CURRICULUM VITAE Dr CRISTINA SOBACCHI

Education and Training
1998. Training in Molecular Biology, Laboratory of Neuroendocrinology, University of Milan
1993-1998. Degree in Pharmaceutical Chemistry and Technologies at the University of Milan

Research and Professional experience
2010-Present. Permanent position at CNR, Institute of Genetic and Biomedical Research (IRGB)
2007-2009. Researcher at CNR, ITB
2005−2007. Post-doc at CNR, ITB in the lab directed by Dr Anna Villa
2001−2003. FIRC Fellowship in the lab directed by Dr Anna Villa on the topic: "Molecular and cellular mechanisms controlling antigen receptor rearrangement and potentially involved in the pathogenesis of lymphoid tumors".
1999−2000. Fellowship in the lab directed by Dr Anna Villa on the topic: Development and application of technologies of industrial interest in the field of genomics (Biosearch Italia S.p.A.)
1998−1999. Fellowship in Molecular Biology, CNR, in the lab directed by Dr Anna Villa, on the topic: Genetic analysis of primary immunodeficiencies.

PATENT
Patent N. 03737357.8-2402-GB0300470. Genetic markers for bone mass- The University Court of the University Aberdeen (Scotland). Date of filing: 04.02.03

AFFILIATIONS TO SCIENTIFIC SOCIETIES AND ACTIVITIES
Member of the European Calcified Tissues Society (ECTS); Member of the ECTS Training Committee (from November 2012). Member of the Forum in Bone and Mineral Research. From October 2013: Member of Ethic Committee Associazione "La Nostra Famiglia", IRCCS Eugenio Medea (as the expert in genetics). Local Organizer of the ECTS PhD Training Course 2015. Member of the Scientific Program Committee for the ECTS 2021 Annual Congress.

HONOURS
2007. Young Investigator Award, from the European Calcified Tissues Society
2009. ECTS/ABBH International Iain Boyle Award

Research interests:
Dr Cristina Sobacchi has been working at CNR for more than 20 years, being actively involved in the study of genetic diseases, in particular primary immunodeficiencies and rare and common bone diseases (such as osteopetrosis and osteoporosis). Since several years, she is an independent researcher, as shown by the number of publications on peer-reviewed journals as senior author and grants awarded. Her main interest is in bone biology, with particular respect to the molecular and cellular mechanisms involved in the pathogenesis of rare and common skeletal diseases and the development of new therapeutic strategies in animal models, by using pharmacological approaches, cell-based approaches (MSC, iPSc) as well as gene editing and gene therapy approaches. She has also been involved in the definition and periodical review of the Consensus guidelines for diagnosis, therapy and follow-up of Osteopetrosis, on behalf of the ESID and the EBMT WP Inborn Errors, as Genetic Consultant.
Dr. Cristina Sobacchi is co-author of total **69** peer-reviewed publications on International Journals in which she features mainly as first and last author (h index: **24**, Source: Scopus) and 3 Book Chapters.

GRANTS
Current Grants:
Ministero della Salute, Ricerca finalizzata ordinaria - "Theory Enhancing". Project RF-2018-12367680: "Dissecting the neglected link between RANKL cytokine and stemness features with high relevance in osteoporosis" Role: Principal Investigator. 3 Years Grant. Founded €450.000

Telethon Rare Diseases and COVID-19, Grant n. GSP20006Covid025: "Assessing the role of the gene associated with Acrofrontofacionasal Dysostosis type 1, NBAS, in the nonsense mediated decay and Golgi-to-ER retrograde transport functions in Sars-CoV-2 infected cells". Role: Principal Investigator. 1 Year Grant. Founded €50.000

Past Grants:
Telethon Grant n.GGP07059: Characterization of the molecular basis of Osteopetrosis: the osteoclast−poor form. Role: Principal Investigator. 1 Year Grant. Founded €40.500

Telethon Grant n.GGP08176: Understanding and treating osteoclast-poor autosomal recessive osteopetrosis: RANKL-RANK axis. Role: Principal Investigator. 2 Years Grant, founded €120.000

Fondazione Cariplo Grant n.2008-2218: Understanding and treating osteoclast-poor autosomal recessive osteopetrosis: RANKL-RANK axis. Role: Principal Investigator. 3 Years Grant, co-founded €232.999,94

"Giovani Ricercatori" Grant GR-2007-686104: HLA-G: a new tolerogenic marker for tolerance induction mediated by regulatory T cells. Role: Head of Unit. 3 Years Grant, founded €150.000

"Giovani Ricercatori" Grant GR-2008-1134625: New therapeutical approaches to RANKL-dependent Autosomal Recessive Osteopetrosis. Role: Principal Investigator.

