

## PERSONAL INFORMATION

EMANUELA SALZANO

📍 Azienda Ospedaliera Ospedali Riuniti Villa Sofia Cervello, PO Cervello,  
Via Trabucco, 180 - 90146 Palermo, Italia  
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### Specific areas of interest:

**Clinical Genetics, Genetics of Intellectual Disability and Autism,  
Dysmorphology,  
Fetal Genetic Disorders and Prenatal Genetic Counseling  
Mosaic Chromosomal Disorders,  
Pallister Killian Syndrome**

## WORK EXPERIENCE

July 2018 until now

Name of employer

### **Clinical Geneticist**

Riuniti Villa Sofia-Cervello Hospitals

Medical Genetic Unit - Regional Referral Center for Genetic and Chromosomal Diseases

Main activities and responsibilities

Principal subjects/occupational skills covered

Genetics of Intellectual disability and Autism, Dysmorphology, Chromosomal Disorders  
Prenatal Genetic Counseling

April 2016 - March 2018

Name of employer

### **Post-doctoral fellow in Medical Genetics**

The Children's Hospital of Philadelphia

Genetic Department – PI Prof. Krantz ID

Main activities and responsibilities

Principal subjects/occupational skills covered

Molecular and Clinical Characterization of Pallister Killian Syndrome (PKS)  
Pallister Killian Syndrome – Cornelia de Lange Syndrome and related disorders.

October 2015- February 2016

Name of employer

### **Pediatric Consultant**

Pediatrician at General Pediatric Services - Local Health Agency of Palermo – ASP Palermo (Italy)

Main activities and responsibilities

General Pediatrics

## RESEARCH EXPERIENCE

April 2022 until now

Scientific and Clinical Research Registry on Pitt-Hopkins Syndrome

Medical Genetic Unit - Regional Referral Center for Genetic and Chromosomal Diseases - Riuniti Villa Sofia-Cervello Hospitals

PI: Pr. M. Piccione

Position: Co-investigator

July 2018 until now

Management and Improvement of multidisciplinary and multispecialistic approach for patients affected by autism spectrum disorders

Medical Genetic Unit - Regional Referral Center for Genetic and Chromosomal Diseases - Riuniti Villa Sofia-Cervello Hospitals

PI: Pr. M. Piccione

Position: Investigator

April 2016-April 2018

Analysis of potential therapeutic compound for PKS through the use of cell-based tissue culture; defining a prenatal profile for PKS; delineating of the PKS clinical features in underrepresented minority populations.

Genetic Department - The Children's Hospital of Philadelphia

PI: Prof. Krantz ID

Position: Res. Post-doctoral Fellow

September 2014-May 2015

Genomic Characterization of patients with ID in the spectrum of Pitt Hopkins Like Syndrome by Next Generation Sequencing Analysis (gene panels/Whole Exome Sequencing).

Institute of Genomic Medicine, A. Gemelli Hospital, Catholic University, Rome, Italy.

PI: Prof. Zollino M

Position: Pediatric Resident

## EDUCATION AND TRAINING

April 2018 – May 2019

Name and type of organization providing education and Training

### II Level Master Degree in Clinical Genetics

University of Siena

- Training in Fetal Genetic Disorders and Prenatal Genetic Screening/ Diagnosis at Medical Genetic Division - Fondazione IRCCS Cà Granda - Maggiore Policlinico Hospital (*Tutor Prof.ssa F. Lalatta*)

May 2010 – May 2015

### Postgraduate Diploma of Specialization in Pediatrics **Level 50/50 cum laude**

With the thesis "Genomic Characterization of patients with Intellectual Disability related to Pitt Hopkins Syndrome phenotype by Next Generation Sequencing"

*Tutors: Prof. G. Corsello; Prof. M. Zollino\*; Dr. M. Piccione*

\* Institute of Genomic Medicine, A. Gemelli Hospital, Catholic University, Rome, Italy.

