

FRANCESCA TONELLI, PhD

Researcher at University of Pavia, Department of Molecular Medicine

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### Researcher (RTDA)

2021-present: Temporary contract as researcher (PON React EU)

Project: Perfluorinated compounds impact on bone cells homeostasis in a zebrafish model for dominant osteogenesis imperfecta.

Supervisor: Prof. Antonella Forlino

University: University of Pavia

### Post doctoral experience

2018-present: Post-Doctoral fellowship

Project: Bone specific delivery of the chemical chaperone 4-phenylbutyrate for osteogenesis imperfecta treatment

Supervisor: Prof. Antonella Forlino

University: University of Pavia

### Education

2014-2017: PhD in Biomedical Sciences

Thesis: OI zebrafish models and their use to develop new pharmacological treatments

University of Pavia

2012-2014: Master degree in biomedical and pharmaceutical Biotechnology

Thesis: Zebrafish Chihuahua: a valid model of osteogenesis imperfecta for the study of new therapeutic approaches

University of Pavia

2009-2012: Bachelor's Degree in biomedical and pharmaceutical Biotechnology

Thesis: Study of expression of collagen receptor DDR1 in human megakaryocytes

2004-2009: High school graduation

School: Liceo scientifico "Galileo Galilei"

## Honors and Awards

New Investigator Award at ICCBH 2019, Salzburg, Austria, 22-25 June 2019

Winner of the "OIF Michael Geisman Fellowship 2019"

## Research interests and technical skills

**Research activity** as PhD student in Biomedical Sciences and post doctoral fellow. My work was focused on the study of molecular defects in macromolecules of skeletal tissue in human diseases. The aim of my project was to generate osteogenesis imperfecta zebrafish models using CRISPR/Cas9 technique and use these models for pharmacological treatments.

### **Teaching activity:**

2020-2021: Contract professor of biochemistry at the University of Pavia.

Co-supervisor of master and PhD thesis students.

### **Skills:**

- Molecular biology: DNA and RNA extraction and purification from cells and tissues, DNA electrophoresis analysis by PCR and RT-qPCR, generation of zebrafish models using the CRISPR-Cas9 strategy, subcloning isolation of plasmidic DNA, *in vitro* transcription.
- Cellular biology: primary cell culture techniques.
- Biochemistry: spectrophotometric techniques, protein extraction and purification from cells and tissues, SDS-PAGE and Western blot.
- Histology: sample preparation and histological staining, optical and fluorescence microscopy, wholemount immunohistochemistry and *in situ* hybridization on zebrafish.
- Animal models: pharmacological treatments using zebrafish, management of zebrafish facility, *in vitro* fertilization and microinjection in zebrafish embryos.

## List of publications and patents

- Cell differentiation and matrix organization are differentially affected during bone formation in osteogenesis imperfecta zebrafish models with different genetic defects impacting collagen type I structure Daponte, V., Tonelli, F., Masiero, C., Syx, D., Exbrayat-Héritier, C., Biggiogera, M., Willaert, A., Rossi, A., Coucke, P.J., Ruggiero, F., Forlino, A. (2023) *Matrix Biology*, 121, pp. 105-126.
- Zebrafish Tric-b is required for skeletal development and bone cells differentiation. Tonelli F, Leoni L, Daponte V, Gioia R, Cotti S, Fiedler IAK, Larianova D, Willaert A, Coucke PJ, Villani S, Busse B, Besio R, Rossi A, Witten PE and Forlino A (2023) *Front. Endocrinol.* 14:1002914.
- Structure, evolution and expression of zebrafish cartilage oligomeric matrix protein (COMP, TSP5). CRISPR-Cas mutants show a dominant phenotype in myosepta. Forte-Gomez HF, Gioia R, Tonelli F, Kobbe B, Koch P, Bloch W, Paulsson M, Zaucke F, Forlino A and Wagener R (2022) *Front. Endocrinol.* 13:1000662.

- Knocking out *TMEM38B* in human foetal osteoblasts hFOB 1.19 by CRISPR/Cas9: A model for recessive OI type XIV. Leoni L, Tonelli F, Besio R, Gioia R, Moccia F, Rossi A, et al. (2021). PLoS ONE 16(9): e0257254
- Zebrafish: A Resourceful Vertebrate Model to Investigate Skeletal Disorders. Tonelli F, Bek JW, Besio R, De Clercq A, Leoni L, Salmon P, Coucke PJ, Willaert A, Forlino A. Front Endocrinol 2020 doi: 10.3389/fendo.2020.00489
- Novel RPL13 variants and variable clinical expressivity in a human ribosomopathy with spondyloepimetaphyseal dysplasia. Costantini A, Alm JJ, Tonelli F, Valta H, Huber C, Tran AN, Daponte V, Kirova N, Kwon YU, Bae JY, Chung WY, Tan S, Sznajder Y, Nishimura G, Näreoja T, Warren AJ, Cormier-Daire V, Kim OH, Forlino A, Cho TJ, Mäkitie O. J Bone Miner Res 2020 doi: 10.1002/jbmr.4177
- Crtap and p3h1 knock out zebrafish support defective collagen chaperoning as the cause of their osteogenesis imperfecta phenotype. Tonelli F, Cotti S, Leoni L, Besio R, Gioia R, Marchese L, Giorgetti S, Gistelinc C, Wagener R, Kobbe B, Larionova D, Fiedler I.A.K., Busse B, Eyre D, Rossi A, Witten PE, Forlino A. Matrix Biology 2020, (90): 40-60. doi: 10.1016/j.matbio.2020.03.004
- Bone biology: insights from osteogenesis imperfecta and related rare fragility syndromes. Besio R, Chow CW, Tonelli F, Marini JC, Forlino A. FEBS J. 2019, (15):3033-3056. doi: 10.1111/febs.14963.
- Steady-State and Pulse-Chase Analyses of Fibrillar Collagen. Forlino A, Tonelli F, Besio R. Methods Mol Biol. 2019;(1952):45-53. doi: 10.1007/978-1-4939-9133-4\_4.
- Severely impaired bone material quality in Chihuahua zebrafish resembles classical dominant human osteogenesis imperfecta. Fiedler I, Schmidt F, Plumeyer C, Milovanovic P, Gioia R, Tonelli F, Bale H, Jähn K, Besio R, Forlino A, Busse B. J Bone Miner Res. 2018, (8):1489-1499. doi: 10.1002/jbmr.3445.
- The chaperone activity of 4PBA ameliorates the skeletal phenotype of Chihuahua, a zebrafish model for dominant osteogenesis imperfecta. Gioia R.\*, Tonelli F.\*, Ceppi I., Biggiogera M., Leikin S., Fisher S., Tenedini E., Yorgan T.A., Schinke T., Tian K., Schwartz J.M., Forte F., Wagener R., Villani S., Rossi A. and Forlino A. Hum Mol Genet 2017, (26):2897-2911. \* first co-authors
- Zebrafish Collagen Type I: Molecular and Biochemical Characterization of the Major Structural Protein in Bone and Skin. Gistelinc C, Gioia R, Gagliardi A, Tonelli F, Marchese L, Bianchi L, Landi C, Bini L, Huysseune A, Witten PE, Staes A, Gevaert K, De Rocker N, Menten B, Malfait F, Leikin S, Carra S, Tenni R, Rossi A, De Paepe A, Coucke P, Willaert A, Forlino A. Sci Rep. 2016, (6):21540.