Telethon Grant n.GGP10116: A pharmacological approach to RANKL-dependent osteoclast-poor Autosomal Recessive Osteopetrosis. Role: Principal Investigator. 2 Years Grant, founded €138.000

Telethon Grant n.GGP12178: Mesenchymal Stem Cell Transplantation as a therapeutic approach to RANKL-dependent Osteopetrosis. Role: Principal Investigator. 2 Years Grant, founded €123.000

Telethon Exploratory Project n.GGP13060: Understanding the genetic basis of Acrofrontofacionasal Dysostosis 1. Role: Principal Investigator. 1 Year Grant, founded €21.350

"Giovani Ricercatori" Grant: Exome sequencing of osteoimmunological diseases: from diagnosis to cure of osteopetrosis and other genetic skeletal diseases. Role: Principal Investigator. 3 Years Grant. Founded €225.987,76

INVITED TALKS

Convegno Scientifico Nazionale "FOCUS ON: OSTEOPOROSI ED ENDOCRINOPATIE - Approccio multidisciplinare per la prevenzione ed il trattamento delle fratture secondarie ad osteoporosi", March 8-9, 2012 - Ancona, Italy. Invited talk: "Forme genetiche con aumento della massa ossea: osteopetrosi ed altre forme"

16º convegno "Patologia immune e malattie orfane" January 17-19, 2013 - Turin, Italy. Invited talk: "Osteopetrosi"

ECTS PhD Training Course September 15-18, 2013 - Hamburg, Germany. Invited talk: "Bone cells: osteoclasts".

5th International Conference on Osteoimmunology: Interactions of the Immune and Skeletal Systems, June 15-20, 2014 – KOS, Greece. Invited talk: "New insights on bone biology from exome sequencing of osteopetrotic patients with atypical presentations"

ECTS PhD Training Course September 13-16, 2015 - Siena, Italy. Local organizer and invited speaker. "Rare bone diseases"

XV Congresso Nazionale SIOMMMS, Basic and Translational Research Symposium, November 12th, 2015 - Bologna, Italy. Invited talk: "Osteopetrosis: diagnosis, prognosis, treatment"

16th Forum in Bone and Mineral Research Meeting, June 15-16, 2016 - Turin, Italy. Invited talk: "Iron metabolism and bone"

ECTS PhD Training Course July 8-11, 2017 - Paris, France. Invited talk: "Bone phenotype and applications to rare diseases".

"Osteoclasts and Bone Resorption in Health and Disease" Meeting, Weizmann Institute of Science, Rehovot, Israel; June 10-12, 2018. Invited talk: "Genetics of Osteopetrosis".

Convegno Auxologico Focus on: Interrelazioni tra syndrome metabolica e fragilità scheletrica: dalla Ricerca di base alle problematiche cliniche. October 11, 2018, Milan. Invited talk: "Stress ossidativo e tessuto osseo".

XVIII Congresso Nazionale SIOMMMS, Basic and Translational Research Symposium, October 26th, 2018 - Naples, Italy. Invited talk: "Osteopetrosis: from gene to therapy"

Master in Medicina Rigenerativa, Humanitas University, Rozzano (MI), Italy, January 16th, 2019. Invited talk: Immunologia e medicina rigenerativa - Ricerca di base.

46th ECTS Congress, Budapest, Hungary, May 11-14, 2019; pre-congress Friday 10 May 2019. Invited talk: “Genetics of Osteopetrosis and Osteoclast Biology”

9th International Conference on Children's Bone Health, Salzburg, Austria, June 22-25, 2019. Invited talk: “Osteoclast disorders”

PUBLICATIONS

1. Santagata S, Besmer E, Villa A, Bozzi F, Allingham JS, Sobacchi C, Hainford DB, Vezzoni P, Nussenzweig MC, Pan ZQ, Cortes P. The RAG1/RAG2 Complex Constitutes a 3’Flap Endonuclease: Implications for junctional diversity in V(D)J and transpositional recombination. Mol Cell 4:1-20, 1999.

