

## CURRICULUM VITAE

NOME COGNOME	<b>Mirella Filocamo</b>	
ISTRUZIONE SCOLASTICA	1970	• Diploma di Maturità Classica-Liceo Classico di Locri (RC)
	1975	• Laurea in Scienze Biologiche-Università di Pisa
	1998	• Specializzazione in Genetica Medica (cum laude) Università di Genova
LINGUA STRANIERA	Inglese	
ATTIVITÀ PROFESSIONALE	1975- a oggi	“Lab. Diagnosi Pre e Postnatale Malattie Metaboliche”- Istituto G. Gaslini Genova
	<i>In qualità di</i>	<ul style="list-style-type: none"> <li>• Borsista (1975-1978)</li> <li>• Assistente Biologo (1978-1991)</li> <li>• Dirigente Biologo 1° livello fascia A (1991-2000)</li> <li>• Responsabile Modulo Dipartimentale di Laboratorio 1/01/2001 - 30/06/2012</li> <li>• Responsabile UOSD - Centro di diagnostica genetica e biochimica delle malattie metaboliche (ex Lab Diagnosi Pre e Postnatale Malattie Metaboliche) dal 1/07/2012 -</li> </ul>
ASSOCIAZIONI	<ul style="list-style-type: none"> <li>• European Working Group on Gaucher Disease (EWGGD)</li> <li>• European Working Group on Lysosomal Disease (ESGLD)</li> <li>• Società Italiana di Genetica Umana (SIGU)</li> <li>• Società Italiana per lo Studio delle Malattie Metaboliche Ereditarie (SISMME)</li> </ul>	
ATTIVITÀ SCIENTIFICA	<ul style="list-style-type: none"> <li>• Coordinatore progetto multicentrico “Telethon Genetic Biobank Network” (<a href="http://biobanknetwork.telethon.it/">http://biobanknetwork.telethon.it/</a>)</li> <li>• Conferimento “Award of 2010 Gaucher Generation grants”</li> <li>• Conferimento “Premio DNA 2013”- 1ª edizione (Ordine Nazionale dei Biologi)</li> <li>• Coautore delle Linee guida “Biobanche Genetiche” pubblicate come inserto redazionale della rivista Analysis N5/6 dicembre 2003</li> <li>• Coautore di Disciplinari SIGU per l’Accreditamento delle Strutture di Genetica Medica- Le Biobanche Genetiche. Analysis 4/5.2009</li> <li>• Coordinatore gruppo di lavoro SIGU per la stesura di Standard per il Sistema di Gestione per la Qualità nelle Biobanche Genetiche</li> <li>• Membro di: <ul style="list-style-type: none"> <li>- Tavolo tecnico per le Biobanche su incarico della Regione Liguria</li> <li>- Tavolo tecnico per la costituzione del Nodo Italiano BBMRI (Biobanking and Biomolecular Resources Research Infrastructure)</li> <li>- BRIF (Bioresource Research Impact Factor), gruppo di lavoro europeo costituito da 34 partecipanti (10 paesi EU) per l’assegnazione di IF alle biorisorse tramite la standardizzazione della loro citazione nelle pubblicazioni</li> </ul> </li> <li>• Docente in corsi di aggiornamento, seminari e convegni nazionali e internazionali</li> <li>• Peer-review di Human Mutation, Journal of Cellular and Molecular Medicine, Clinical Genetics, Clinica Chimica Acta, BBA-Mol Basis Dis, Blood Cell Mol Dis, European Journal of Human Genetics, American Journal of Medical Genetics, Molecular Genetics and Metabolism, J Inherit Metab Dis, Cellular and Molecular Life Sciences, PLOS ONE, Orphanet Journal of Rare Diseases, Clinica Chimica Acta, Metabolic Brain Disease, Human Genetics, Neuromuscular Disorders</li> <li>• Autore/Coautore di capitoli di libri/enciclopedie scientifiche e di articoli scientifici (peer-reviewed) su riviste impattate</li> </ul>	

### **Pubblicazioni: libri/enciclopedie:**

- 1 Borrone C, **Filocamo M.** La diagnosi prenatale. In: "Dal DNA alle malattie ereditarie" di G. Romeo. Casa Editrice Ambrosiana.1983, Cap.16

