Dr. Maria Chiara Pelleri

PERSONAL DETAILS

Name	Maria Chiara Pelleri
Date of Birth	May 27, 1983
Place of Birth	Ravenna, Italy
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EDUCATION

INSTITUTION AND LOCATION	DEGREE	Completion Date MM/YYYY	FIELD OF STUDY
University of Bologna, Italy	PhD	04/2011	Science of human development and movement.
Queen Mary University of London, UK Prof. Greg Elgar Lab	Foreign experience as PhD student	04/2009	Functional genomics
University of Bologna, Italy	M.D.	10/2007	Medical Biotechnology
University of Bologna, Italy	B.S.	09/2005	Biotechnology

A. Personal Statement

My long term research interests involve the development of a comprehensive understanding of the underlying genetic mechanisms of Down syndrome with the ultimate goal of identifying possible new therapeutic approaches.

My academic training and research experience have provided me with an excellent background in multiple biological disciplines including molecular biology, genetics and bioinformatics.

In particular, during my PhD and first years as a Post-Doc, I was published in the field of genomics, developing innovative and integrated approaches for structural and functional characterization of the human genome, with particular attention to human chromosome 21.

In the last five years, my research interest focused on Down syndrome and I have been involved in a project led by Prof. Pierluigi Strippoli with the goal of identifying new therapeutic targets for intellectual disability in DS.

In particular, I have contributed to the draft of the project, with the coordination of the procedures for Ethics Committee Approval. Then, my research focused on a systematic analysis of partial trisomy 21 cases with or without DS. A highly restricted DSCR (HR-DSCR) of only 34 kb on distal 21q22.13 was identified as the minimal region whose duplication is shared by all DS subjects and is absent in all non-DS subjects. HR-DSCR is proposed as a candidate for the typical DS features, the intellectual disability and some facial phenotypes. My current research consists in searching for HR-DSCR functional loci for understanding the fundamental genotype-phenotype relationships in DS.

B. Positions

Positions and Employment

2018-present	Senior assistant professor Department of Experimental, Diagnostic and Specialty Medicine - DIMES, University of Bologna, Italy Academic discipline: BIO/13 Applied Biology
2017-2018	Junior assistant professor Department of Experimental, Diagnostic and Specialty Medicine - DIMES, University of Bologna, Italy Academic discipline: BIO/13 Applied Biology
2015-2017	Post-Doc, Tutor Prof. Pierluigi Strippoli, University of Bologna, Italy
2008-2015	Post-Doc, Tutor Prof. Flavia Frabetti, University of Bologna, Italy

Career disruption

16 June 2014 – 23 November 2014	Maternity leave
18 August 2012 – 23 January 2013	Maternity leave
06 November 2010 – 06 April 2011	Maternity leave

Other Experience and Professional Memberships

2017-present	Lecturer, Applied Biology and Genetics Single cycle degree/Combined Bachelor and Master in Medicine and Surgery, University of Bologna, Italy Bachelor's Degree in Professional Education, School of Medicine, University of Bologna Bachelor's Degree in Dietetics, School of Medicine, University of Bologna
2012-present	Teaching Assistant in the exam boards for Biology and Genetics classes, Single cycle degree programs in Medicine and Surgery, School of Medicine, University of Bologna
2012-present	Co-supervisor of 5 theses (B.Sc, M.Sc., Medicine and Surgery)
2008-present	Tutor, Biology and Genetics laboratory, Single cycle degree programs in Medicine and Surgery, School of Medicine, University of Bologna
2013-2014	Lecturer, Genetics Bachelor's Degree in Nursing, School of Medicine, University of Bologna
2015-present	Member, Associazione Italiana di Biologia e Genetica (A.I.B.G.)
2016-present	Member, European Society of Human Genetics (ESHG)
2016-present	Member, Trisomy 21 Research Society (T21RS)

C. Contribution to Science

During my PhD period and then as a Post-Doc (Tutor Prof. Flavia Frabetti), my research interests were focused on structural and functional characterization of new human genes with particular attention to human chromosome 21 (Hsa21) genes.

