CURRICULUM VITAE ET STUDIORUM

PERSONAL DATA:

Name:	Annalisa Frattini
Date and place of Birth:	27th, December 1962. Varese, Italy
Nationality:	Italian
Home address:	via alpestre, 22, 21100 Varese, Italy
Office address:	via Fantoli, 15/16,20090 Milan – Italy via JH Dunant, 5,21100 Varese - Italy
tel. (office)	+39 0332 217113
fax. (office)	+39 0332 397119
e-mail:	annalisa.frattini@irgb.cnr.it

EDUCATIONAL TRAINING:

1983:	High School Diploma in Humanities, Institute "E. Cairoli" Varese.
1989:	Degree in Biological Science, University of Milan.
1994:	Specialization Diploma in Biotechnological Applications, University of Milan.

RESEARCH AND PROFESSIONAL EXPERIENCE:

1987 - 1989:	Internship in Entomology Department of European Community Center of Ispra, Varese, under Prof. R. Cavalloro.
1989 - 1994:	Internship in Human Genome Unit at Institute of Advanced Biomedical Technologies (ITBA), in Milan, under Dr. P. Vezzoni.
1995 - 2008*:*	Researcher at the Human Genome Department of the Institute of Biomedical Technologies (ITB) of National Council of Research, in Milan.
2009 - at present:	Researcher at Institute of Genetic and Biomedical Research (IRGB) of National Council of Research. Collaboration with the Department of Medicine (DMC), University of Insubria, Varese, Italy.
2014:	National scientific license in Applied Biology

Initially, I was involved in the Italian Human Genome Project and my interests were devoted to investigating the genes located in the long arm of the human X chromosome. During this period, I also developed know-how in bioinformatics. I have contributed to identifying the genes involved in two diseases: the autosomal form of T-B+ Severe Combined Immunodeficiency (T-B+ SCID) and Omenn Syndrome.

We have identified the first gene responsible for a subset of patients affected by autosomal recessive osteopetrosis, (OP) a severe bone disease with a fatal outcome. We have demonstrated that the subunit a3 of the V-ATPase complex, coded by TCIRG1 gene (also called ATP6i), is defective in a subset of patients affected by OPTB1. This result points to the acidification of the extracellular microenvironment as one of the more sensitive steps of bone resorption by osteoclasts and opens the way to early diagnosis and, possibly, to therapy of this disease. We have also clarified the role of the chloride channel 7 (a CLC Cl^-/H^+ antiporter coded by the ClCN7 gene) as responsible for a subset of the recessive form of osteopetrosis (OPTB 4) with primary neural defect and of the intermediate form of OP. We have also identified the third gene responsible for recessive osteopetrosis (OPTB5), the OSMT1 gene. OSTM1 is mutated in few OP patients with severe central nervous system involvement leading to a very poor prognosis. We have analyzed more than 130 cases of recessive OP, about 30% of them do not show mutations in these three genes, so we are looking for the new responsible gene/genes with wide genome scan in consanguineous families and with the candidate gene approach.

As the only available treatment of the severe form of osteopetrosis is bone marrow transplantation, which usually does not cure all the effects of the disease, we have used an *in utero* transplant approach in the *oc/oc* mouse model with the aim of curing all the stigmata of the disease. Our results clearly demonstrated that in utero hematopoietic stem cell transplantation can prevent fatal defects of osteopetrosis and that this approach may be directly applicable to humans.

In addition, we used this approach in the murine model BrtlIV that reproduces the molecular, biochemical, and radiographic features of osteogenesis imperfecta type IV, to test the efficacy of the *in utero* mesenchymal stem cell transplantation.

We identified the gene responsible for a form of osteopetrosis (OPTB2) due to the lack of mature osteoclasts, the osteoclast-poor osteopetrosis. The gene codes for RANKL (the TNF ligand family, receptor activator of nuclear factor-KAPPAB ligand; TNFSF11) the key molecule for the differentiation of the osteoclasts from precursor cells in the myeloid lineage.

Mutations in this gene prevent the interaction of the RANKL with its receptor RANK present in the plasmatic membrane of the osteoclasts precursors, with the consequence of the lack of the activation of the specific gene involved in the cellular differentiation.
Moreover, we demonstrated that mutations also in the RANK gene are responsible for a subset of patients affected by osteoclast-poor osteopetrosis (OPTB7) characterized by hypogammaglobulinemia.
In collaboration with Prof. Amling (Center for Biomechanics, Experimental Trauma Surgery & Skeletal Biology; Department of Trauma, Hand, and Reconstructive Surgery, University Medical Center Hamburg-Eppendorf, Hamburg, Germany), the study of murine models of osteopetrosis (tcirg1$^{-/-}$ and src$^{-/-}$) defined a new relation between bone metabolism and the uptake of calcium at gastric level.
In collaboration with the Human Genomic group of at the Department of Clinical Medicine, University of Insubria, Varese, we evaluated the CNVs (Copy Number Variations) in patients affected by the dominant form of osteopetrosis (OPTB4) and in patients affected by olfactory neuroblastoma.
We analyzed the genomic profile of different batches of HeLa cell lines through the analysis of the karyotypes and a-CGH and their expression profiles. Our results underlined that the high genomic instability of different of HeLa clones invalidate their gene expression profiling.
We analyzed the data of the exome sequencing of a patient affected by neutropenia and bone marrow chromosomes instability.
In collaboration with the Department of Biotechnologies and Life Science, University of Insubria, Varese we evaluated the paracrine effects of human adipocytes in the differentiation of LECs (lymphatic endothelial cells) obtained from rat diaphragm.

Actually, in collaboration with Dr. R. Valli (DMC, University of Insubria), and Dr. I. Villa (Bone Metabolism Unit, San Raffaele Scientific Institute, Milan) we are defining the molecular pathways of the bone defects in patients affected by Schwachman-Diamond syndrome (SDS, OMIM: 260400), a rare monogenic inherited multisystemic disorder principally characterized by exocrine pancreatic dysfunction, skeletal defects characterized by metaphyseal and thoracic dysplasia, early onset of low bone turnover osteoporosis, and bone marrow failure, with the predisposition toward myelodysplasia syndrome or acute myeloid leukaemia.
At the same time, we are evaluating the effect of drugs able to bypass nonsense mutations (carried by about 90% of SDS patients) in lymphoblastic cell lines and in primary cultures of osteoblasts derived from bone biopsies of SDS patients.

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