Name and type of organization providing education

University of Palermo. Faculty of Medicine and Surgery. Maternal and Child Health Department. Pediatric residency training program

Principal subjects/occupational skills covered

**Diagnosis and Management of Genetic Syndromes and other Rare Genetic Diseases. CGH – array data analysis. Next-Generation Sequencing analysis.**

Pediatric Genetics Residency Training

- Clinical Genetic Trainee at Medical Genetic Service of Palermo Polyclinic University Hospital and at Centre for Prevention Control and Cure of Down's Syndrome and Genetic Diseases of Palermo Villa Sofia-Cervello Hospital (May 2010-May 2015)
- Clinical Genetic Trainee at Human Genetic Department of Catholic University Medical School, Rome (September 2014- December 2014).

Other Residency Training Attended

- **Neonatology and Neonatal Intensive Care Unit – "Paolo Giaccone" Polyclinic University Hospital, Palermo** (August 2010-October 2010, February 2011-April 2011, December 2011- March 2012, February 2013, February 2014-May 2015)
- **Pediatric Nephrology Unit** - A.R.N.A.S. Civico - Di Cristina Benfratelli Hospital, Palermo (November 2013-January 2014)
- **Pediatric Onco-hematology Unit** - A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (August 2013-October 2013)
- **Pediatric Endocrinology** - A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (May 2013-July 2013)
- **General Pediatrics practitioner**, Palermo (March 2013-April 2013)
- **Infectious Disease Unit** - A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (November 2012- January 2013)

- **Cystic Fibrosis Regional Centre** A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (August 2012 - October 2012)
- **Centre for inborn metabolic diseases** - Pediatric Clinic Unit - A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (April 2012-July 2012)
- **Pediatric Gastroenterology** – Pediatric Clinic Unit – A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (August 2011-November 2011)
- **Pediatric Diabetology** – Pediatric Clinic Unit- A.R.N.A.S. Civico Di Cristina Benfratelli Hospital, Palermo (May 2011-July 2011, November 2010-Janary 2011)

**P-BLS Course** – Pediatric Residency Program, Maternal and Child Health Department. University of Palermo - April 2015

**Neonatal Resuscitation Course according to 2010 Italian Society of Neonatology Guidelines** - Pediatric Residency Program, Maternal and Child Health Department. University of Palermo - April 2015

October 2008 - April 2010

Name and type of organization providing education and training

**Pediatric Genetics Trainee**

Centre for Prevention Control and Cure of Down’s Syndrome and Genetic Diseases of Palermo, Maternal and Child Health Department. University of Palermo

November 2008 – February 2009

Name and type of organization providing education and training

Principal subjects/occupational skills covered

**Professional Habilitation**

University of Palermo. Faculty of Medicine and Surgery

General Practice, General Surgery, Internal Medicine.

**Level 270/270**

September 2002 – September 2008

Name and type of organization providing education and training

Principal subjects/occupational skills covered

**Medicinae Doctor**

with the thesis “Small Supernumerary Marker Chromosomes: clinical and cytogenetic evaluation of nine patients”. Admitted to “A. Albanese” Prize contest

*Tutor: Dr. M. Piccione*

**Level 110/110 cum laude**

University of Palermo. Faculty of Medicine and Surgery

- October 2006-September 2008 - Internship, *Pediatrics Department, Neonatal Intensive Care Unit*, Faculty of Medicine and Surgery, University of Palermo.

- October 2006-September 2008 - Internship, *Centre for Diagnosis and Care of Down’s Syndrome and Rare Genetic Diseases*, Pediatrics Department, Faculty of Medicine and Surgery, University of Palermo

September 1997- June 2002

Name and type of organization providing education and training

Principal subjects/occupational skills covered

**Diploma of Secondary School in Classical Studies** **Level 100/100 with honors**

Secondary School “Liceo Classico Statale G. Garibaldi”, Palermo, Italy.