- 2 Gatti R, Lombardo C, **Filocamo M**, Borrone C and Porro E. Comparison of the activities of 15 lysosomal enzymes in chorionic villi and in cultured amniotic fluid cells. First trimester fetal diagnosis. Ed. by M. Fraccaro et al. Springer Verlag. 1985; p.238.
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- 5 **Filocamo M**, Morrone A: Lysosomal storage disorders - epidemiology, biochemistry, and genetics: how to read and interpret biochemical and molecular tests. In: Lysosomal storage disorders: early diagnosis and new treatments. Mariani Foundation Paediatric Neurology Series 23; 2010.
- 6 Dagna Bricarelli F, Baldo C, Rossi M, Bellomo R, **Filocamo M**: Le Biobanche genetiche: diagnosi e ricerca. In Scienza, Tecnologia e Diritto, Ed. Amon, 2011, p.109
- 7 **Filocamo M**, Cooper DN, Di Rocco M: Mucopolysaccharide Storage Disorders. In Encyclopedia of Life Sciences (ELS). John Wiley & Sons, Ltd: Chichester. 2011. DOI: 10.1002/9780470015902.a0006095
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#### **Publicazioni (peer-reviewed): riviste internazionali**

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- 2 Gatti C, Borrone C, Torreblanca J, Cavalieri S, De Martini I, **Filocamo M**, Antelo y MC. Características biológicas de las Mucopolisidosis II y III. Annales Espanoles de Pediatria. 1979;12: 563-574.
- 3 Durand P, Borrone C, Gatti R, **Filocamo M**. Results of prenatal diagnosis for genetic metabolic diseases in Italy. Recent Advance in Prenatal Diagnosis, Ed. J. Wiley and Sons New York. 1981, p. 249.
- 4 Gatti R, Borrone C, Salemi D, **Filocamo M**, Sanna G, Potier M, Durand P. Sialidosis: clinical and biochemical studies of distinct phenotypes. Perspectives in Inherited Metabolic Diseases. 1981; 4:365-378.
- 5 **Filocamo M**, Di Rocco M, Rolando S, Schiappapietra M, Costantino G, Fucosidosis: review of personal experience, *Pediatr Med Chir*, 1982;4:185-194.
- 6 Cerruti Mainardi P, Gatti R, Javarone A, **Filocamo M**, Levis F, Borrone C, Mannosidosis, Study of two families and prenatal diagnosis, *Pediatr Med Chir* 1982;4:203-214.
- 7 Garibaldi LR, Canini S, Superti-Furga A, Lamedica G, **Filocamo M**, Marchese N, Borrone C, Galactosemia caused by generalized uridine diphosphate galactose-4-epimerase deficiency, *J Pediatr*, 1983;103:927-930.
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- 10 Pendola F, Borrone C, **Filocamo M**, Lituanica M, Steinmann B and Superti-Furga A. Radiological "metamorphosis" in a patient with severe congenital osteogenesis imperfecta. *Eur.J.Pediatr*. 1990; 149:403-405.
- 11 Tenni R, Biglino P, Dyne K, Rossi A, **Filocamo M**, Pendola F, Brunelli P, Borrone C and Cetta G. Phenotypic Variability and Abnormal Type I Collagen Unstable at Body Temperature in a Family with Mild Dominant Osteogenesis Imperfecta. *Inher. Metab. Dis*. 1991; 14:189-201.
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- 14 Caruso U, Cerone R, Schiaffino MC, Minniti G, Romano C, **Filocamo M**, Colombo JP. Prenatal Diagnosis of Argininemia: Experience on two pregnancies in the same family. *International Pediatrics Suppl.* 1994; 2:9.
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- 27 Romano M, Danek G, Baralle F, Mazzotti R and **Filocamo M**. Functional characterisation of the novel mutation IVS 8 (-11delC)(-14T->A) in the intron 8 of the glucocerebrosidase gene of two Italian siblings with Gaucher disease type I. *Blood Cells, Molecules and Disease.* 2000;26:171-176.
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- 34 Stroppiano M, Bonuccelli G, Corsolini F and **Filocamo M**. Aberrant splicing at catalytic site environment as cause of infantile onset glycogen storage disease type II (GSDII): molecular identification of a novel IVS9 (+2GT→GC) in combination with rare IVS10 (+1 GT→CT). *AJMG*. 2001;101:55-58.
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- 47 Parazzini C, Arena S, Marchetti L, Menni F, **Filocamo M**, Verheijen FW, Mancini GM, Triulzi F, Parini R. Infantile Sialic Acid Storage Disease: Serial Ultrasound and Magnetic Resonance Imaging Features. *AJNR* 2003; 24: 398-400.
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Autorizzo il trattamento dei miei dati personali ai sensi del Decreto Legislativo 30 giugno 2003, n. 196 "Codice in materia di protezione dei dati personali (facoltativo)".

Mirella Filocamo

Genova, 018/09/2017

