

CURRICULUM VITAE

GAIA ROVERSI

PERSONAL DATA

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E-mail	gaia.roversi@unimib.it
citizenship	Italian
date and place of birth	08/02/74, Ferrara, Italy

EDUCATION AND WORK EXPERIENCE

- From October 2015, ASST-Monza, San Gerardo Hospital, Unit of Medical Genetics, **Genetic counselor**
- From November 2011, University of Milano-Bicocca- School of Medicine and Surgery, **Researcher** in Medical Genetics
- February 2009-October 2011, Fondazione IRCSS Istituto Nazionale dei Tumori, Milan, Unit of Medical Genetics, **Genetic counselor** for cancer predisposition syndrome
- November 2006-November 2008, University of Milan, San Paolo Hospital, Department of Medicine, Surgery and Dentistry, **Temporary Research Fellow**; assigned project :”The role of cancer stem cells in gliomagenesis”, supervision of Professor Lidia Larizza
- November 2004-November 2006, University of Milan, San Paolo Hospital, Department of Medicine, Surgery and Dentistry, **Temporary Research Fellow**; assigned project :” The WNT signaling pathway and its role in human brain tumors”, supervision of Professor Lidia Larizza
- 2000-2004 **Post Graduate School of Specialization in Medical Genetics** (University of Milan)
- 2000 **University Degree in Medicine and Surgery** (University of Milan).

SCIENTIFIC ACTIVITY

- Mutational targeted screening and genomic characterization by array CGH of **Thyroid Cancers**. Search for genes involved in cancer predisposition to Familial Non- Medullary Thyroid carcinoma (FNMTC) by exome sequencing.
- Genomic characterization by array CGH of **glioma** cell lines. Hunt for candidate gene involved in gliomagenesis. Setting of cancer stem cell lines from glioma cell lines and primary glioma.
- Genomic characterization by array CGH and FISH of MI-4 glioblastoma cell line. Cytogenomic characterization by array CGH of the **acute myeloid leukemia** Kasumi-1 cell line and its immunophenotypic subpopulations
- Molecular cloning, characterization and expression studies of **MARK4 gene** during both gliomagenesis and central nervous system development.
- Mutational screening and expression studies of the **RECQL4 gene**, involved in the chromosomal instability syndrome Rothmund-Thomson (RTS). Hunt for new candidate gene in **Rothmund-Thomson syndrome**
- Application of **chromosomal instability** tests: spontaneous and *aphidicolin* (APH) induced chromosomal breakage and cytokinesis-block micronucleous (CBMN) assay on peripheral blood and lymphoblastoid cell lines of RTS patients
- Identification of the disease gene involved in the **Clericuzio-type poikiloderma with neutropenia syndrome** by autozygosity mapping and linkage analysis of an inbred Italian family.
- Array CGH characterization and FISH mapping of interstitial deletions in **Cornelia de Lange** patients: hunt for candidate genes.
- Mutational screening of the **c-KIT oncogene** at somatic and constitutional level by automated sequencing in GIST tumors and in patients affected by piebaldism
- studies about expression and RNA editing of the hematopoietic cell **phosphatase PTPN6** patients affected by acute myeloid leukemia.

MAIN TECHNICAL SKILLS AND COMPETENCES:

Main technical skills and competence: Molecular biology in cancer genetics
Genetic oncological counseling

- **Conventional cytogenetics:** Metaphase Chromosome Spread Preparation. Karyotype analysis with QFQ banding. Induction of fragile sites in lymphocytes by aphidicolin.
- **Molecular cytogenetics:** one and dual color fluorescence in situ hybridization (FISH) of metaphase chromosomes or interphase nuclei. Oligonucleotide and BAC- based array comparative genomic hybridization (CGH).
- **Molecular Biology:** DNA and RNA extraction from whole blood, cultured cells, fresh tissues, Formalin-fixed, paraffin-embedded (FFPE) tissues. Plasmid and BAC DNA isolation by miniprep. Molecular cloning. PCR, RT-PCR, TaqMan Real Time PCR. Agarose and acrylamide electrophoresis. Mutational screening by automated sequencing, DDGE, CDGE, TGCE. Microsatellite Analysis, Taqman RT-PCR.
- **Cellular biology:** preparation of bacterial culture and cell culture (cancer cell lines, fibroblast, lymphoblastoid cell lines). Isolation and expansion of cancer stem cells from glioma cell lines. Cytokinesis-block micronucleus (CBMN) assay in human lymphocytes.
- **Bioinformatics:** Use of the most common bioinformatics tools (NCBI, UCSC, Ensemble, BLAST, splice site prediction websites)

