PERSONAL INFORMATION

Edoardo Errichiello

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DEGREES

Ph.D. in Genetics, Molecular and Cellular Biology
Specialization in Clinical Pathology
M.Sc. Sanitary Biology
B.Sc. Molecular Sciences

WORK EXPERIENCE

November 2019 -
Senior Clinical Researcher
Unit of Cytogenetics and Molecular Genetics, Department of Laboratory Medicine, IRCCS Mondino Foundation, Pavia, Italy

March 2018 -
Non-tenured Assistant Professor of Medical Genetics
Unit of Medical Genetics, Department of Molecular Medicine, University of Pavia

November 2013 – February 2017
PhD/PostDoc
Unit of Medical Genetics, Department of Molecular Medicine, University of Pavia

November 2012 - October 2013
Visiting Scientist
Laboratory of Neurogenetics, National Institutes of Health (NIH), Bethesda, USA

October 2011 - October 2012
Research Fellow
Unit of Molecular Genetics, Department of Laboratory Medicine, Regina Margherita Children's Hospital, Torino

April 2011 – September 2011
Visiting Scientist
Unit of Oncogenomics, Kimmel Cancer Center, Thomas Jefferson University, Philadelphia, USA

March 2008 - March 2011
Research fellow
Unit of Stem Cell Transplantation and Cell Therapy, Department of Pediatric Onco-Hematology, Regina Margherita Children's Hospital, Torino

November 2007 - February 2008
Biologist trainee
Unit of Molecular Pathology, Institute for Cancer Research and Treatment, Candiolo

EDUCATION AND TRAINING

November 2013 – January 2017

Ph.D. in Genetics, Molecular and Cellular Biology

University of Pavia

Thesis' title: "*SMARCA4* inactivating mutations cause concomitant Coffin-Siris syndrome, microphthalmia and small cell carcinoma of the ovary, hypercalcemic type"

March 2008 - March 2013

Specialization in Clinical Pathology

University of Torino

Thesis' title: "Lo stress ossidativo contribuisce alla progressione della poliposi associata a *MUTYH* tramite specifiche mutazioni del DNA mitocondriale e del pathway MAPK-ERK" (70/70 cum laude)

November 2005 - November 2007

M.Sc. in Sanitary Biology

University of Torino

Thesis' title: "Pathways genetici alternativi nelle poliposi del colon" (110/110 cum laude)

November 2005 - November 2007

B.Sc. in Molecular Sciences

University of Torino

Thesis' title: "ROS-mediated mutations in mitochondrial DNA in colorectal cancerogeneis" (110/110 cum laude)

PERSONAL SKILLS AND COMPETENCES

Mother tongue: Italian

Other languages

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	C2	C2	C2	C2	C2
French	A2	A2	A2	A2	A2

A1/A2: Basic user - B1/B2: Intermediate user - C1/C2: Advanced user

Organisational skills and competences: Organization of research and clinical activities. Validation and standardization of molecular and cytogenetic methods

Social skills and competences: Ability to work as a part of a team, to plan, organize and prioritize work activities

Technical skills and competences: *Molecular Biology:* DNA/RNA/microRNA extraction from different biological matrices (blood, buffy coat, saliva, buccal swabs, hair, sputum, formalin fixed paraffin embedded - and frozen tissues, cell culture fluid and cells,…), DNA/RNA quality check (NanoDrop, Qubit, Agilent 2100 Bioanalyzer, Fragment Analyzer), direct sequencing, Next Generation Sequencing (ChiP-seq, RNA-seq, whole exome and

genome sequencing), genotyping, Real-Time PCR, Allele Specific-PCR, microsatellite analysis, Southern Blot, Methylation Specific PCR, Competitive Allele Specific PCR (castPCR), pyrosequencing, chromosome X inactivation assay

Cytogenetics: karyotyping, Fluorescent in situ hibridization (FISH), Flow-FISH

Cell Biology: cell culture, cell separation, freezing/thawing cells, immunohistochemistry immunofluorescence, flow cytometry

