CURRICULUM VITAE
Dr Livia Garavelli

PERSONAL INFORMATION

Name / Surname Livia Garavelli
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Nationality

Date of birth

EDUCATION AND TRAINING

1993	Specialty Board in Genetics (University of Ferrara)
1989-1993	Student at the Genetic Institute, University of Ferrara *Fields of interest:* growth retardation in genetic syndromes, congenital malformations in twins, genetic counselling
1985	Specialty Board in Pediatrics (University of Parma)
1980-1985	Student at the Clinical Genetic Surgery, Pediatric Department, University of Parma *Fields of interest:* Turner syndrome, clinical genetics
1981	M.D. degree (University of Parma)

PROFESSIONAL EXPERIENCE

2007-present	Medical doctor permanent position, Director of the Clinical Genetics Unit, Maternal and Child Health Department, AUSL-IRCCS of Reggio Emilia (formerly Obstetric and Pediatric Department, Arcispedale S. Maria Nuova, Reggio Emilia), Italy *Fields of interest:* clinical genetics, postnatal diagnosis of genetic syndromes, follow-up of children with MCA/MR, hereditary hearing loss, genetic counselling, Mowat-Wilson syndrome, skeletal dysplasias
1987-2006	Medical doctor permanent position at Pediatric Department, Arcispedale S. Maria Nuova, Reggio Emilia,Italy *Fields of interest:* clinical genetics, postnatal diagnosis of genetic syndromes, follow-up of children with MCA/MR, hereditary hearing loss, Mowat-Wilson syndrome
2012-present	Professor for the Course of Medical Genetics, University of Modena and Reggio Emilia, Italy
2002-2014	Professor for the Course of Syndromology, Medical School of Pediatrics, University of Parma, Italy

AWARDS, HONORS, GRANTS AND OTHER INFORMATION

2018	National Scientific Qualification for the role of Second Degree Professor in the Field of Medical Genetics
2015	Telethon Foundation 1-year grant for the project "Genotype-phenotype correlation for magnetic resonance imaging features of Mowat-Wilson syndrome with *Zeb2* mutation/deletion; future discoveries of the role of the gene Zeb2 in the development of the human Brain"; position: Coordinator
2013-present	Member of the Working Group for Quality Assessment in Genetic Counselling, Genetic Sevices Quality Committee, European Society of Human Genetics
2009	Paul Harris Fellow Award for the Clinical Genetics Activity
1999	Quality Award for Innovative Quality in Clinical Genetics, Arcispedale Santa Maria Nuova, Reggio Emilia, Italy
2012-present	Editorial Board Member of Molecular Syndromology
????-present	Member of the Scientific Committee of the Italian Association of Mowat-Wilson syndrome
other memberships:	Italian Society of Pediatrics (SIP), Italian Society of Human Genetics (SIGU) International Skeletal Displasia Society (ISDS)

PUBLICATIONS

Author/coauthor of 112 publications on peer-reviewed journals

h-index: 25 (Scopus, Author Id 56009178700)

PUBLICATIONS ON INDEXED JOURNALS, LAST 15 YEARS:

1. De Bernardi, ML., Ivanovski, I., Caraffi SG., Maini I., Street ME., Bayat A., Zollino M., Lepri FR., Gnazzo M., Errichiello E., Superti-Furga A., Garavelli L.. Prominent and elongated coccyx, a new manifestation of KBG syndrome associated with novel mutation in ANKRD11 Am J Med Genet A. 2018 Aug 8. doi: 10.1002/ajmg.a.40386.
2. Spunton M., Garavelli L., Cerruti Mainardi P., Emmig U., Finale E., Guala A.. Anesthesia in Mowat-Wilson syndrome:informations on 11 Italian patients Pediatr Rep. 2018 Mar 29;10(1):7514. doi: 10.4081/pr.2018.7514. eCollection 2018 Mar 22.
3. Ivanovski, I., Djuric, O., Caraffi, S. G., Santodirocco, D., Pollazzon, M., Rosato, S., . . . Garavelli, L. (2018) Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. Genetics in Medicine. doi: 10.1038/gim.2017.221. [Epub ahead of print]
4. Ivanovski, I., Akbaroghli, S., Pollazzon, M., Gelmini, C., Caraffi, S. G., Mansouri, M., Garavelli L., . . . Hennekam, R. C. (2018). Van Maldergem syndrome and Hennekam syndrome: Further delineation of allelic phenotypes. American Journal of Medical Genetics, Part A, 176(5), 1166-1174. doi:10.1002/ajmg.a.38652
5. Maini, I., Farnetti, E., Caraffi, S. G., Ivanovski, I., De Bernardi, M. L., Gelmini, C., . . . Garavelli, L. (2018). A novel CCND2 mutation in a previously reported case of megalencephaly and perisylvian polymicrogyria with postaxial polydactyly and hydrocephalus. Neuropediatrics, 49(3), 222-224. doi:10.1055/s-0038-1641722
6. Maini, I., Ivanovski, I., Djuric, O., Caraffi, S. G., Errichiello, E., Marinelli, M., . . . Garavelli, L. (2018). Prematurity, ventricular septal defect and dysmorphisms are independent predictors of pathogenic copy number variants: A retrospective study on array-CGH results and phenotypical features of 293 children with neurodevelopmental disorders and/or multiple congenital anomalies. Italian Journal of Pediatrics, 44(1) doi:10.1186/s13052-018-0467-z