AWARD: October 2009, Milan, “Nastro Rosa Estèe Lauder Companies” award organized by LILT (Lega Italiana per la Lotta contro i Tumori)

PUBLICATIONS

1. Imperatore V, Pinto AM, Gelli E, Trevisson E, Morbidoni V, Frullanti E, Hadjistilianou T, De Francesco S, Toti P, Gusson E, Roversi G, Accogli A, Capra V, Mencarelli MA, Renieri A, Ariani F. **Parent-of-origin effect of hypomorphic pathogenic variants and somatic mosaicism impact on phenotypic expression of retinoblastoma.** Eur J Hum Genet. 2018 Apr 17. [Epub ahead of print]
2. Smith A, Galli M, Piga I, Dentì V, Stella M, Chinello C, Fusco N, Leni D, Manzoni M, Roversi G, Garancini M, Pincelli AI, Cimino V, Capitoli G, Magni F, Pagni F. **Molecular signatures of medullary thyroid carcinoma by matrix-assisted laser desorption/ionisation mass spectrometry imaging.** J Proteomics. 2018 Mar 24. pii: S1874-3919(18)30122-2
3. Manzoni M, Roversi G, Di Bella C, Pincelli AI, Cimino V, Perotti M, Garancini M, Pagni F. **Solid cell nests of the thyroid gland: morphological, immunohistochemical and genetic features.** Histopathology. 2016 May;68(6):866-74.
4. Bentivegna A*, Roversi G*, et al: **The Effect of Culture on Human Bone Marrow Mesenchymal Stem Cells: Focus on DNA Methylation Profiles.** Stem Cells Int. 2016 Published online 2016 Jan 6.
5. La Verde N, Corsi F, Moretti A, Peissel B, Dalu D, Girelli S, Fasola C, Gambaro A, Roversi G, Azzollini J, Radice P, Pensotti V, Farina G, Manoukian S. **A targeted approach to genetic counseling in breast cancer patients: the experience of an Italian local project.** Tumori. 2016 Jan-Feb;102(1):45-50.
6. Roversi G. et al; **Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors.** Sci Rep. 2015 Oct 20;5:15454.
7. Peterlongo P. et al: **FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor.** Hum Mol Genet. 2015 Sep 15;24(18):5345-55.
8. Pagni F, L'Imperio V, Bono F, Garancini M, Roversi G, De Sio G, Galli M, Smith AJ, Chinello C, Magni F. **Proteome analysis in thyroid pathology.** Expert Rev Proteomics. 2015 Aug;12(4):375-90.
9. among collaborators: Kuchenbaecker KB. et al: **Identification of six new susceptibility loci for invasive epithelial ovarian cancer.** Nat Genet. 2015 Feb;47(2):164-71.
10. Kuchenbaecker KB, et al; **Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers.** Breast Cancer Res. 2014 Dec 31;16(6):3416.
11. Colombo EA, Fontana L, Roversi G, Negri G, Castiglia D, Paradisi M, Zambruno G, Larizza L.: **Novel physiological RECQL4 alternative transcript disclosed by molecular characterisation of Rothmund-Thomson Syndrome sibs with mild phenotype.** Eur J Hum Genet. 2014 Feb 12.
12. Mainini V, Pagni F, Garancini M, Giardini V, De Sio G, Cusi C, Arosio C, Roversi G, Chinello C,