Haematology: analysis of blood smears, coagulation tests

Other expertise: Parasitology, Toxicology, urine test, ELISA, HPLC, cytological analysis

Computer skills and competences

SELF-ASSESSMENT				
Elaboration	Communication	Content creation	Security	Problem resolution
Advanced	Advanced	Advanced	Advanced	Advanced

Advanced knowledge of graphical (e.g., Office, Photoshop), bioinformatics and statistics (e.g., R) programs. Experience in using genetic softwares and bioinformatic tools (Gene Mapper, SeqScape, Sequencer, Chrome Lite, BLAST, Primer 3, gPLINK, UCSC, variant prioritization tools and databases for NGS data analysis, …).

Advanced knowledge of programs for the analysis of nucleotide sequences (Sequencer, SeqScape, SeqScanner) and fragments (PeakScanner, GeneMapper), design and in silico validation of primers (Primer3, BLAST). Knowledge of bioinformatics tools for the annotation and interpretation of germline and somatic variants and for the prioritization of gene variants (e.g., UCSC, HGMD, IGV, Alamut, Mutation Taster, Panther, Phenomizer, Endeavor, ToppGene, GeneDistiller, ExAC, gnomAD, COSMIC, ...). Knowledge of programs for the analysis and filtering of NGS data (exome/genomes, gene panels, RNA-seq, 10X). Knowledge of bioinformatics databases used in clinical genetics (DECIPHER, OMIM, ...) and for interactive pathway analysis (pathway enrichment, PPI networks, ...), RNA-seq, etc ...

Editorial activity

Reviewer for the following journals: Journal of Medical Genetics, European Journal of Human Genetics, European Journal of Medical Genetics, Clinical Genetics, World Journal of Surgical Oncology, Brain and Development, Life (MDPI), Journal of the Neurological Sciences, Frontiers in Genetics, Frontiers in Immunology, OncoTargets and Therapy, JSM Gastroenterology and Hepatology, Journal of Laboratory and Clinical Medicine, The Journal of Pediatric Genetics, European Journal of Paediatric Neurology, Medical Sciences, Pediatric Research. Guest Editor for: Frontiers in Genetics, Medical Sciences, OBM Genetics, Computational and Mathematical Methods in Medicine.

External abstract reviewer per la conferenza “European Human Genetics (ESHG) 2019” (Göteborg)

Awards

Gigi Ghirotti (2011)
SIGU (Italian Society of Human Genetics) Claudio Castellan – Best oral communication in Clinical Genetics (2018)

BIBLIOGRAPHY

Tzialla C, Arossa A, Mannarino S, Orcesi S, Veggiotti P, Fiandrino G, Zuffardi O, **Errichiello E**. SCN2A and arrhythmia: A potential correlation? A case report and literature review. Eur J Med Genet. 2022;65(12):104639. doi: 10.1016/j.ejmg.2022.104639.

Caretto A, **Errichiello E**, Patricelli MG, Zuffardi O, Cristel G, Ravelli S, Sirtori M, Scavini M, Bosi E, Martinenghi S. Transcutaneous electrical stimulation therapy and genetic analysis in Dercum's disease: A pilot study. Medicine (Baltimore). 2021;100(51):e28360. doi: 10.1097/MD.0000000000028360.

Berlincioni V, Catania C, Acerbi F, Spinillo A, Arossa A, Kurtas NE, **Errichiello E**, Zuffardi O. Boundaries and precision medicine in consanguineous migrant couples Genetic counseling after the identification of fetal pathologies. Book chapter: Borders, Migration and Globalization: An Interdisciplinary Perspective. 1st Edition, 2021, Routledge (eBook ISBN9781003106517).

