

Matteo DELLA MONICA

PUBBLICAZIONI

- 1 "Il tempo di protrombina nel monitoraggio della terapia anticoagulante", estratto da "Tempo Medico", suppl. al n.5, maggio 1985.
- 2 "Expression of the autosomal folate-sensitive fragile sites in ten kindreds with Martin-Bell syndrome", Annales De Genetique, 29, n.1, 59-61, 1986.
- 3 "Incidenza di riarrangiamenti cromosomici nelle coppie con aborti spontanei (studio di 148 coppie:1980-1984)" Giornale It.di Ost. e Gin., Vol VII n.11-Nov .1986
- 4 "Sindrome di Down in Campania: 11 anni di osservazioni (1975-1986)", Atti del II Corso di aggiornamento per i Consultori Familiari, Aprile-Dicembre 1986.
- 5 "Auxological evaluation of a case of trisomy 4p from paternal balanced translocation.Considerations on the preferential rearrangement of the 4p arm with the acrocentric groups and chromosome n.12", Acta Medica Auxologica, Vol.18, n. 3, pg. 169-178, 1986.
- 6 "Two sibs affected by Pendred's syndrome in a family with recurrent goiter", estratto da Minerva Endocrinologica, Vol.12, N., pg.29-32, Gennaio-Marzo 1987.
- 7 "Report of three cases with Tricho-Rhino-Phalangeal Syndrome Type I (Two Cases) and Type II (One Case)", Australas Radiol,32,:338-342,1988.
- 8 "Malattie genetiche ed infezioni trasmissibili: valore decisionale dei dati di laboratorio", Patologia Clinica, Vol.III N.5, Ottobre, pg.325-331,1988.
- 9 "Studio di alcune malattie diagnistiche di rilevanza sociale:il ritardo mentale, la distrofia muscolare di Duchenne, le ittiosi e le cecità ereditarie.Obiettività in Irpinia, prospettive di diagnosi e prevenzione" (Gennaio 88-luglio 90) Atti del Programma di Ricerca
- 10 "Diagnosi prenatale e screening per la sindrome di Down", Notiziario dell'Associazione della Sindrome di Down, pg.18-20, Gennaio 1991.
- 11 "Sindrome cardio-facio-cutanea (CFC): un nuovo caso italiano", estratto da Rivista Italiana di Pediatria, Vol.17, N.2, pg220-222, Aprile 1991.
- 12 "Suscettibilità alle infezioni della persona Down" Notiziario dell'Associazione della Sindrome di Down, 1993 pg.27- 30.
- 13 "Sindrome di Tel Hashomer", Riv Ital Pediatr,1994; 20:572-575.
- 14 "Prenatal diagnosis of Femur-Fibula-Ulna Complex by ultrasonography in a male fetus at 24 weeks of gestation", Prenatal Diagnosis vol.14:502-505 ,1994.
- 15 "La sindrome di Smith-Magenis:rara o sottodiagnosticata? Presentazione di un caso diagnosticato all'età di due mesi". Riv.Ital.Pediatr (IJP)1995,21:731-735