Classical Studies, Maths, Physics and Biology.

## RESEARCH GRANTS AND AWARDS

**PKS Research Fellowship 2017 at Children’s Hospital of Philadelphia**  
funded by PKS Kids USA Foundation (Gross amount \$ 58.000,00)

**Italian Society of Pediatrics “Pioneers of Pediatrics” Research Grant 2016 Award** funded by Italian Society of Pediatrics (Gross amount € 10.000,00)

**PKS Kids Italia Onlus Research Grant 2015 Award at Children’s Hospital of Philadelphia** funded by PKS Kids Italia Onlus (Gross amount € 30.000,00)

**Pitt Hopkins Syndrome Best Thesis 2015 Award** funded by Pitt Hopkins Syndrome Italian Association - ONLUS

**Best Oral Communication:** 5<sup>th</sup> SIMMESN/SIMGePeD National Congress Nov 26-28, 2013, Naples, SIMGePeD Session.

*“Noonan Syndrome and congenital heart defects: genotype-phenotype correlation”.*

**Best Oral Communication:** 39<sup>th</sup> SIP/SIN/SIMEUP, Regional Congress Nov 11-13, 2010, Palermo, SIP Session.

*“Small supernumerary markers chromosome in patients with neuromotor delay”.*

## RESEARCH AND LABORATORY SKILLS

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- Experimental design, Data Manager (Laboratory and Patients Med. Records), Data Analysis and Interpretation
- Cell Culture Procedures (Feeding, subculturing, havervesting, freezing)
- DNA isolation
- RNA isolation
- Bisulfite Conversion of DNA
- Digital Droplet PCR (ddPCR)Technology (Copy Number Variation Assay)
- RT, qPCR protocols

## COURSES IN CLINICAL GENETICS

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- ❖ **Multidisciplinary Fetal Diagnostics Winter School** – ERN Ithaca - Edition 2023, Paris, December 7, 2023.
- ❖ **XV Italian Society of Human Genetics Conference**, September 2022
- ❖ **Neurocutaneous Syndromes**, Fondazione Mariani Course, October 2021
- ❖ **Fetal Malformation 1.0.**, Medical Channel On Line Course, October 2021
- ❖ **CNVs Interpretation Course**, Italian Society of Human Genetics, December 2020
- ❖ **Skeletal Dysplasias Course**, Italian Society of Human Genetics, December 2020
- ❖ **XXIII Italian Society of Human Genetics Conference**, November 2020
- ❖ **Next Generation Sequencing in Diagnosis. From Exome to Genome Sequencing. Omics Technologies**, Rome, October 22-23, 2020
- ❖ **XXII Italian Society of Human Genetics Conference**, Rome, November 13-16, 2019
- ❖ **38th David W. Smith Workshop on Malformations and Morphogenesis**, Stowe, VT, August 26-29
- ❖ **Deciphering Beckwith-Wiedemann Syndrome**, Philadelphia, July 21 -23, 2017

- ❖ **2016 PKS Kids Face2Face Meeting**, Chicago, July, 2016
- ❖ **The Annual Philadelphia Genetic Meeting, Philadelphia**, June 8, 2016
- ❖ **“Congenital Heart Diseases: from diagnosis to prophylaxis of respiratory tract infections”** Palermo, October 8, 2015.
- ❖ **Course on Rare Diseases and Clinical Genetics - XII National ONSP Days**, Padova 30 Sept - 1 Oct 2015.
- ❖ **E-Learning Course “Approach to Rare Diseases”**, Provider Ospedale Pediatrico Bambino Gesù IRCCS, 1 Oct 2014 – 30 Sept 2015.
- ❖ **Course on Labiopalatoschisis** Palermo 8 May 2015.
- ❖ **Italian Society of Pediatric Genetic Diseases and Congenital Disabilities “What the Pediatrician should Know about genetic syndromes”** Roma 4 June 2015.
- ❖ **“Neuromuscular Diagnosis in Medical Genetics” Italian Society of Human Genetics Course** Roma 10 Dec 2014.
- ❖ **Italian Society of Human Genetics National “Next Generation Sequencing in Medical Genetics”** Bologna 30-31 Oct 2014.
- ❖ **“Diagnosis of Intellectual Disability” Italian Society of Human Genetics Course** Roma 19 Sept 2014.
- ❖ **Course on Metabolic Diseases “1° Seminar on Lysosomal Storage Diseases”** Bologna 19-20 Mar 2014.
- ❖ **Expert Meeting on Rare Diseases** Palermo 5 Oct 2012.
- ❖ **Course on “Diagnosis e Clinical Assistance in Rare Diseases for Pediatrician and General Practitioner”** Palermo 21 Jan 2012.
- ❖ **E-Learning Course in Pediatric Genetics V Online Edition – Metodological Approach to the child with Complex Malformative Syndrome.** Fondazione Mariani, 15 Feb – 11 Apr 2010.