Liu S, Aldinger KA, Cheng CV, Kiyama T, Dave M, McNamara HK, Zhao W, Stafford JM, Descostes N, Lee P, Caraffi SG, Ivanovski I, **Errichiello E**, Zweier C, Zuffardi O, Schneider M, Papavasiliou AS, Perry MS, Humberson J, Cho MT, Weber A, Swale A, Badea TC, Mao CA, Garavelli L, Dobyns WB, Reinberg D. NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. Mol Cell. 2021;81(22):4663-4676.e8. doi: 10.1016/j.molcel.2021.09.020.

Carbone R, Rovedatti L, Lenti MV, Furlan D, **Errichiello E**, Gana S, Luinetti O, Arpa G, Alvisi C, De Grazia F, Valente EM, Sessa F, Paulli M, Vanoli A, Di Sabatino A. Histologic heterogeneity and syndromic associations of non-ampullary duodenal polyps and superficial mucosal lesions. Dig Liver Dis. 2021:S1590-8658(21)00126-2. doi: 10.1016/j.dld.2021.03.011.

Bonometti A, Lobascio G, Boveri E, Cesari S, Lecca M, Arossa A, Spinillo A, **Errichiello E**, Paulli M. Acute megakaryoblastic leukemia with a novel GATA1 mutation in a second trimester stillborn fetus with trisomy 21. Leuk Lymphoma. 2021:1-4. doi: 10.1080/10428194.2021.1907377.

Errichiello E, Giorda R, Gambale A, Iolascon A, Zuffardi O, Giglio S. RB1CC1 duplication and aberrant overexpression in a patient with schizophrenia: further phenotype delineation and proposal of a pathogenetic mechanism. Mol Genet Genomic Med. 2021;9(1):e1561. doi: 10.1002/mgg3.1561.

Errichiello E, Malara A, Grimod G, Avolio L, Balduini A, Zuffardi O. Low penetrance COL5A1 variants in a young patient with intracranial aneurysm and very mild signs of Ehlers-Danlos syndrome. Eur J Med Genet. 2021;64(1):104099. doi: 10.1016/j.ejmg.2020.104099.

Errichiello E, Mina T, Morbini P, Zecca M, Zuffardi O. FANCA, TP53, and del(5q)/RPS14 alterations in a patient with T-cell non-Hodgkin lymphoma and concomitant Fanconi anemia and Li-Fraumeni syndrome. Cancer Genet. 2020;S2210-7762(20)30278-7. doi: 10.1016/j.cancergen.2020.10.003.

Errichiello E, Arossa A, Iasci A, Villa R, Ischia B, Pavesi MA, Rizzuti T, Bedeschi MF, Zuffardi O. An additional piece in the TBX6 gene dosage model: A novel nonsense variant in a fetus with severe spondylocostal dysostosis. Clin Genet. 2020;98(6):628-629. doi: 10.1111/cge.13854.

Todisco M, Gana S, Cosentino G, **Errichiello E**, Arceri S, Avenali M, Valente EM, Alfonsi E. KCTD17-related myoclonus-dystonia syndrome: clinical and electrophysiological findings of a patient with atypical late onset. Parkinsonism Relat Disord. 2020;78:129-133. doi: 10.1016/j.parkreldis.2020.07.026.

Maini I, **Errichiello E**, Caraffi SG, Rosato S, Bizzarri V, Pollazzon M, Trimarchi G, Contrò G, Cavirani B, Gelmini C, Napoli M, Moratti C, Pascarella R, Rizzi S, Fusco C, Zuffardi O, Garavelli L. Improving the phenotype description of Basel-Vanagaite-Smirin-Yosef syndrome, MED25-related: polymicrogyria as a distinctive neuroradiological finding. Neurogenetics. 2021;22(1):19-25. doi: 10.1007/s10048-020-00625-2.

Bossi G, **Errichiello E**, Zuffardi O, Marone P, Monzillo V, Barbarini D, Vergori A, Bassi LA, Rispoli GA, De Amici M, Zecca M. Disseminated Mycobacterium Avium Infection in a Child with Complete Interferon-γ Receptor 1 Deficiency due to Compound Heterozygosis of IFNGR1 for a Subpolymorphic Copy Number Variation and a Novel Splice-Site Variant. J Pediatr Genet. 2020;9(3):186-192. doi: 10.1055/s-0039-1700803.

