

CICCONE ROBERTO – PUBLICATIONS

- 1: Della Mina E, Borghesi A, Zhou H, Bougarn S, Boughorbel S, Israel L, Meloni I, Chrabieh M, Ling Y, Itan Y, Renieri A, Mazzucchelli I, Basso S, Pavone P, Falsaperla R, Ciccone R, Cerbo RM, Stronati M, Picard C, Zuffardi O, Abel L, Chaussabel D, Marr N, Li X, Casanova JL, Puel A. Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. *Proc Natl Acad Sci U S A*. 2017 Jan 24;114(4):E514-E523.

- 2: Vetro A, Iascone M, Limongelli I, Ameziane N, Gana S, Della Mina E, Giussani U, Ciccone R, Forlino A, Pezzoli L, Rooimans MA, van Essen AJ, Messa J, Rizzuti T, Bianchi P, Dorsman J, de Winter JP, Lalatta F, Zuffardi O. Loss-of-Function FANCL Mutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. *Hum Mutat*. 2015 May;36(5):562-8.

- 3: Bersano A, Zuffardi O, Pantoni L, Quaglini S, Ciccone R, Vetro A, Persico A, Denaro MF, Micieli G; SVE-LA project collaborators. Next generation sequencing for systematic assessment of genetics of small-vessel disease and lacunar stroke. *J Stroke Cerebrovasc Dis*. 2015 Apr;24(4):759-65.

- 4: Decio A, Tonduti D, Pichiecchio A, Vetro A, Ciccone R, Limongelli I, Giorda R, Caffi L, Balottin U, Zuffardi O, Orcesi S. A novel mutation in COL4A1 gene: a possible cause of early postnatal cerebrovascular events. *Am J Med Genet A*. 2015 Apr;167A(4):810-5.

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- 8: Novara F, Simonati A, Sicca F, Battini R, Fiori S, Contaldo A, Criscuolo L, Zuffardi O, Ciccone R. MECP2 duplication phenotype in symptomatic females: report of three further cases. *Mol Cytogenet*. 2014 Jan 28;7(1):10.

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- 36: Rossi E, Verri AP, Patricelli MG, Destefani V, Ricca I, Vetro A, Ciccone R, Giorda R, Toniolo D, Maraschio P, Zuffardi O. A 12Mb deletion at 7q33-q35 associated with autism spectrum disorders and primary amenorrhea. *Eur J Med Genet*. 2008 Nov-Dec;51(6):631-8.
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