During this period, I became competent in the development and usage of innovative computational biology tools for structural characterization of genomic regions (Casadei et al., *Genomics*, 2012; Piovesan et al., *Genomics*, 2013; Piovesan et al., *Mamm. Genome*, 2014) and meta-analysis of data (Lenzi et al., *BMC Genomics*, 2011). I performed molecular biology experiments (nucleic acid extraction, RT-PCR, Real-time PCR) and cell cultures for structural characterization and gene expression studies related to new genes on Hsa21 (Facchin et al. *PloS One*, 2011; Casadei et al., *Mol. Biol. Rep.*, 2014).

I acquired experience in functional genomics techniques applied to Zebrafish (*Danio rerio*): molecular biology experiments (RNA extraction from Zebrafish embryos and tissues; RT-PCR) were performed for the identification of ideal housekeeping genes in Zebrafish (Casadei et al., *Gene Expr Patterns*, 2011), while microinjection techniques were applied to Zebrafish embryos for the analysis of conserved non-coding elements (CNEs) associated with vertebrate development (Doglio et al., *PloS Genet.*, 2013). Currently, my expertise in using Zebrafish is applied to functional genomics experiments for the purpose of characterizing Hsa21 genes involved in complex diseases.

Finally, I was involved in a systematic study presenting an estimation of human total cell number (3.72±0.81x1013) (Bianconi et al., *Annals of Human Biology,* 2013). The results provide for the first time a reference point for studies of biology and human disease.

Under the guidance of Prof. Strippoli, I have recently started a new project as a Post-Doc, directing my knowledge in the field of genomics to the specific topic of Down Syndrome.

I am experienced and competent in research design: I have actively participated in drafting the project (Strippoli et al., *Science PostPrint*, 2013) and I have managed the research project submission to the Ethics Committee of the Policlinico S. Orsola-Malpighi in Bologna which gave their approval in July 2013.

I have extensive experience in bioinformatics tools used for structural characterization of genomic regions (Piovesan et al., *DNA Res.*, 2015; Piovesan et al., *Database (Oxford),* 2016; Vitale et al., *Int J Mol Med.* 2017) and gene expression meta-analysis, with particular attention to Hsa21 genes (Pelleri et al., *BMC Med. Genomics*, 2014; Caracausi et al., *Neurogenetics*, 2014; Caracausi et al., *Hippocampus*, 2016; Mariani et al., *PLoS One*, 2016; Caracausi et al., *J. Cell Physiol.*, 2017; Caracausi et al., *Mol Med Rep*, 2017*;* Vitale et al., *BMC Genomics*, 2017)
To validate the results, independent confirmation by Real-Time reverse transcription polymerase chain reaction (RT-PCR) was performed.

Recently, I have focused my interests on a systematic meta-analysis of all reported partial trisomy 21 cases. The study supported the idea that not all Hsa21 loci are required for the manifestation of DS, rather suggesting a small region on 21q22.13 (highly restricted Down Syndrome critical region, HR-DSCR) as critical to the DS core phenotype (intellectual disability and *facies*) (Pelleri et al., *Hum. Mol. Genet.*, 2016). This result paves the way for the research, in this region, of genes associated to Down syndrome, the function of which might become the target for specific therapy.

Moreover, the same framework can be used for the research of genotype-phenotype correlations regarding other signs and symptoms. We have published an analysis of PT21 cases for the identification of critical regions on Hsa21 associated to congenital heart disease (CHD) in DS, one of the most frequent comorbidities in DS (Pelleri et al., *Genomics*, 2017).

Finally, an important collaboration with one of the leading groups in Italy in the field of metabolomics (Prof. Paola Turano, CERM, University of Florence, Italy) allowed us to publish the first study of metabolomics through Nuclear Magnetic Resonance (NMR) applied to plasma and urine samples of 67 DS subjects and 29 control subjects (Caracausi et al., *Sci Rep*, 2018). Our

study reveals that DS subjects present some alterations in metabolic pathways, in particular, several significantly altered metabolites are produced at the beginning or during the Krebs cycle. The NMR approach used resulted extremely powerful in providing an efficient, high-throughput untargeted picture of the metabolic fingerprint of the DS subjects.
The genetic origin of this metabolic profile possibly related to the HR-DSCR and correlations with intellectual disability in DS are still under investigation.