2. Villa A, Bozzi F, Sobacchi C, Strina D, Fasth A, Pasic S, Notarangelo LD, Vezzoni P. Prenatal diagnosis of RAG-deficient Omenn syndrome. Prenatal Diagnosis 20:56-59, 2000.

3. Frattini A, Orchard PJ, Sobacchi C, Giliani S, Abinun M, Mattsson JP, Keeling DJ, Andersson AK, Wallbrandt P, Zecca L, Notarangelo LD, Vezzoni P and Villa A. Defects in the TCIRG1-encoded 116kD subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genet 25:343-346, 2000.

4. Gomez CA, Ptaszek LM, Villa A, Bozzi A, Sobacchi C, Brooks EG, Notarangelo LD, Spanopoulou E, Pan ZQ, Vezzoni P, Cortes P and Santagata S. Mutations in conserved regions of the predicted RAG2 kelch repeats block initiation of V(D)J recombination and result in primary immunodeficiencies. Mol Cell Biol 20: 5653-5664, 2000.

5. Santagata S, Villa A, Sobacchi C, Cortes P, Vezzoni P. The genetical and biochemical basis of Omenn Syndrome. Immunol Reviews 178:64-74, 2000.

6. Santagata S, Gomez CA, Sobacchi C, Bozzi F, Abinun M, Pasic S, Cortes P, Vezzoni P, Villa A. N-terminal RAG1 frameshift mutations in Omenn syndrome: internal methionine usage leads to partial V(D)J recombination activity and reveals a fundamental role in vivo for the N-terminal domains. Proc Natl Acad Sci U S A 20:5653-5664, 2000.

7. Villa A, Sobacchi C, Notarangelo LD, Bozzi F, Abinun M, Abrahamsen TG, Arkwright PD, Baniyash M, Brooks EG, Conley ME, Cortes P, Duse M, Fasth A, Filipovich AM, Infante AJ, Jones A, Mazzolari E, Muller SM, Pasic S, Rechavi G, Sacco MG, Santagata S, Schroeder ML, Seger R, Strina D, Ugazio U, Väliaho J, Vihinen M, Vogler LB, Ochs H, Vezzoni P, Friedrich W, Schwarz K. V(D)J recombination defects in lymphocytes due to RAG mutations: a severe immunodeficiency with a spectrum of clinical presentations. Blood 97:81-88, 2001.

8. Villa A, Sobacchi C, Vezzoni P. Recombination activating gene and its defects. Curr Opin Allergy Clin Immunol 1:491-295, 2001.

9. Sobacchi C, Frattini A, Orchard P, Porras O, Tezcan I, Andolina M, Babul-Hirji R, Baric I, Canham N, Chitayat D, Dupuis-Girod S, Ellis I, Etzioni A, Fasth A, Fisher A, Gerritsen B, Gulino V, Horwitz E, Klamroth V, Lanino E, Mirolo M, Musio A, Matthijs G, Nonomaya S, Notarangelo LD, Ochs HD, Superti Furga A, Valiaho J, van Hove JL, Vihinen M, Vujic D, Vezzoni P, Villa A. The mutational spectrum of human malignant autosomal recessive osteopetrosis. Hum Mol Genet 10:1767-1773, 2001.

10. Villa A, Sobacchi C, Vezzoni P. Omenn Syndrome in the context of other B cell negative-severe combined immunodeficiencies. Isr Med Assoc J 4:218-221, 2002.

11. Frattini A, Pangrazio A, Susani L, Sobacchi C, Mirolo M, Abinum M, Andolina M, Flanagan A, Horwitz EM, Mihci E, Notarangelo LD, Ramenghi U, Teti A, Van Hove1 J, Vujic D, Young T, Albertini A, Orchard PJ, Vezzoni P and Villa A. Chloride channel ClCN7 mutations are responsible for severe recessive, dominant and intermediate osteopetrosis. J Bone Miner Res 18:1740-1747, 2003.

12. Sobacchi C, Vezzoni P, Reid DM, McGuigan FEA, Frattini A, Mirolo M, Albagha OME, Musio A, Villa A, Ralston SH. Association between a polymorphism affecting an AP1 binding site in the promoter of the TCIRG1 gene and bone mass in women. Calcif Tissue Int 74:35-41, 2004.