- 16 "Novel Findings in a Patient With Weaver or a Weaver-Like Syndrome", Am J of Med Genet 63:378-381 (1996)
- 17 "The Williams syndrome: a Italian collaborative study", estratto da Minerva Pediatrica, vol.48.pg421-28 (ottobre 1996).
- 18 "A case of Short Rib Syndrome Without Polydactyly ia a Stillborn:A New Type?", Birth Defects:Original Article series, volume 30,Number 1, pages 95-101, 1996 March of dimes Birth Defects Foundation
- 19 "Malformation Syndromes With Kidney Dysplasia", Birth Defects:Original Article series, volume 30,Number 1, pages 379-395, 1996 March of dimes Birth Defects Foundation
- 20 "Sindrome di Dubowitz", Linee Guida Assistenziali Nel Bambino con Patologia Malformativa e Metabolica, vol.III (1997)
- 21 "Sindrome di Marfan", Linee Guida Assistenziali Nel Bambino con Patologia Malformativa e Metabolica, vol.III (1997)
- 22 "Sindrome di Stickler (Arto-Oftalmopatia progressiva ereditaria)", Linee Guida Assistenziali Nel Bambino con Patologia Malformativa e Metabolica, vol.III (1997)
- 23 "Sindrome di Dubowitz:difficoltà diagnostiche", Riv Ital Pediatr(IJP)1997;23:441-444.
- 24 "Linkage of DFNB1 to Non-Syndromic Neurosensory Autosomal-Recessive Deafness in Mediterranean Families", Eur J Hum Genet 1997;5:83-88
- 25 "Connexin26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans", Human Molecular Genetics,1997,vol.6,No.9,1605-1609.
- 26 "Mosaic trisomy 17 in amniocytes: phenotypic outcome, tissue distribution, and uniparental disomy studies". Eur J Hum Genet. 1999 May-Jun;7(4):421-6.
- 27 "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;122(3):261-5.
- 28 " A family with X-linked recessive fusion of metacarpals IV and V". Am J Med Genet A. 2004 Feb 1;124(4):407-10.
- 29 "Mental retardation, Robin sequence, and brachydactyly: further confirmation of a new syndrome. Am J Med Genet A. 2004 Apr 15;126(2):204-7.
- 30 "Ocular proptosis, brachycephaly, broad thumb and big toe at birth. Neonata con proptosi oculare, brachicefalia, alluce largo e pollice grande alla nascita" Ital J Pediatr 2004;30:137-139
- 31 "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. Review.

- 32 "Congenital scoliosis associated with agenesis of the uterine cervix. Case report". BMC Womens Health. 2004 Jun 30;4(1):4.
- 33 "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.
- 34 "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.
- 35 "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
- 36 "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;143(22):2733-7.
- 37 "Al-Awadi/Raas-Rothschild syndrome: two new cases and review". **Am J Med Genet A. 2007 Dec 15;143A(24):3169-74.**
- 38 "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.
- 39 "Prenatal diagnosis of 46,XX testicular DSD. Molecular, cytogenetic, molecular-cytogenetic, and ultrasonographic evaluation" Prenat Diagn. 2009 Oct;29(10):998-1001.
- 40 "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.
- 41 "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.
- 42 "Mutation spectrum of MLL2 in a cohort of Kabuki syndrome patients". Orphanet J Rare Dis. 2011 Jun 9;6:38.
- 43 "Two novel patients with Bohring-Opitz syndrome caused by de novo ASXL1 mutations". Am J Med Genet A. 2012 Apr;158A(4):917-21.
- 44 "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. Epub 2012 Jun 7. PMID:22678594 [PubMed - indexed for MEDLINE]
- 45 "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;161(4):809-16.

46. "Science, art, and mystery 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;161(11):2920-9.
47. "Clinical and molecular characterization of Rubinstein-Taybi syndrome patients carrying distinct novel mutations of the EP300 gene". Clin Genet. 2014 Jan 29. doi: 10.1111/cge.12348. [Epub ahead of print] PMID:24476420[PubMed - as supplied by publisher]
- 48 "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. Epub 2013 Mar 4.PMID:23460545 [PubMed - in process]
- 49 "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 Jan 14. [Epub ahead of print] PMID:24381304 [PubMed - as supplied by publisher]
- 50 "Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of Kabuki Syndrome Patients." Hum Mutat. 2014 Mar 13. doi: 10.1002/humu.22547. [Epub ahead of print] PMID:24633898 [PubMed - as supplied by publisher]
- 51 "Clinical and molecular characterization of Rubinstein-Taybi syndrome patients carrying distinct novel mutations of the EP300 gene". Clin Genet. 2014 Jan 29. doi: 10.1111/cge.12348. [Epub ahead of print]
- 52 "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

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Ha curato i seguenti capitoli nei testi riportati:

1. Le sindromi genetiche associate a sordità. Adriana de Filippis. L'impianto cocleare. Manuale Operativo. Masson, Milano, 2002.
2. Il bambino con sindrome malformativa nella storia. Il neonato in Europa tra storia e attualità. Choes, Usmate (MI), 2007.
3. Il neonato malformato nel corso della storia in "Figli della Dea madre. Storia dei neonati nel Mediterraneo" a cura di Vassilios Fanos e Murat Yurdakok.Hygeia Press, maggio 2011.