi: 10.1002/ajmg.a.33035. [ABSTRACT]

Lonardo F, Cantalupo G, Ciavarella M, **Monica MD**, Lombardi C, Maioli M, Masella L, Nazzaro A, Scarano G. Prenatal diagnosis of 46,XX testicular DSD. Molecular, cytogenetic, molecular-cytogenetic, and ultrasonographic evaluation. Prenat Diagn. 2009 Oct;29(10):998-1001. doi: 10.1002/pd.2329. [ABSTRACT]

Rendina D, De Filippo G, Viceconti R, Soscia E, Sirignano C, Salvatore M, **Della Monica M**, Scarano G, Mossetti G. Interleukin (IL)-6 and receptor activator of nuclear factor (NF)-kappaB ligand (RANKL) are increased in the serum of a patient with primary pachydermoperiostosis. Scand J Rheumatol. 2008 May-Jun;37(3):225-9. doi: 10.1080/03009740701772457. [ABSTRACT]

Della Monica M, Lonardo F, Faravelli F, Pierluigi M, Luquetti DV, De Gregori M, Zuffardi O, Scarano G. A case of autism with an interstitial 1q deletion (1q23.3-24.2) and a de novo translocation of chromosomes 1q and 5q. Am J Med Genet A. 2007 Nov 15;143A(22):2733-7. [ABSTRACT]

Lonardo F, Parenti G, Luquetti DV, Annunziata I, **Della Monica M**, Perone L, De Gregori M, Zuffardi O, Brunetti-Pierri N, Andria G, Scarano G. Contiguous gene syndrome due to an interstitial deletion in Xp22.3 in a boy with ichthyosis, chondrodysplasia punctata, mental retardation and ADHD. Eur J Med Genet. 2007 Jul-Aug;50(4):301-8. [ABSTRACT]

Lonardo F, Sabba G, Luquetti DV, **Monica MD**, Scarano G. Al-Awadi/Raas-Rothschild syndrome: two new cases and review. Am J Med Genet A. 2007 Dec 15;143A(24):3169-74. [ABSTRACT]

Della Monica M, Nazzaro A, Lonardo F, Ferrara G, Di Blasi A, Scarano G. Prenatal ultrasound diagnosis of cloacal exstrophy associated with myelocystocele complex by the 'elephant trunk-like' image and review of the literature. Prenat Diagn. 2005 May;25(5):394-7. [ABSTRACT]

Cecconi M, Forzano F, Milani D, Cavani S, Baldo C, Selicorni A, Pantaleoni C, Silengo M, Ferrero GB, Scarano G, **Della Monica M**, Fischetto R, Grammatico P, Majore S, Zampino G, Memo L, Cordisco EL, Neri G, Pierluigi M, Bricarelli FD, Grasso M, Faravelli F. Mutation analysis of the NSD1 gene in a group of 59 patients with congenital overgrowth. Am J Med Genet A. 2005 Apr 30;134(3):247-53. [ABSTRACT]

Nazzaro A, **Della Monica M**, Lonardo F, Di Blasi A, Baffico M, Baldi M, Nazzaro G, De Placido G, Scarano G. Prenatal ultrasound diagnosis of a case of Pfeiffer syndrome without cloverleaf skull and review of the literature. Prenat Diagn. 2004 Nov;24(11):918-22. [ABSTRACT]

Gurrieri F, Scarano G, Garavelli L, **Della Monica M**, Lonardo F, Cuda D, Banchini G, Opitz JM, Neri G. Mental retardation, Robin sequence, and brachydactyly: further confirmation of a new syndrome. Am J Med Genet A. 2004 Apr 15;126A(2):204-7. [ABSTRACT]

Lonardo F, **Della Monica M**, Riccardi G, Riccio I, Riccio V, Scarano G. A family with X-linked recessive fusion of metacarpals IV and V. Am J Med Genet A. 2004 Feb 1;124A(4):407-10. [ABSTRACT]