## COURSES IN BASIC/TRASLATIONAL RESEARCH

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- ❖ **Science Research Writing for Non-Native Speakers of English (BPP)**, Philadelphia, February 2017
- ❖ **Responsible Conduct of Research (RCR) training**, Philadelphia, April 2017
- ❖ **Conflict of interest Training**, Philadelphia, 2017
- ❖ **CITI Protection of Human Research Subjects Training**, March 2016

## INVITED SPEAKER

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**3<sup>RD</sup> European Workshop on Pallister Killian Syndrome**, Sassuolo, Sept 6-8, 2019

**Congenital Heart Campus 2019**, Siracusa (CT) 25 May, 2019.

**Course on Genetics of Neurodevelopmental Disorders**, Villa Nave Palermo (PA) 10 Nov, 2018.

**2<sup>nd</sup> European Workshop on Pallister Killian Syndrome**, Cervia (RA) 29 Sep-1 Oct, 2017.

## EDITORIAL

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### Manuscripts ad hoc reviewer:

American Journal of Medical Genetics Part A

Genes

Journal of Pediatric Genetics

Medicina

Molecular Cytogenetics

**MEMBERSHIPS  
IN PROFESSIONAL  
ORGANIZATIONS**

- SIGU (Italian Society of Human Genetics)
- SIP (Italian Society of Pediatrics)
- PKS Kids Italia Onlus

**LANGUAGE SKILLS**

MOTHER TONGUE

**ITALIAN**

OTHER LANGUAGES

**ENGLISH**

UNDERSTANDING		SPEAKING		WRITING
Listening	Reading	Spoken interaction	Spoken production	
C1	C1	C1	C1	C1

October 2016 - November 2016

ESL Course –Strategically Speaking: building fluency for Confident Communication Language Connected Biomedical Postdoctoral Program – University of Pennsylvania

October 1999 – February 2002

English Course - Satisfactorily completed 7° (upper-intermediate) level of ten. International House English Language Centre, Palermo, Italy.

**COMPUTER SKILLS**

Good Experience in using Microsoft Office (Excel, Word, PowerPoint) and GraphPad Prism. Proficiency in using internet as research tool.

**ADDITIONAL INFORMATION**

Volunteer at the “First Aid - Pediatric Ambulatory for Immigrants”, Centro Agape, Medical Community Service, Arcidiocesi and Caritas of Palermo (2007-2008)

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## PUBLICATIONS

### Book Chapter

- **Salzano, E.**, Raible, S.E. and Krantz, I.D. (2021). Pallister Killian Syndrome - In Cassidy and Allanson's Management of Genetic Syndromes (eds J.C. Carey, A. Battaglia, D. Viskochil and S.B. Cassidy), pp. 717-733, first published: 30 October 2020

-Fragapane T, **Salzano E**, Vecchio D and Maria Piccione, Neurofibromatosis Type 2- In Vestibular Schwannoma: Pathophysiology, Diagnosis and Treatment (eds. Francesco Dispenza, MD, PhD, Francesco Martines, MD, PhD) Chapter 3, Publication Date: April 2020. ISBN: 978-1-53617-191-4.

