SCIENTIFIC PROFILE

My scientific activity is mainly in the field of translational medicine with a strong mission towards precision medicine. My research activity is focused on red cell pathologies, normal and pathologic erythropoiesis, hemoglobinopathies such as β-thalassemic syndromes and sickle cell disease (SCD) and hereditary rare anemias (i.e.: stomatocytosis, CDAII). An additional area of investigation is chorea-acanthocytosis and McLeod syndrome, which are neurodegenerative disorders involving also the erythroid compartment (Blood 118: 5652-63, 2011; Blood 128: 2976, 2016; J Neurosci 36: 12027, 2016). Through highly productive collaborations and keywork in my laboratory, my research has resulted in a number of breakthrough technologies and discoveries.

- My original finding of Src tyrosine kinase in normal and pathologic red cells has opened new field of investigation in red cell pathologies and diseased erythropoiesis, facilitating characterization of the functional interplay between iron and erythropoiesis (i.e.: J. Clin. Invest. 99:220-227, 1997; Hematologica 82: 648-653, 1997; Blood 118: 5652-63, 2011; PlosOne May 1 10: e0125580, 2012; Cell Rep: 411-21, 2016; Haematologica 96: 1595, 2011; Haematologica 91: 1336, 2006; Blood 86: 4050, 1995; ARS 2015-2018).
- My major novel contributions in field of thalassemic syndromes and erythropoiesis include identification of the role of oxidative stress in thalassemia and stress erythropoiesis such as in response to abnormalities in iron homeostasis (i.e. Cell Rep: 411-21, 2016; Haematologica 96: 1595, 2011; Haematologica 91: 1336, 2006; Blood 86: 4050, 1995; Blood 84: 315, 1994).
- I have established novel methodologies for erythroid profiling and sorting that allow for the first time, to study highly purified erythroid precursors and identified new cytoprotective pathways involved in pathological erythropoiesis (i.e.: Haematologica 92:1319-1326, 2007; Haematologica 94: 1049-1059, 2009; Haematologica 96: 1595-604, 2011; Blood 121: e43-49, 2013; Haematologica. 99: 267-75, 2014). An international collaboration allowed us to develop new possible therapeutic approach with PEP1-fusion protein Prx2 (ARS 23: 1284, 2015).
- I have also contributed to the progress in phosphoproteomic analysis of erythroid cells, using β-thalassemia as disease model (i.e.: Blood 118: 5652-63, 2011; Proteomics, 8: 4695- 708, 2008; PLoS One 7: e31015, 2012).
- My major novel contributions in field of SCD are: (i) the identification of new molecules blocking red cell dehydration in SCD transfer to clinical use such as senicapoc (phase II-phase III trial), Mg-pidolate (patent), calpain 1 inhibitor (under pre-clinical development), hemopexin (Circulation 127: 1317-29, 2013). (ii) the identification of novel mechanisms involved in SCD red cell functional crosstalk with microcirculation (J. Clin. Invest. 92: 520-526, 1993 and J. Clin. Invest. 93: 1670-1676, 1994; Blood 87: 1186-1195, 1996; Blood 88: 2738-2744, 1996; J. Clin. Invest. 100: 1847-1852, 1997; Blood 94: 4307-4313,1999; Blood 97: 1451-1457, 2001; Blood 101: 2412-2418, 2003; FASEB J 2012 Oct 19 doi: fj 12-217836); (iii) models of SCA related organ damages with generation of mouse model mimicking organ damage of SCD human counterpart (Blood 101: 2412-2418, 2003; FASEBJ, 22: 1849-1860, 2008; J Clin Invest, 118: 1924-1933, 2008; Haematologica 96: 24-32,

2011; Haematologica May 1 doi: 2015.124586, 2015); (iv) identification of the mechanisms involved in bone disease related to SCD (Blood 126: 2320, 2016).

- My major novel contributions in field of rare anemia. I have identified and contributed to the identification of rare anemias and to their functional characterization (i.e.: Blood 106: 4359-66, 2005, Haematologica 94: 1049-1059, 2009; Am J Hematology 88: 66-78, 2013; Blood 121: 3925-35, 2013; Haematologica Epub 2016.142372, 2016).