Giardino D, Finelli P, Gottardi G, De Canal G, **Della Monica M**, Lonardo F, Scarano G, Larizza L. Narrowing the candidate region of Albright hereditary osteodystrophy-like syndrome by deletion mapping in a patient with an unbalanced cryptic translocation t(2;6)(q37.3;q26). Am J Med Genet A. 2003 Oct 15;122A(3):261-5. [ABSTRACT]

Genuardi M, Tozzi C, Pomponi MG, Stagni ML, **Della Monica M**, Scarano G, Calvieri F, Torrisi L, Neri G. Mosaic trisomy 17 in amniocytes: phenotypic outcome, tissue distribution, and uniparental disomy studies. Eur J Hum Genet. 1999 May-Jun;7(4):421-6. [ABSTRACT] [FULL TEXT] [PDF]

Grifa A, Wagner CA, D'Ambrosio L, Melchionda S, Bernardi F, Lopez-Bigas N, Rabionet R, Arbones M, **Monica MD**, Estivill X, Zelante L, Lang F, Gasparini P. Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. Nat Genet. 1999 Sep;23(1):16-8. [ABSTRACT]

Gasparini P, Estivill X, Volpini V, Totaro A, Castellvi-Bel S, Govea N, Mila M, **Della Monica M**, Ventruto V, De Benedetto M, Stanziale P, Zelante L, Mansfield ES, Sandkuijl L, Surrey S, Fortina P. Linkage of DFNB1 to non-syndromic neurosensory autosomal-recessive deafness in Mediterranean families. Eur J Hum Genet. 1997 Mar-Apr;5(2):83-8. [ABSTRACT]

Franceschini P, Guala A, Vardeu MP, Signorile F, Franceschini D, Mastroiacovo P, Gianotti A, Livini E, Lalatta F, Selicorni A, Andria G, Scarano G, **Della Monica M**, Rizzo R, Zelante L, Stabile M, Gabrielli O, Neri G. The Williams syndrome: an Italian collaborative study. Minerva Pediatr. 1996 Oct;48(10):421-8. [ABSTRACT]

Scarano G, **Della Monica M**, Lonardo F, Neri G. Novel findings in a patient with Weaver or a Weaver-like syndrome. Am J Med Genet. 1996 May 17;63(2):378-81. [ABSTRACT]

Scarano G, **Della Monica M**, Capece G, Lonardo F, Neri G, Maroteaux P. A case of short-rib syndrome without polydactyly in a stillborn: a new type? Birth Defects Orig Artic Ser. 1996;30(1):95-101. [ABSTRACT]

Genuardi M, Scarano G, Tozzi C, Chinca M, **Della Monica M**, Martini-Neri ME, Picardi P, Neri G. Malformation syndromes with kidney dysplasia. Birth Defects Orig Artic Ser. 1996;30(1):379-95. [ABSTRACT]

Capece G, Fasolino A, **Della Monica M**, Lonardo F, Scarano G, Neri G. Prenatal diagnosis of femur-fibula-ulna complex by ultrasonography in a male fetus at 24 weeks of gestation. Prenat Diagn. 1994 Jun;14(6):502-5. [ABSTRACT]

Stabile M, Ametrano O, Della Bruna M, **Della Monica M**, Lonardo F, Strisciuglio P. Report of three cases with tricho-rhino-phalangeal syndrome type I (two cases) and type II (one case). Australas Radiol. 1988 Aug;32(3):338-42. [ABSTRACT]

Della Bruna M, Mastro AA, Lonardo F, Ametrano O, **Della Monica M**, D'Avanzo MG, Bianco A, Ventruto V. Two sibs affected by Pendred's syndrome in a family with recurrent goiter. Minerva Endocrinol. 1987 Jan-Mar;12(1):29-32. [ABSTRACT]

Ventruto V, Stabile M, Gentile G, de Tollis G, Colantuoni M, Sirone P, Perone L, Lonardo F, **Della Monica M**, Fiore M. Expression of the autosomal folate-sensitive fragile sites in ten kindreds with Martin-Bell syndrome. Ann Genet. 1986;29(1):59-61. [ABSTRACT]