### PEER-REVIEWED JOURNAL

- Karimi, K., Mol, M. O., Haghshenas, S., Relator, R., Levy, M. A., Kerkhof, J., ... **Salzano E.**, ... & Sadikovic, B. (2024). Identification of DNA methylation episcapature for the intellectual developmental disorder, autosomal dominant 21 syndrome caused by variants in the CTCF gene. *Genetics in Medicine*, 26(3):101041.

- Leone, M. P., Morlino, S., Nardella, G., Pracella, R., Giachino, D., Celli, L., ... **Salzano E.**, ... & Castori, M. (2023). Specifications and validation of the ACMG/AMP criteria for clinical interpretation of sequence variants in collagen genes associated with joint hypermobility. *Human Genetics*, 142(6), 785-808.

- **Salzano, E.**, Niceta, M., Pizzi, S., Radio, F. C., Busè, M., Mercadante, F., ... & Piccione, M. (2023). Case report: Novel compound heterozygosity for pathogenic variants in MED23 in a syndromic patient with postnatal microcephaly. *Frontiers in neurology*, 14, 84.

- Vinciguerra, M., Leto, F., Cassarà, F., Tartaglia, V., Malacarne, M., Coviello, D., ...**Salzano E.**, ... & Giambona, A. (2023). Incidental Detection of a Chromosomal Aberration by Array-CGH in an Early Prenatal Diagnosis for Monogenic Disease on Coelomic Fluid. *Life*, 13(1), 20.

- Mercadante, F., Piro, E., Busè, M., **Salzano, E.**, Ferrara, A., Serra, G., ... & Piccione, M. (2022). Cutis verticis gyrate and Noonan Syndrome: report of two cases with pathogenic variant in SOS1 gene. *Italian Journal of Pediatrics*, 48(1), 1-7.

- Poeta, L., Malacarne, M., Padula, A., Drongitis, D., Verrillo, L., Lioi, M. B., ... & Miano, M. G. (2022). Further Delineation of Duplications of ARX Locus Detected in MALE Patients with Varying Degrees of Intellectual Disability. *International journal of molecular sciences*, 23(6), 3084. doi: 10.3390/ijms23063084

- Mercadante, F., Busè, M., **Salzano, E.** Fragapane, T., Palazzo, D., Malacarne, M., & Piccione, M. 12q14.3 microdeletion involving HMGA2 gene cause a Silver-Russell syndrome-like phenotype: a case report and review of the literature. *Italian Journal of Pediatrics* volume 46, Article number: 108 (2020).

- Dowsett, L., Porras, A. R., Kruszka, P., Davis, B., Hu, T., Honey, E., ... & **Salzano, E.**, Jackson, L., Dearthoff, M., Kline, A., Summar, M., Muenke, M., Linguraru M.G., Krantz, I.D. Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics Part A*. (2019).

- **Salzano, E.**, Raible, S. E., Kaur, M., Wilkens, A., Sperti, G., Tilton, R. K., ... & Conlin, L. K. Prenatal profile of Pallister-Killian syndrome: Retrospective analysis of 114 pregnancies, literature review and approach to prenatal diagnosis. *American Journal of Medical Genetics Part A*. 2018,176(12), 2575-2586.

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- Piccione, M., & **Salzano, E.** (2017). *Mulibrey nanism. Atlas of Genetics and Cytogenetics in Oncology and Haematology.*

- Piccione, M., & **Salzano, E.** (2016). *Denys-Drash syndrome (DDS). Atlas of Genetics and Cytogenetics in Oncology and Haematology.*

- Giuffrè M, Mocerì G, Vecchio D, Antona V, **Salzano E**, Corsello G *Intellectual disability in developmental age. Italian Journal of Pediatrics.* 2015, 41 (Suppl 2): A38.

