

## CURRICULUM VITAE

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INDIRIZZO		
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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://www.biobanknetwork.com/">http://www.biobanknetwork.com/</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</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 e al. Springer Verlag. 1985; p.238.
- 3 **Filocamo M**, Tonlorenzi R, Stroppiano M, Corsolini F, De Biasio P. Lysosomal Enzyme Activities on Cultured Chorionic Villi (CCV) for First Trimester Prenatal Diagnosis of I - Cell disease; in Early Fetal Diagnosis: Recent Progress and Public Health Implications, Ed.: M. Macek, M.A. Ferguson-Smith, M.Spala. Karolinum-Charles University Press, Prague. 1992; p.401.
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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

**Pubblicazioni (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 Mucopolipidosis II y III. Annales Espanoles de Pediatria. 1979;12: 563-574.
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- 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.
- 8 Gatti R, Borrone C, **Filocamo M**, Pannone N and Di Natale P. Prenatal diagnosis of Mucopolysaccharidosis I: a special difficulty arising from an unusually low enzyme activity in mother's cells. *Prenatal Diagnosis*. 1985; 5:149-154.
- 9 Gatti R, Lombardo C, **Filocamo M**, Borrone C, Porro E. Comparative study of 15 lysosomal enzyme in chorionic villi and cultured amniotic fluid cells. *Prenatal Diagnosis*. 1985; 5:329-336.
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- 35 **Filocamo M**, Bonuccelli G, Corsolini F, Mazzotti R, Cusano R and Gatti R. Molecular analysis of 40 Italian patients with Mucopolysaccharidosis type II: new mutations in the iduronate-2-sulfatase gene. *Hum Mutat*. 2001;18: 164-165.
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- 41 **Filocamo M**, Mazzotti R, Stroppiano M, Seri M, Giona F, Parenti G, Regis S, Corsolini F, Zoboli S and Gatti R. Analysis of the Glucocerebrosidase Gene and Mutation Profile in 144 Italian Gaucher Patients. *Human Mutation*. 2002; 20:234-235.
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