Complete List of Published Work:

1. Piovesan A, **Pelleri MC**, Antonaros F, Strippoli P, Caracausi M, Vitale L.
On the length, weight and GC content of the human genome.
BMC Res Notes, 12(1):106, 2019.

2. **Pelleri** MC, Cattani C, Vitale L, Antonaros F, Strippoli P, Locatelli C, Cocchi G, Piovesan A, Caracausi M.
Integrated Quantitative Transcriptome Maps of Human Trisomy 21 Tissues and Cells.
Front Genet, 9:125, 2018.

3. Caracausi M, Ghini V, Locatelli C, Mericio M, Piovesan A, Antonaros F, **Pelleri MC**, Vitale L, Vacca RA, Bedetti F, Mimmi MC, Luchinat C, Turano P, Strippoli P, Cocchi G.
Plasma and urinary metabolomic profiles of Down syndrome correlate with alteration of mitochondrial metabolism.
Sci Rep, 8:2977, 2018.

4. Vitale L, Piovesan A, Antonaros F, Strippoli P, **Pelleri MC**, Caracausi M.
A molecular view of the normal human thyroid structure and function reconstructed from its reference transcriptome map.
BMC Genomics, 18:739, 2017.

5. Caracausi M, Piovesan A, Antonaros F, Strippoli P, Vitale L, **Pelleri MC**.
Systematic identification of human housekeeping genes possibly useful as references in gene expression studies.
Mol Med Rep, 16:2397-2410, 2017.

6. **Pelleri MC**, Gennari E, Locatelli C, Piovesan A, Caracausi M, Antonaros F, Rocca A, Donati CM, Conti L, Strippoli P, Seri M, Vitale L, Cocchi G.
Genotype-phenotype correlation for congenital heart disease in Down syndrome through analysis of partial trisomy 21 cases.
Genomics, 109:391-400, 2017

7. Vitale L, Caracausi M, Casadei R, **Pelleri MC**, Piovesan A.
Difficulty in obtaining the complete mRNA coding sequence at 5' region (5' end mRNA artifact): causes, consequences in biology and medicine and possible solutions for obtaining the actual amino acid sequence of proteins (Review).
Int J Mol Med, 39:1063-1071, 2017

8. Caracausi M, Piovesan A, Vitale L, **Pelleri MC**.
Integrated Transcriptome Map Highlights Structural and Functional Aspects of the Normal Human Heart.

J Cell Physiol, 232:759-770, 2017.

9. Piovesan A, Caracausi M, Antonaros F, **Pelleri MC**, Vitale L.
GeneBase 1.1: a tool to summarize data from NCBI gene datasets and its application to an update of human gene statistics.
Database: The Journal of Biological Databases and Curation, pii: baw153. doi: 10.1093/database/baw153, 2016.

10. Mariani E, Frabetti F, Tarozzi A, **Pelleri MC**, Pizzetti F, Casadei R.
Meta-Analysis of Parkinson's Disease Transcriptome Data Using TRAM Software:
Whole Substantia Nigra Tissue and Single Dopamine Neuron Differential Gene Expression.
PLoS One, 11(9):e0161567, 2016.

11. **Pelleri MC**, Cicchini E, Locatelli C, Vitale L, Caracausi M, Piovesan A, Rocca A, Poletti G, Seri M, Strippoli P, Cocchi G.
Systematic reanalysis of partial trisomy 21 cases with or without Down syndrome suggests a small region on 21q22.13 as critical to the phenotype.
Hum Mol Genet 25:2525-2538, 2016.

12. Caracausi M, Rigon V, Piovesan A, Strippoli P, Vitale L, **Pelleri MC**.
A quantitative transcriptome reference map of the normal human hippocampus.
Hippocampus, 26:13-26, 2016.