Errichiello E, Zagnoli-Vieira G, Rizzi R, Garavelli L, Caldecott KW, Zuffardi O. Characterization of a novel loss-of-function variant in TDP2 in two adult patients with spinocerebellar ataxia autosomal recessive 23 (SCAR23). J Hum Genet. 2020;65(12):1135-1141. doi: 10.1038/s10038-020-0800-4.

Errichiello E, Dardiotis E, Mannino F, Paloneva J, Mattina T, Zuffardi O. Phenotypic Expansion in Nasu-Hakola Disease: Immunological Findings in Three Patients and Proposal of

a Unifying Pathogenic Hypothesis. Front Immunol. 2019;10:1685. doi: 10.3389/fimmu.2019.01685.

Andolfo I, De Rosa G, **Errichiello E**, Manna F, Eleni Rosato B, Gambale A, Vetro A, Calcaterra V, Pelizzo G, De Franceschi L, Zuffardi O, Russo R, Iolascon A. PIEZO1 Hypomorphic Variants in Congenital Lymphatic Dysplasia Cause Shape and Hydration Alterations of Red Blood Cells. Front. Physiol. doi: https://doi.org/10.3389/fphys.2019.00258.

Berlincioni V, Catania C, Acerbi F, Spinillo A, Arossa A, Kurtas N, **Errichiello E**, Zuffardi O. Confini e medicina di precisione in coppie consanguinee di migranti: la ricezione della consulenza genetica successiva all'identificazione di patologie fetali. Confini, Giappichelli Editore, pagg 99-108.

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Kurtas NE, Xumerle L, Leonardelli L, Delledonne M, Brusco A, Chrzanowska K, Schinzel A, Larizza D, Guerneri S, Natacci F, Bonaglia MC, Reho P, Manolakos E, Mattina T, Soli F, Provenzano A, Al-Rikabi AH, **Errichiello E**, Nazaryan-Petersen L, Giglio S, Tommerup N, Liehr T, Zuffardi O. Small supernumerary marker chromosomes: A legacy of trisomy rescue? Hum Mutat. 2019;40(2):193-200.

Bonaglia MC, Kurtas NE, **Errichiello E**, Bertuzzo S, Beri S, Mehrjouy MM, Provenzano A, Vergani D, Pecile V, Novara F, Reho P, Di Giacomo MC, Discepoli G, Giorda R, Aldred MA, Santos-Rebouças CB, Goncalves AP, Abuelo DN, Giglio S, Ricca I, Franchi F, Patsalis P, Sismani C, Morí MA, Nevado J, Tommerup N, Zuffardi O. De novo unbalanced translocations have a complex history/aetiology. Hum Genet. 2018;137(10):817-829.

De Bernardi ML, Ivanovski I, Caraffi SG, Maini I, Street ME, Bayat A, Zollino M, Lepri FR, Gnazzo M, **Errichiello E**, Superti-Furga A, Garavelli L. Prominent and elongated coccyx, a new manifestation of KBG syndrome associated with novel mutation in ANKRD11. Am J Med Genet A. 2018;176(9):1991-1995.

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Maini I, Ivanovski I, Djuric O, Caraffi SG, **Errichiello E**, Marinelli M, Franchi F, Bizzarri V, Rosato S, Pollazzon M, Gelmini C, Malacarne M, Fusco C, Gargano G, Bernasconi S, Zuffardi O, Garavelli L. Prematurity, ventricular septal defect and dysmorphisms are independent predictors of pathogenic copy number variants: a retrospective study on array-CGH results and phenotypical features of 293 children with neurodevelopmental disorders and/or multiple congenital anomalies. Ital J Pediatr. 2018;44(1):34.

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Kurtas N, Arrigoni F, **Errichiello E**, et al. Chromothripsis and ring chromosome 22: a paradigm of genomic complexity in the Phelan-McDermid syndrome (22q13 deletion syndrome). J Med Genet. 2018;55(4):269-277.