- Caria P, Vanni R, Magni F.: **An Alternative Approach in Endocrine Pathology Research: MALDI-IMS in Papillary Thyroid Carcinoma.** *Endocr Pathol.* 2013 Dec;24(4):250-3.
13. Larizza L, Roversi G, Verloes A.: **Clinical utility gene card for: Rothmund-Thomson syndrome.** *Eur J Hum Genet.* 2013 Jul;21(7).
 14. Manoukian S, Peissel B, Frigerio S, Lecis D, Bartkova J, Roversi G, Radice P, Bartek J, Delia D. **Two new CHEK2 germ-line variants detected in breast cancer/sarcoma families negative for BRCA1, BRCA2, and TP53 gene mutations.** *Breast Cancer Res Tr.* 2011 Nov;130(1):207-15.
 15. Bonifaci N, Palafox M, Pellegrini P, Osorio A, Benítez J, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Roversi G, Barile M, Viel A, Mariette F, Bernard L, Radice P, Kaufman B, Laitman Y, Milgrom R, Friedman E, Sáez ME, Climent F, Soler MT, Diez O, Balmaña J, Lasa A, Ramón y Cajal T, Miramar MD, de la Hoya M, Pérez-Segura P, Caldés T, Moreno V, Urruticoechea A, Brunet J, Lázaro C, Blanco I, Pujana MA, González-Suárez E. **Evidence for a link between TNFRSF11A and risk of breast cancer.** *Breast Cancer Res Tr.* 2011 Oct;129(3):947-54.
 16. Martrat G, Maxwell CM, Tominaga E, Porta-de-la-Riva M, Bonifaci N, Gómez-Baldó L, Bogliolo M, Lázaro C, Blanco I, Brunet J, Aguilar H, Fernández-Rodríguez J, Seal S, Renwick A, Rahman N, Kühl J, Neveling K, Schindler D, Ramírez MJ, Castellà M, Hernández G; EMBRACE, Easton DF, Peock S, Cook M, Oliver CT, Frost D, Platte R, Evans DG, Lalloo F, Eeles R, Izatt L, Chu C, Davidson R, Ong KR, Cook J, Douglas F, Hodgson S, Brewer C, Morrison PJ, Porteous M, Peterlongo P, Manoukian S, Peissel B, Zaffaroni D, Roversi G, et al. **Exploring the link between MORF4L1 and risk of breast cancer.** *Breast Cancer Res.* 2011 Apr 5;13(2):R40.
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 18. Concolino D, Roversi G, Muzzi GL, Sestito S, Colombo EA, Volpi L, Larizza L, Strisciuglio P. **Clericuzio-type poikiloderma with neutropenia syndrome in three sibs with mutations in the C16orf57 gene: delineation of the phenotype.** *Am J Med Genet A.* 2010 Oct;152A(10):2588-94
 19. Catucci I, Verderio P, Pizzamiglio S, Manoukian S, Peissel B, Zaffaroni D, Roversi G, Ripamonti CB, Pasini B, Barile M, Viel A, Giannini G, Papi L, Varesco L, Martayan A, Riboni M, Volorio S, Radice P, Peterlongo P. **The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers.** *Breast Cancer Res Tr.* 2011 Feb;125(3):855-60
 20. Pedranzini L, Mottadelli F, Ronzoni S, Rossella F, Ferracin M, Magnani I, Roversi G, Colapietro P, Negrini M, Pelicci PG, Larizza L. **Differential cytogenomics and miRNA signature of the Acute Myeloid Leukaemia Kasumi-1 cell line CD34+38- compartment.** *Leukemia Res.* 2010 Oct;34(10):1287-95
 21. Larizza L, Roversi G, Volpi L. **Rothmund-Thomson syndrome.** *Orphanet J Rare Dis.* 2010 Jan 29;5:2. Review
 22. Volpi L*, Roversi G*, Colombo EA, Leijsten N, Concolino D, Calabria A, Mencarelli MA, Fimiani M, Macciardi F, Pfundt R, Schoenmakers EF, Larizza L.: **Targeted next-generation sequencing appoints c16orf57 as clericuzio-type poikiloderma with neutropenia gene.** *Am J Hum Genet.* 2010 Jan;86(1):72-6. Epub 2009 Dec 10. Erratum in: *Am J Hum Genet.* 2010 Sep 10;87(3):445
 23. Magnani I, Novielli C, Bellini M, Roversi G, Bello L, Larizza L.: **Multiple localization of endogenous MARK4L protein in human glioma.** *Cell Oncol.* 2009;31(5):357-70
 24. Gervasini C, Pfundt R, Castronovo P, Russo S, Roversi G, Masciadri M, Milani D, Zampino G, Selicorni A, Schoenmakers E, Larizza L.: **Search for genomic imbalances in a cohort of 24 Cornelia de Lange patients negative for mutations in the NIPBL and SMC1L1 genes.** *Clin Genet.* 2008 Dec;74(6):531-8
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 27. Larizza L, Magnani I, Roversi G.: **Rothmund-Thomson syndrome and RECQL4 defect: Splitting and lumping.** *Cancer Lett.* 2006 Jan 28;232(1):107-20. Review
 28. Roversi G, Pfundt R, Moroni RF, Magnani I, van Reijmersdal S, Pollo B, Straatman H, Larizza L, Schoenmakers EF. **Identification of novel genomic markers related to progression to glioblastoma through genomic profiling of 25 primary glioma cell lines.** *Oncogene.* 2006 Mar 9;25(10):1571-83

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30. Beghini A, Castorina P, Roversi G, Modiano P, Larizza L.: **RNA processing defects of the helicase gene RECQL4 in a compound heterozygous Rothmund-Thomson patient.** Am J Med Genet. A 2003 Jul 30;120A(3):395-9
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34. Beghini A, Ripamonti CB, Peterlongo P, Roversi G, Cairoli R, Morra E, Larizza L.: **RNA hyperediting and alternative splicing of hematopoietic cell phosphatase (PTPN6) gene in acute myeloid leukemia.** Hum Mol Genet. 2000 Sep 22;9(15):2297-304

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