13. Susani L, Pangrazio A, Sobacchi C, Taranta A, Mortier G, Savarirayan R, Villa A, Orchard P, Vezzoni P, Albertini A, Frattini A, Pagani F. TCIRG1-dependent recessive osteopetrosis: mutation analysis, functional identification of the splicing defects, and in vitro rescue by U1 snRNA. Hum Mutat 24:225-235, 2004.

14. Musio A, Marrella V, Sobacchi C, Rucci F, Fariselli L, Giliani S, Lanzi G, Notarangelo LD, Delia D, Colombo R, Vezzoni P, Villa A. Damaging-agent sensitivity of Artemis-deficient cell lines. Eur J Immunol 35:1250-1256, 2005.

15. Frattini A, Blair HC, Sacco MG, Cerisoli F, Faggioli F, Cato EM, Pangrazio A, Musio A, Rucci F, Sobacchi C, Sharrow AC, Kalla SE, Bruzzone MG, Colombo R, Magli MC, Vezzoni P, Villa A. Rescue of ATPa3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation in utero. Proc Natl Acad Sci U S A. 102:14629-14634, 2005.

16. Rucci F, Cattaneo L, Marrella V, Sacco MG, Sobacchi C, Lucchini F, Nicola S, Della Bella S, Villa ML, Imberti L, Gentili F, Montagna C, Tiveron C, Tatangelo L, Facchetti F, Vezzoni P, Villa A. Tissue-specific sensitivity to AID expression in transgenic mouse models. Gene 377:150-158, 2006.

17. Sobacchi C, Marrella V, Rucci F, Vezzoni P, Villa A. RAG-dependent primary immunodeficiencies. Hum Mut 27:1174-1184, 2006.

18. Sobacchi C, Frattini A, Guerrini M, Abinun M, Pangrazio A, Susani L, Bredius R, Mancini G, Cant A, Bishop N, Grabowski P, Del Fattore A, Messina C, Errigo G, Coxon FP, Scott DI, Teti A, Rogers MJ, Vezzoni P, Villa A, Helfrich MH. Osteoclast-poor human osteopetrosis due to mutations in RANKL. Nature Genet 39:960-962, 2007.

19. Frattini A, Vezzoni P, Villa A, Sobacchi C. The dissection of human autosomal recessive osteopetrosis identifies an osteoclast-poor form due to RANKL deficiency. Cell Cycle 6:3027-3033, 2007.

20. Matangkasombut P, Pichavant M, Saez DE, Giliani S, Mazzolari E, Finocchi A, Villa A, Sobacchi C, Cortes P, Umetsu DT, Notarangelo LD. Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. Blood 111:271-274, 2008.

21. Marrella V, Poliani PL, Sobacchi C, Grassi F, Villa A. Of Omenn and mice. Trends Immunol 29:133-140, 2008.

22. Guerrini MM, Sobacchi C, Cassani B, Abinun M, Kilic SS, Pangrazio A, Moratto D, Mazzolari E, Clayton-Smith J, Orchard P, Coxon FP, Helfrich MH, Crockett JC, Mellis D, Vellodi A, Tezcan I, Notarangelo LD, Rogers MJ, Vezzoni P, Villa A, Frattini A. Human osteoclast-poor osteopetrosis with hypogammaglobulinemia due to TNFRSF11A (RANK) mutations. Am J Hum Genet 83:64-76, 2008.

23. Pangrazio A, Caldana ME, Sobacchi C, Panaroni C, Susani L, Mihci E, Cavaliere ML, Giliani S, Villa A and Frattini A. Characterization of a novel Alu-Alu recombination-mediated genomic deletion in the TCIRG1 gene in five osteopetrotic patients. J Bone Miner Res 24:162-167, 2009.

24. Villa A, Pangrazio A, Caldana E, Guerrini M, Vezzoni P, Frattini A, Sobacchi C. Prognostic potential of precise molecular diagnosis of Autosomal Recessive Osteopetrosis with respect to the outcome of bone marrow transplantation. Cytotechnology 58:57-62, 2008.