- Corsello G, **Salzano E**, Vecchio D, Antona V, Grasso M, Malacarne M, Carella M, Palumbo P, Piro E, Giuffrè M. *Paternal uniparental disomy chromosome 14-like syndrome due a maternal de novo 160 kb deletion at the 14q32.2 region not encompassing the IG- and the MEG3-DMRs: Patient report and genotype-phenotype correlation. Am J Med Genet A.* 2015. 167(12), 3130-3138.

- Piccione M, **Salzano E**, Vecchio D, Ferrara D, Malacarne M, Pierluigi M, Ferrara I, Corsello G. *4p16.1-p15.31 duplication and 4p terminal deletion in a 3-years old Chinese girl: Array-CGH, genotype-phenotype and neurological characterization, European Journal of Paediatric Neurology (2015).*

- Vecchio D, **Salzano E**, Piro E, Insinga V, Piccione M, Giuffrè M, Ferrara I, Corsello G *Unusual paroxysmal autonomic manifestations in a 22 month old girl. Euromediterranean Biomedical Journal* 2014, 9(S1):75-95.

- Vecchio D, **Salzano E**, Piro E, Insinga V, Piccione M, Giuffrè M, Ferrara I, Corsello G *Unusual paroxysmal autonomic manifestations in a 22 month old girl Euromediterranean Biomedical Journal* 2014, 9(S1):75-95.

- Piccione M, Vecchio D, **Salzano E**, Corsello G. *Delineating a new critical region for juvenile myoclonic epilepsy at the 22q11.2 chromosome. Epilepsy Behav.* 2013. 29(3):587-8.

- Vecchio D, Piccione M, Giuffrè M, Piro E, **Salzano E**, Ferrara D, Insinga V, Corsello G. *An acute headache due a nodular swelling of the skull. EuroMediterraneanBiomedical Journal.* 2013. 8(S1): 99-100.

- Piccione M, Antona V, Graziano L, Consiglio V, **Salzano E**, Vecchio D, Lauricella SA, Corsello G. *X-Linked Intellectual Disability. Acta Medica Mediterranea.* 2013. 29: 77-8.

- Piro E, Consiglio V, Agrifoglio M, Sireci F, Ballacchino A, Salvago P, Martines F, Graziano F, Busè M, Sanfilippo C, Vecchio D, **Salzano E.** *Diagnosis and follow-up of Complex Congenital Malformations/MentalRetardation (MRA/MR). Acta Medica Mediterranea.* 2013. 29: 321-2.

- Vecchio D, **Salzano E**, Vecchio A, Roccella M. *A rare unbalanced translocation 1;18 in a child with epilepsy, mild dysmorphology and mental retardation. Minerva Pediatr.* 2012. 64(3): 365-367.

## ORAL PRESENTATIONS

- Piccione M, Antona R, **Salzano E**, Cavani S, Malacarne M, Morreale Bubella, Pierluigi M, Viaggi C.D, Corsello G. *Array CGH and Clinical characterization in a patient with subtelomeric 6p deletion without ocular dysgenesis. 2011. Am J Med Genet Part A 158A:150–154.*



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-Vecchio D, **Salzano E**, Vecchio A, Di Filippo T, Roccella M. A case of femoral-facial syndrome in a patient with autism spectrum disorders. 2011. *Minerva Pediatr.* 63(4): 341-4.

- Vecchio D, Moncada A, Giambona A, **Salzano E**, Piccione M. Solving the Complexity in Diagnosis of Autosomal Recessive Primary Microcephaly (McpH) Through a Comprehensive Next Generation Sequencing (NGS) Disease-Customized Genes Panel. Clinical Report and Genotype-Phenotype Correlation of a homogeneous Patients' Cohort. *Pediatrics, Cogent Medicine* (2017), 4: 1408251.

