

## Elena ANDREUCCI

### PUBBLICAZIONI

1- Ricci U, Sani I, Guarducci S, Biondi C, Pelagatti S, Lazzerini V, Brusaferrri A, Lapini M, Andreucci E, Giunti L, Giovannucci Uzielli ML. "Infrared Fluorescent automated detection of thirteen short tandem repeat polymorphisms and one gender-determining system of the CODIS core system". Electrophoresis 2000; 21: 3564-70

2- Giunti L, Pelagatti S, Lazzerini V, Guarducci S, Lapi E, Coviello S, Cecconi A, Ombroni L, Andreucci E, Sani I, Brusaferrri A, Lasagni A, Ricotti G, Giometto B, Nicolao P, Gasparini P, Granatiero M, Giovannucci Uzielli ML. "Spectrum and distribution of MECP2 Mutations in 64 Italian Rett syndrome girls: tentative genotype/phenotype correlation". Brain and Development 2001; 23: S242-5

3- Savino M, d'Apolito M, Formica V, Baorda F, Mari F, Renieri A, Carabba E, Tarantino E, Andreucci E, Belli S, Lo Muzio L, Dallapiccola B, Zelante L, Savoia A. "Spectrum of PTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: identification of thirteen novel alleles" Human Mutation 2004Nov;24(5):441

4- Giunti L, Cetica V, Ricci U, Giglio S, Sardi I, Paglierani M, Andreucci E, Sanzo M, Forni M, Buccoliero AM, Genitori L, Genuardi M. "Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome." European Journal of Human Genetics 2009Jul;17(7):919-27

5- Carboni I, Andreucci E, Caruso MR, Ciccone R, Zuffardi O, Genuardi M, Pela I, Giglio S. "Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H<sup>+</sup>-ATPase genes." Nephrology Dialysis Transplantation 2009Sep;24(9):2734-8

6- Andreucci E, Bianchi B, Carboni I, Lavoratti G, Mortilla M, Fonda C, Bigozzi M, Genuardi M, Giglio S, Pela I. "Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity". Pediatric Nephrology 2009Nov;24(11):2147-53

7- Iannicelli M, Brancati F, Mougou-Zerelli S, Mazzotta A, Thomas S, Elkhartoufi N, Travaglini L, Gomes C, Ardissino GL, Bertini E, Boltshauser E, Castorina P, D'Arrigo S, Fischetto R, Leroy B, Loget P, Bonnière M, Starck L, Tantau J, Gentilin B, Majore S, Swistun D, Flori E, Lalatta F, Pantaleoni C, Penzien J, Grammatico P, International JSRD Study Group, Dallapiccola B, Gleeson JG, Attie-Bitach T, Valente EM. " Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies". Hum Mutat.2010May;31(5):E1319-31

8- Bedeschi MF, Bianchi V, Gentilin B, Colombo L, Natacci F, Giglio S, Andreucci E, Trespidi L, Acaia B, Superti Furga A, Lalatta F. "Prenatal manifestation and management of a mother and child

affected by spondyloperipheral dysplasia with a C-propeptide mutation in COL2A1: case report". Orphanet J Rare Dis. 2011 Feb 28;6(1):7. [Epub ahead of print]

9- Bowen ME, Boyden ED, Holm IA, Campos-Xavier B, Bonafé L, Furga AS, Ikegawa S, Cormier-Daire V, Bovée JV, Pansuriya TC, De Sousa SB, Savarirayan R, Andreucci E, Vikkula M, Garavelli L, Pottinger C, Ogino T, Sakai A, Regazzoni BM, Wuyts W, Sangiorgi L, Pedrini E, Zhu M, Kozakewich HP, Kasser JR, Seidman JG, Kurek KC, Warman ML. "Loss-of-function mutations in PTPN11 cause Metachondromatosis, but not Ollier Disease or Maffucci Syndrome". PLoS Genet 2011; 7(4): E1002050

10- Andreucci E, Aftimos S, Alcausin M, Haan E, Hunter W, Kannu P, Kerr B, McGillivray G, Gardner RM, Patricelli MG, Sillence D, Thompson E, Zacharin M, Zankl A, Lamandé SR, Savarirayan R. "TRPV4 related skeletal dysplasias: a phenotypic spectrum highlighted by clinical, radiographic, and molecular studies in 21 new families". Orphanet J Rare Dis. 2011 Jun 9;6:37

11- Vultaggio A, Matucci A, D'Elios MM, Andreucci E, Giglio S, Annunziato F, Zupo S, Maggi E. "Multiorgan Infiltration by CD8+ T Cells and 1p;16p Translocation in a Patient with Hypogammaglobulinemia and a Reduced Number of B Cells ". Int Arch Allergy Immunol. 2012 Jan 26;158(2):206-210

12- Piccione M, Serra G, Sanfilippo C, Andreucci E, Sani I, Corsello G. "A new mutation in EDA gene in X-linked hypohidrotic ectodermal dysplasia associated with keratoconus." Minerva Pediatr. 2012 Feb;64(1):59-64.

13- Garavelli L, Gargano G, Simonte G, Rosato S, Wischmeijer A, Melli N, Braibanti S, Gelmini C, Forzano F, Pietrobono R, Pomponi MG, Andreucci E, Toutain A, Superti-Furga A, Neri G. "Simpson-Golabi-Behmel syndrome type 1 in a 27-week macrosomic preterm newborn: The diagnostic value of rib malformations and index nail and finger hypoplasia." Am J Med Genet A. 2012 Sep;158A(9):2245-9

14- 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.

15- Artuso R, Provenzano A, Mazzinghi B, Giunti L, Palazzo V, Andreucci E, Blasetti A, Chiuri RM, Gianiorio FE, Mandich P, Monami M, Mannucci E, Giglio S. "Therapeutic implications of novel mutations of the RFX6 gene associated with early-onset diabetes." Pharmacogenomics J. 2015 Feb;15(1):49-54.

16- Straniero L, Rimoldi V, Soldà G, Mauri L, Manfredini E, Andreucci E, Bargiacchi S, Penco S, Gesu GP, Del Longo A, Piozzi E, Asselta R, Primignani P. "Two novel splicing mutations in the SLC45A2 gene cause Oculocutaneous Albinism Type IV by unmasking cryptic splice sites." J Hum Genet. 2015 May 28 [Epub ahead of print]

17- 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):1682-1683