25. Villa A, Guerrini MM, Cassani B, Pangrazio A, Sobacchi C. Infantile malignant, autosomal recessive osteopetrosis: the rich and the poor. Calcif Tissue Int 84:1-12, 2009.

26. Pangrazio A, Pusch M, Caldana E, Frattini A, Lanino E, Tamhankar PM, Phadke S, Lopez AG, Orchard P, Mihci E, Abinun M, Wright M, Vettenranta K, Bariae I, Melis D, Tezcan I, Baumann C, Locatelli F, Zecca M, Horwitz E, Mansour LS, Van Roij M, Vezzoni P, Villa A, Sobacchi C. Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Hum Mutat 31:E1071-80, 2010.

27. Cassani B, Poliani PL, Moratto D, Sobacchi C, Marrella V, Imperatori L, Vairo D, Plebani A, Giliani S, Vezzoni P, Facchetti F, Porta F, Notarangelo LD, Villa A, Badolato R. Defect of regulatory T cells in patients with Omenn syndrome. J Allergy Clin Immunol 125:209-216, 2010.

28. Prontera P, Rogaia D, Sobacchi C, Tavares VL, Mazzotta G, Passos-Bueno MR, Donti E. Craniometaphyseal dysplasia with severe craniofacial involvement shows homozygosity at 6q21-22.1 locus. Am J Med Genet A 155:1106-1108, 2011.

29. Pangrazio A, Boudin E, Piters E, Damante G, Iacono NL, D'Elia AV, Vezzoni P, Van Hul W, Villa A, Sobacchi C. Identification of the first deletion in the LRP5 gene in a patient with Autosomal Dominant Osteopetrosis type I. Bone 49:568-571, 2011.

30. Sfaihi L, Aissa K, Sobacchi C, Kamoun F, Gorbel M, Hachicha M. Choanal atresia: Think about osteopetrosis. International Journal of Pediatric Otorhinolaryngology Extra 6:422-424, 2011.

31. Pangrazio A, Cassani B, Guerrini MM, Crockett JC, Marrella V, Zammataro L, Strina D, Schulz A, Schlack C, Kornak U, Mellis DJ, Duthie A, Helfrich MH, Durandy A, Moshous D, Vellodi A, Chiesa R, Veys P, Iacono NL, Vezzoni P, Fischer A, Villa A, Sobacchi C. RANK-dependent autosomal recessive osteopetrosis: characterisation of 5 new cases with novel mutations. J Bone Miner Res 27:342-351, 2012.

32. Pangrazio A, Caldana ME, Lo Iacono N, Mantero S, Vezzoni P, Villa A, Sobacchi C. Autosomal recessive osteopetrosis: report of 41 novel mutations in the TCIRG1 gene and diagnostic implications. Osteoporosis Int 23:2713-2718, 2012.

33. Pangrazio A, Frattini A, Valli R, Maserati E, Susani L, Vezzoni P, Villa A, Al-Herz W, Sobacchi C. A homozygous contiguous gene deletion in chromosome 16p13.3 leads to autosomal recessive osteopetrosis in a Jordanian patient. Calcif Tissue Int 91:250-254, 2012.

34. Lo Iacono N, Blair HC, Poliani PL, Marrella V, Ficara F, Cassani B, Facchetti F, Fontana E, Guerrini MM, Traggiai E, Schena F, Paulis M, Mantero1 S, Inforzato A, Valaperta S, Pangrazio A, Crisafulli L, Maina V, Kostenuik P, Vezzoni P, Villa A, Sobacchi C. Osteopetrosis rescue upon RANKL administration to Rankl-/- mice: a new therapy for human RANKL-dependent ARO. J Bone Miner Res 27:2501-2510, 2012.

35. Mégarbané A, Pangrazio A, Villa A, Chouery E, Maarawi J, Sabbagh S, Lefranc G, Sobacchi C. Homozygous stop mutation in the SNX10 gene in a consanguineous Iraqi boy with osteopetrosis and corpus callosum hypoplasia. Eur J Med Genet 56:32-35, 2013.

36. Pangrazio A, Fasth A, Sbardellati A, Orchard PJ, Kasow KA, Raza J, Albayrak C, Albayrak D, Vanakker OM, De Moerloose B, Vellodi A, Notarangelo LD, Schlack C, Strauss G, Kühl J-S, Caldana E, Lo Iacono N, Susani L, Kornak U, Schulz A, Vezzoni P, Villa A, Sobacchi C. SNX10 mutations define a subgroup of human Autosomal Recessive Osteopetrosis with variable clinical severity. J Bone Miner Res 28:1041-1049, 2013.