- Dowsett LW, Noon SE, **Salzano E**, Deardorff MA, Kline AD, Kruzka P, Muenke M, Krantz ID. Cornelia De Lange Syndrome in Diverse Populations. Proceedings of 38th David W. Smith Workshop on Malformations and Morphogenesis, Stowe, VT, August 26-29<sup>th</sup>, 2017.

- Maccora I, Piro E, Puccio G, Li Puma L, Angileri V, **Salzano E**, Campo C, Giuffrè M, Arculeo P, Pappalardo MP, Vitaliti M, Corsello G. Developmental abnormalities of the CNS: epidemiological investigation in a sample of newborns from 2010 to 2014. Proceeding of XL National Pediatric Neurology Italian Society, Palermo Nov 2014.

- **Salzano E**, Lo Presti C, Giuffrè M, Carta M, Schierz IAM, Li Voti G, Vecchio D, Corsello G. Apple-peel digiuno-ileal atresia: a rare neonatal onset of cystic fibrosis *Riflessioni Universo Pediatria Anno X, Speciale Suppl. al N. 1 - March 2015 ISSN 2039-8344.*

- Lo Presti C, **Salzano E**, Curtopelle G, Provenzano C, Gabriele B, Mocerì G, Corsello G. CMV Congenital Infection involving intestinal tract in a term newborn. *Riflessioni Universo Pediatria Anno X, Speciale Suppl. al N. 1 – March 2015 ISSN 2039-8344.*

- Vecchio D, **Salzano E**, Piro E, Piccione M, Maranto M, Corsello G. Temporal epilepsy and autonomic paroxysmal in preschool age: clinical report. Proceedings of 70° Italian Pediatric Society Congress, Palermo June 11-14, 2014.

- **Salzano E**, Genova S, Matina F, Insinga V, Maniscalchi V, La Placa S, Piccione M, Giuffrè M, Corsello G. Noonan Syndrome and congenital heart defects: genotype-phenotype correlation. Proceedings of 5<sup>th</sup> Congresso Nazionale Congiunto SIMMESN/SIMGePeD, SIMGePeD Session, Naples Nov 26-28, 2013.

- Accomando S, **Salzano E**, Gennaro P, Alga P, Tricarico A, Lombardo A, Corsello G. Cutaneous manifestations in pediatric celiac disease. Proceedings of 54<sup>th</sup> Annual Meeting European Society for Pediatric Research. Porto, Oct 10-14, 2013.

- **Salzano E**, Corsello G. A short stature critical region: *PAR 1 (Xp22.33 – Yp11.32)*. Proceedings of Palermo Pediatria- 3rd Edition – Interactive Clinical Cases, Palermo June 2013.

- Termini L, **Salzano E**, Ficili F, Traverso G, Giordano G, Riva S, Scivares M, Corsello G, Di Girgenti C, Collura M. Neonatal cholestasis: atypical presentation of cystic

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## POSTER PRESENTATIONS

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fibrosis. Oral Communication. Proceedings of 8<sup>th</sup> Congresso Nazionale Società Italiana Fibrosi Cistica, Tirrenia, Nov 14-17, 2012.

- *Gagliano F, Accomando S, Vergara B, **Salzano E**, Maniscalchi E, Ferraro F, Bonanno M, Galione G, Salli A, Scavone V, Troisi M, Corsello G.* Acute idiopathic thrombocytopenic purpurain children: role of viral infections. Proceedings of 40<sup>th</sup> Congresso Regionale SIP, Catania, Nov 17-19, 2011.

- *Piccione M, **Salzano E**, Consiglio V, Malacarne M, Corsello G.* Small supernumerary markers chromosome in patients with neuromotor delay. Proceedings of 39<sup>th</sup> Congresso Regionale SIP, SIN SIMEUP Nov 11-13, 2010.