13. Piovesan A, Caracausi M, Ricci M, Strippoli P, Vitale L, **Pelleri MC**.
Identification of minimal eukaryotic introns through GeneBase, a user-friendly tool for parsing the NCBI Gene databank.
DNA Res, 22:495-503, 2015.

14. **Pelleri MC**, Piovesan A, Caracausi M, Berardi A, Vitale L, Strippoli P.
Integrated differential transcriptome maps of Acute Megakaryoblastic Leukemia (AMKL) in children with or without Down Syndrome (DS).
BMC Med Genomics, 7(1):63, 2014.

15. Caracausi M, Vitale L, **Pelleri MC**, Piovesan A, Bruno S, Strippoli P.
A quantitative transcriptome reference map of the normal human brain.
Neurogenetics, 15:267-287, 2014.

16. Casadei R, **Pelleri MC**, Vitale L, Facchin F, Canaider S, Strippoli P, Vian M, Piovesan A, Bianconi E, Piva F, Frabetti F.
Characterization of human gene locus CYYR1: a complex multi-transcript system.
Mol Biol Rep, 41:6025-6038, 2014.

17. Piovesan A, Caracausi M, **Pelleri MC**, Vitale L, Martini S, Bassani C, Gurioli A, Casadei R, Soldà G, Strippoli P.
Improving mRNA 5' coding sequence determination in the mouse genome.
Mamm Genome, 25:149-159, 2014.

18. Strippoli P, **Pelleri MC**, Caracausi M, Vitale L, Piovesan A, Locatelli C, Mimmi MC, Berardi AC, Ricotta D, Radeghieri A, Barisani D, Basik M, Monaco MC, Ghezzo A, Seri M, Cocchi G.

An integrated route to identifying new pathogenesis-based therapeutic approaches for trisomy 21 (Down Syndrome) following the thought of Jérôme Lejeune.
Science Postprint 1(1): e00010. doi:10.14340/spp.2013.12R0005, 2013.

19. Doglio L, Goode DK, **Pelleri MC**, Pauls S, Frabetti F, Shimeld SM, Vavouri T, Elgar G.
Parallel Evolution of Chordate Cis-Regulatory Code for Development.
PLoS Genet, 9(11):e1003904, 2013.

20. Bianconi E, Piovesan A, Facchin F, Beraudi A, Casadei R, Frabetti F, Vitale L, **Pelleri MC**, Tassani S, Piva F, Perez-Amodio S, Strippoli P, Canaider S.
An estimation of the number of cells in the human body.
Ann Hum Biol, 40:463-471, 2013.

21. Piovesan A, Vitale L, **Pelleri MC**, Strippoli P.
Universal tight correlation of codon bias and pool of RNA codons (codonome): the genome is optimized to allow any distribution of gene expression values in the transcriptome from bacteria to humans.
Genomics, 101:282-289, 2013.
I.F. = 2.801

22. Casadei R, Piovesan A, Vitale L, Facchin F, **Pelleri MC**, Canaider S, Bianconi E, Frabetti F, Strippoli P.
Genome-scale analysis of human mRNA 5′ coding sequences based on expressed sequence tags (EST) database.
Genomics, 100:125-130, 2012.

23. Facchin F, Vitale L, Bianconi E, Piva F, Frabetti F, Strippoli P, Casadei R, **Pelleri MC**, Piovesan A, Canaider S.
Complexity of bidirectional transcription and alternative splicing at human RCAN3 locus.
PLoS One, 6(9):e24508, 2011.

24. Lenzi L, Facchin F, Piva F, Giulietti M, **Pelleri MC**, Frabetti F, Vitale L, Casadei R, Canaider S, Bortoluzzi S, Coppe A, Danieli GA, Principato G, Ferrari S, Strippoli P.
TRAM (Transcriptome Mapper): database-driven creation and analysis of transcriptome maps from multiple sources.
BMC Genomics, 12:121, 2011.

25. Casadei R, **Pelleri MC**, Vitale L, Facchin F, Lenzi L, Canaider S, Strippoli P, Frabetti F.
Identification of housekeeping genes suitable for gene expression analysis in the zebrafish.
Gene Expr Patterns, 11:271-276, 2011.