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Selected peer reviewed publications in the last 5 years

1. **Dardis A**, Buratti E. Impact, Characterization, and Rescue of Pre-mRNA Splicing Mutations in Lysosomal Storage Disorders. Genes (Basel). 2018 Feb 6;9(2).
2. Zampieri S, Cattarossi S, Bembi B, **Dardis A**. GBA Analysis in Next-Generation Era: Pitfalls, Challenges, and Possible Solutions. J Mol Diagn. 2017 Sep;19(5):733-741
3. Goina E, Peruzzo P, Bembi B, **Dardis A***, Buratti E*. Glycogen reduction in myotubes of late-onset Pompe disease patients using antisense technology. Molecular Therapy. 2017 Sep 6;25(9):2117-2128. *** Co-corresponding authors.**
4. **Dardis A**, Zampieri S, Canterini S, Newell KL, Stuani C, Murrell JR, Ghetti B, Fiorenza MT, Bembi B, Buratti E. Altered localization and functionality of TAR DNA Binding Protein 43 (TDP-43) in niemann- pick disease type C. Acta Neuropathol Commun. 2016 May 18;4(1):52
5. Milena Romanello[1], Stefania Zampieri[1], Nadia Bortolotti[2], Laura Deroma[1], Annalisa Sechi[1], Agata Fiumara[3], Rossella Parini[4], Barbara Borroni[5], Francesco Brancati[6], Amalia Bruni[7], Cinzia V. Russo[8], Andrea Bordugo[9], Bruno Bembi[1], **Andrea Dardis**[1.] Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3β,5α,6β-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease Due to NPC1 and SMPD1 Mutations.. Clinica Chimica Acta, 2016 Jan 11;455:39-45.
6. Stefania Zampieri[1], Mirella Filocamo[2], Annalisa Pianta[1], Susanna Lualdi[2], Laura Gort[3], Maria Jose Coll[3], Richard Sinnott[4], Tarekegn Geberhiwot[5], Bruno Bembi[1], Andrea **Dardis.** SMPD1 Mutation Update: Database and comprehensive analysis of published and novel variants.. Human Mutation, 2016 Feb;37(2):139-47.
7. Malini E, Zampieri S, Deganuto M, Romanello M, Sechi A, Bembi B, **Dardis A**. Role of LIMP-2 in the intracellular trafficking of β-glucosidase in different human cellular models. FASEB J. 2015 Sep;29(9):3839-52.
8. Zampieri S, Bianchi E, Cantile C, Saleri R, Bembi B, **Dardis A**. Characterization of a spontaneous novel mutation in the NPC2 gene in a cat affected by Niemann Pick type C disease. PLoS One. 2014 Nov 14;9(11):e112503.
9. Platt FM, Wassif C, Colaco A, **Dardis A**, Lloyd-Evans E, Bembi B, Porter FD. Disorders of cholesterol metabolism and their unanticipated convergent mechanisms of disease. Annu Rev Genomics Hum Genet. 2014;15:173-94.
10. **Dardis A**, Zanin I, Zampieri S, Stuani C, Pianta A, Romanello M, Baralle FE, Bembi B, Buratti E. Functional characterization of the common c.-32-13T>G mutation of GAA gene: identification of potential therapeutic agents. Nucleic Acids Res. 2014 Jan;42(2):1291-302.