- *Serruto M, Cigna V, Orlandi E, Schillaci G, Colavito D, Mercadante F, Fragapane T, Salzano E, Piccione M.*

Diagnosi prenatale di Sindrome di Bardet-Biedl: segni ultrasonografici, revisione della letteratura e correlazione genotipo-fenotipo fetale di variante patogenetica del gene *BBS10*. Proceedings of 75<sup>th</sup> Congresso Italiano di Pediatria, Bologna, May 29-June 1<sup>st</sup>, 2019

- *Moncada A, Vecchio D, **Salzano E**, Buse' M, Passarello C, Giambona A, Maggio A, Piccione M.* Diagnosis of Autosomal Recessive Primary Microcephaly (MCPH) in a cohort of patients through a Comprehensive Next Generation Sequencing (Ngs). Proceedings of the XX Italian Society of Human Genetics National Conference, Naples, November 15-18, 2017.

- ***Salzano E**, Noon SE, Dowsett LK, Wilkens A, Muenke M, Izumi K, Krant ID.* Pallister-Killian Syndrome Across Different Ancestry Groups. Proceedings of 38th David W. Smith Workshop on Malformations and Morphogenesis, Stowe, VT, August 26-29<sup>th</sup>, 2017.

- *Vecchio D, Antona V, Giuffrè M, Lagalla L, **Salzano E**, Insinga V, D'Anna A, Malacarne M, Corsello G.* 17p13.3 microduplication syndrome: characterization of a new critical region for a clinical variant with cleft palate. Proceedings of 72° Italian Pediatric Society Congress, Florence 16 November 2016.

- *Vecchio D, Antona V, Giuffrè M, Lagalla L, **Salzano E**, Piro E, Malacarne M, Corsello G.* 1q21duplication syndrome: new 6Mb rearrangement with variable penetrance and familial clinical expression. Proceedings of 72° Italian Pediatric Society Congress, Florence 16 November 2016.

- *Vecchio D, Antona V, Giuffrè M, **Salzano E**, Lagalla L, Pisaneschi E, Selicorni A, Corsello G.* New NPBL gene mutation in a patient affected by CDLS: case report and genotype-phenotype correlation. Proceedings of 72° Italian Pediatric Society Congress, Florence 16 November 2016.

- *Vecchio D, Piccione M, D'Adamo P, Mignogna M, **Salzano E**, Giuffrè M, Antona V, Caputo V, Pizzuti A, Nardello R, Piro E, Capobianco E, Corsello G.* Intellectual disability, epilepsy and mild dysmorphisms due 22q11.2 distal duplication: clinical and molecular characterization of a 0.5 mb minimal critical region. (2016). doi:10.1007/s00431-016-2785-8

- *Moceri G, Vecchio D, **Salzano E**, Antona V, Lauricelli F, Piro E, Giuffrè M, Corsello G.*

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Early Diagnosis of Syndromic Intellectual Disability. Proceedings of XXI Italian Society of Neonatology Congress Palermo 24-26 September 2015.

-Falcone V, Alletto A, La Placa S, **Salzano E**, Antona V, Piccione M, Giuffrè M, Corsello G. Pulmonary Stenosis and Facial Dysmorphic Traits: neonatal diagnosis and follow-up in Noonan Syndrome. Proceedings of XXI Italian Society of Neonatology Congress Palermo 24-26 September 2015.

- Antona V, **Salzano E**, Vecchio D, Mocerì G, Piro E, Giuffrè M, Corsello G. Ichthyosis and Intellectual Disabilities: non casual association. Proceedings of 71° Italian Pediatric Society Congress, Rome 4-6 June 2015.

-Piro E, **Salzano E**, Antona V, Vecchio D, Tiziano D, Corsello G. Severe Hypotonia in newborn with arthrogryposis: clinical report and diagnostic matters. Proceedings of 70° Italian Pediatric Society Congress, Palermo 11-14 June 2014.

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Palermo, August 01, 2024

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Autorizzo il trattamento dei miei dati personali ai sensi del Regolamento UE 2016/679.

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