37. Lo Iacono N, Pangrazio A, Abinun M, Bredius R, Zecca M, Blair HC, Vezzoni P, Villa A, Sobacchi C. RANKL Cytokine: From Pioneer of the Osteoimmunology Era to Cure for a Rare Disease. Clin Dev Immunol 2013:412768, 2013.

38. Sobacchi C, Schulz A, Coxon FP, Villa A, Helfrich MH. Human Osteopetrosis: genetics, treatment and new insights into osteoclast function. Nat Rev Endocrinol 9:522-536, 2013.

39. Pangrazio A, Puddu A, Oppo M, Valentini M, Zammataro L, Vellodi A, Gener B, Llano-Rivas I, Raza J, Atta I, Vezzoni P, Superti-Furga A, Villa A, Sobacchi C. Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 59C:122-126, 2013.

40. Pasic S, Vujic D, Veljković D, Slavkovic B, Mostarica-Stojkovic M, Minic P, Minic A, Ristic G, Giliani S, Villa A, Sobacchi C, Lilić D, Abinun M.Severe Combined Immunodeficiency in Serbia and Montenegro Between Years 1986 and 2010: A Single-Center Experience. J Clin Immunol 34:304-308, 2014.

41. Sobacchi C, Pangrazio A, López AG, Gómez DP, Caldana ME, Susani L, Vezzoni P, Villa A. As Little as Needed: The Extraordinary Case of a Mild Recessive Osteopetrosis Due to a Novel Splicing Hypomorphic Mutation in the TCIRG1 Gene. J Bone Miner Res 29:1646-1650, 2014.

42. Gannagé-Yared MH, Makrythanasis P, Chouery E, Sobacchi C, Mehawej C, Santoni FA, Guipponi M, Antonarakis SE, Hamamy H, Mégarbané A. Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia. Bone 68:142-145, 2014.

43. Palagano E, Blair HC, Pangrazio A, Tourkova I, Strina D, Angius A, Cuccuru G, Oppo M, Uva P, Van Hul W, Boudin E, Superti-Furga A, Faletra F, Nocerino A, Ferrar MC, Grappiolo G, Monari M, Montanelli A, Vezzoni P, Villa A, Sobacchi C. Buried in the middle, but guilty: intronic mutations in the TCIRG1 gene cause human Autosomal Recessive Osteopetrosis. J Bone Miner Res 30:1814-1821, 2015.

44. Neri T, Muggeo S, Paulis M, Caldana ME, Crisafulli L, Strina D, Focarelli ML, Faggioli F, Recordati C, Scaramuzza S, Scanziani E, Mantero S, Buracchi C, Sobacchi C, Lombardo A, Naldini L, Vezzoni P, Villa A, Ficara F. Targeted gene correction in osteopetrotic induced pluripotent stem cells for the generation of functional osteoclasts. Stem Cell Rep 5:558-568, 2015.

45. Marrella V, Lo Iacono N, Fontana E, Sobacchi C, Sic H, Schena F, Castiello MC; Cassani B, Poliani PL, Vezzoni P, Traggiai E, Villa A. Expansion of IL-10 secreting B cells counteracts the autoimmunity propensity of Rankl-/- mice. J Immunol 194:4144-4153, 2015. Endocrinol 2015:265151, 2015.

46. Rauner M, Coudert A, Sobacchi C, Del Fattore A. The endocrine role of the skeleton. Int J Endocrinol 2015:265151, 2015.

47. Sobacchi C, Villa A, Schulz A, Kornak U. CLCN7-Related Osteopetrosis. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016. 2007 Feb 12 [updated 2016 Jun 9].