Cattaneo M, La Sala L, Rondinelli M, **Errichiello E**, Zuffardi O, Puca AA, Genovese S, Ceriello A. A donor splice site mutation in CISD2 generates multiple truncated, non-functional isoforms in Wolfram syndrome type 2 patients. BMC Med Genet;18(1):147.

Errichiello E, Mustafa N, Vetro A, Notarangelo LD, de Jonge H, Rinaldi B, Vergani D, Giglio SR, Morbini P, Zuffardi O. SMARCA4 inactivating mutations cause concomitant Coffin-Siris syndrome, microphthalmia and small-cell carcinoma of the ovary hypercalcaemic type. J Pathol. 2017;243(1):9-15.

Errichiello E, Venesio T. Mitochondrial DNA variants in colorectal carcinogenesis: Drivers or passengers? J Cancer Res Clin Oncol. 2017;143(10):1905-1914.

Errichiello E, Casati B, Zuffardi O. Patologia ereditaria da gain of function. Prospettive in Pediatria. 2017; 47(185): 1-9. ISSN 0301-3642.

Errichiello E, Vetro A, Mina T, Wischmeijer A, Berrino E, Carella M, Romagnoli M, Sacchini P, Venesio T, Zecca M, Zuffardi O. Whole exome sequencing in the differential diagnosis of Diamond-Blackfan anemia: clinical and molecular study of three patients with novel RPL5 and mosaic RPS19 mutations. Blood Cells, Molecules and Diseases. Blood Cells Mol Dis. 2017; 64:38-44.

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Errichiello E, Balsamo A, Cerni M, Venesio T. Mitochondrial variants in MT-CO2 and D-loop instability are involved in MUTYH-associated polyposis. J Mol Med (Berl). 2015;93(11):1271-81.

Renton AE, Pliner HA, Provenzano C, Evoli A, Ricciardi R, Nalls MA, Marangi G, Abramzon Y, Arepalli S, Chong S, Hernandez DG, Johnson JO, Bartoccioni E, Scuderi F, Maestri M, Gibbs JR, **Errichiello E**, Chiò A, Restagno G, Sabatelli M, Macek M, Scholz SW, Corse A, Chaudhry V, Benatar M, Barohn RJ, McVey A, Pasnoor M, Dimachkie MM, Rowin J, Kissel J, Freimer M, Kaminski HJ, Sanders DB, Lipscomb B, Massey JM, Chopra M, Howard JF Jr, Koopman WJ, Nicolle MW, Pascuzzi RM, Pestronk A, Wulf C, Florence J, Blackmore D, Soloway A, Siddiqi Z, Muppidi S, Wolfe G, Richman D, Mezei MM, Jiwa T, Oger J, Drachman DB, Traynor BJ. A genome-wide association study of myasthenia gravis. JAMA Neurol. 2015;72(4):396-404.

Venesio T, Balsamo A, **Errichiello E**, Ranzani GN, Risio M. Oxidative DNA damage drives carcinogenesis in MUTYH associated polyposis by specific mutations of mitochondrial and MAPK genes. Modern Pathology 2013; 26(10):1371-81.

Mereuta OM, Baldovino S, **Errichiello E**, Binello GB, Restagno G, Battaglia GG, Mazzucco G, Roccatello D. Systemic AA amyloidosis as a unique manifestation of a combined mutation of TNFRSF1A and MEFV genes. Amyloid 2013; 20(2):122-6.

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Gunetti M, Noghero A, Molla F, Staszewsky LI, de Angelis N, Soldo A, Russo I, **Errichiello E**, Frasson C, Rustichelli D, Ferrero I, Gualandris A, Berger M, Geuna M, Scacciatella P, Basso G, Marra S, Bussolino F, Latini R, Fagioli F. Ex vivo-expanded bone marrow CD34(+) for acute myocardial infarction treatment: in vitro and in vivo studies. Cytotheraphy 2011;13(9):1140-52.