48. Palagano E, Susani L, Menale C, Ramenghi U, Berger M, Uva P, Oppo M, Vezzoni P, Villa A, Sobacchi C. Synonymous Mutations Add a Layer of Complexity in the Diagnosis of Human Osteopetrosis. J Bone Miner Res. 2016 Jul 28. doi: 10.1002/jbmr.2929. [Epub ahead of print]

49. Lleo A, Bian Z, Zhang H, Miao Q, Yang F, Peng Y, Chen X, Tang R, Wang Q, Qiu D, Fang J, Sobacchi C, Villa A, Di Tommaso L, Roncalli M, Gershwin ME, Ma X, Invernizzi P. Quantitation of the Rank-Rankl Axis in Primary Biliary Cholangitis. PLoS One. 2016 Sep 15;11(9):e0159612.

50. Weisz Hubshman M, Basel-Vanagaite L, Krauss A, Konen O, Levy Y, Garty BZ, Smirin-Yosef P, Maya I, Lagovsky I, Taub E, Marom D, Gaash D, Shichrur K, Avigad S, Hayman Manzur L, Villa A, Sobacchi C, Shohat M, Yaniv I, Stein J. Homozygous Deletion of RAG1, RAG2 and 5' region TRAF6 Causes Severe Immune Suppression and Atypical Osteopetrosis. Causes Severe Immune Suppression and Atypical Osteopetrosis. Clin Genet. 2016 Nov 3. doi: 10.1111/cge.12916. [Epub ahead of print]

51. Schena F, Menale C, Caci E, Diomede L, Palagano E, Recordati C, Sandri M, Tampieri A, Bortolomai I, Capo V, Pastorino C, Bertoni A, Gattorno M, Martini A, Villa A, Traggiai E, Sobacchi C. Murine Rankl-/- Mesenchymal Stromal Cells display an osteogenic differentiation defect improved by a RANKL-expressing lentiviral vector. Stem Cells 35(5):1365-1377, 2017. doi: 10.1002/stem.2574.

52. Palagano E, Slatter MA, Uva P, Menale C, Villa A, Abinun M, Sobacchi C. Hematopoietic Stem Cell Transplantation corrects Osteopetrosis in a child carrying a novel homozygous mutation in the FERMT3 gene. Bone 97:126-129, 2017.

53. Sobacchi C, Palagano E, Villa A, Menale C. Soluble Factors on Stage to Direct Mesenchymal Stem Cells Fate. Front Bioeng Biotechnol. 5:32, 2017.

54. Woodcock TM, Frugier T, Nguyen TT, Semple BD, Bye N, Massara M, Savino B, Besio R, Sobacchi C, Locati M, Morganti-Kossmann MC. The scavenging chemokine receptor ACKR2 has a significant impact on acute mortality rate and early lesion development after traumatic brain injury. PLoS One 12(11):e0188305, 2017.

55. Palagano E, Menale C, Sobacchi C, Villa A. Genetics of osteopetrosis. Curr Rep Osteopor 16(1):13-25, 2018.

56. Hernández-Martínez C, Guzmán-Martínez MN, Scheffler-Mendoza S, Espinosa-Padilla SE, Sobacchi C, Blancas-Galicia L. [Identification of new mutations in TCIRG1 as a cause of infantile malignant osteopetrosis in two Mexican patients]. Rev Alerg Mex 65(1):108-116, 2018.

57. Palagano E, Zuccarini G, Prontera P, Borgatti R, Stangoni G, Elisei S, Mantero S, Menale C, Forlino A, Uva P, Oppo M, Vezzoni P, Villa A, Merlo GR, Sobacchi C. Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. Bone 114:125-136, 2018.

58. Menale C, Campodoni E, Palagano E, Mantero S, Erreni M, Inforzato A, Fontana E, Schena F, van't Hof R, Sandri M, Tampieri A, Villa A, Sobacchi C. MSC-seeded biomimetic scaffolds as a factory of soluble RANKL in Rankl-deficient osteopetrosis. Stem Cell Transl Med, 2018.

59. Sobacchi C, Erreni M, Strina D, Palagano E, Villa A, Menale C. 3D Bone Biomimetic Scaffolds for Basic and Translational Studies with Mesenchymal Stem Cells. Int J Mol Sci 19(10), 2018.

60. Penna S, Capo V, Palagano E, Sobacchi C, Villa A. One Disease, Many Genes: Implications for the Treatment of Osteopetroses. Front Endocrinol (Lausanne) 10:85, 2019.

61. Sobacchi C, Menale C, Villa A. The RANKL-RANK Axis: A Bone to Thymus Round Trip. Front Immunol 10:629, 2019.

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