7. Bonatti, F., Adorni, A., Matichecchia, A., Mozzoni, P., Uliana, V., Pisani, F., . . . Percesepe, A. (2017). Patterns of novel alleles and genotype/phenotype correlations resulting from the analysis of 108 previously undetected mutations in patients affected by neurofibromatosis type I. International Journal of Molecular Sciences, 18(10) doi:10.3390/ijms18102071
8. Garavelli, L., Ivanovski, I., Caraffi, S. G., Santodirocco, D., Pollazzon, M., Cordelli, D. M., . . . Paciorkowski, A. R. (2017). Neuroimaging findings in mowat-wilson syndrome: A study of 54 patients. Genetics in Medicine, 19(6), 691-700. doi:10.1038/gim.2016.176
9. Ivanovski, I., Ješić, M., Ivanovski, A., Garavelli, L., & Ivanovski, P. (2017). Metabolically based liver damage pathophysiology in patients with urea cycle disorders - A new hypothesis. World Journal of Gastroenterology, 23(44), 7930-7938. doi:10.3748/wjg.v23.i44.7930
10. Palazzo, V., Provenzano, A., Becherucci, F., Sansavini, G., Mazzinghi, B., Orlandini, V., . . . Giglio, S. (2017). The genetic and clinical spectrum of a large cohort of patients with distal renal tubular acidosis. Kidney International, 91(5), 1243-1255. doi:10.1016/j.kint.2016.12.017
11. Pizzamiglio, M. R., Piccardi, L., Bianchini, F., Canzano, L., Palermo, L., Fusco, F., . . . Ursini, M. V. (2017). Cognitive-behavioural phenotype in a group of girls from 1.2 to 12 years old with the incontinentia pigmenti syndrome: Recommendations for clinical management. Applied Neuropsychology: Child, 6(4), 327-334. doi:10.1080/21622965.2016.1188388
12. Dauwerse, J. G., Van Belzen, M., Van Haeringen, A., Van Santen, G., Van De Lans, C., Rahikkala, E., . . . Peters, D. (2016). Analysis of mutations within the intron20 splice donor site of CREBBP in patients with and without classical RSTS. European Journal of Human Genetics, 24(11), 1639-1643. doi:10.1038/ejhg.2016.47
13. Garavelli, L., Maini, I., Baccilieri, F., Ivanovski, I., Pollazzon, M., Rosato, S., . . . Tartaglia, M. (2016). Natural history and life-threatening complications in myhre syndrome and review of the literature. European Journal of Pediatrics, 175(10), 1307-1315. doi:10.1007/s00431-016-2761-3
14. Maini, I., Ivanovski, I., Iodice, A., Rosato, S., Pollazzon, M., Mussini, M., . . . Garavelli, L. (2016). Endocrinological abnormalities are a main feature of 17p13.1 microduplication syndrome: A new case and literature review. Molecular Syndromology, 7(6), 337-343. doi:10.1159/000450718
15. Parenti, I., Gervasini, C., Pozojevic, J., Wendt, K. S., Watrin, E., Azzollini, J., . . . Kaiser, F. J. (2016). Expanding the clinical spectrum of the 'HDAC8-phenotype' - implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 89(5), 564-573. doi:10.1111/cge.12717
16. Rosato, S., Syx, D., Ivanovski, I., Pollazzon, M., Santodirocco, D., De Marco, L., . . . Malfait, F. (2016). RIN2 syndrome: Expanding the clinical phenotype. American Journal of Medical Genetics, Part A, 170(9), 2408-2415. doi:10.1002/ajmg.a.37789
17. Van Karnebeek, C. D. M., Bonafé, L., Wen, X. -., Tarailo-Graovac, M., Balzano, S., Royer-Bertrand, B., . . . Superti-Furga, A. (2016). NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 48(7), 777-784. doi:10.1038/ng.3578
18. Caciotti, A., Tonin, R., Rigoldi, M., Ferri, L., Catarzi, S., Cavicchi, C., . . . Morrone, A. (2015). Optimizing the molecular diagnosis of GALNS: Novel methods to define and characterize morquio-A syndrome-associated mutations. Human Mutation, 36(3), 357-368. doi:10.1002/humu.22751
19. Gannon, T., Perveen, R., Schlecht, H., Ramsden, S., Anderson, B., Kerr, B., . . . Clayton-Smith, J. (2015). Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 23(9), 1165-1170. doi:10.1038/ejhg.2014.248
20. Garavelli, L., Cordeddu, V., Errico, S., Bertolini, P., Street, M. E., Rosato, S., . . . Tartaglia, M. (2015). Noonan syndrome-like disorder with loose anagen hair: A second case with neuroblastoma. American Journal of Medical Genetics, Part A, 167(8), 1902-1907. doi:10.1002/ajmg.a.37082
21. Graziano, C., Wischmeijer, A., Pippucci, T., Fusco, C., Diquigiovanni, C., Nõukas, M., . . . Seri, M. (2015). Syndromic intellectual disability: A new phenotype caused by an aromatic amino acid decarboxylase gene (DDC) variant. Gene, 559(2), 144-148. doi:10.1016/j.gene.2015.01.026
22. Ivanovski, I., Garavelli, L., Djurić, O., Ćirović, A., Škorić, D., & Ivanovski, P. I. (2015). Mitotic crossover promotes leukemogenesis in children born with TEL-AML1 via the generation of loss of heterozygosity at 12p. Pediatria Medica e Chirurgica, 37(2), 1-3. doi:10.4081/pmc.2015.112
23. Maas, S. M., Shaw, A. C., Bikker, H., Lüdecke, H. -., van der Tuin, K., Badura-Stronka, M., . . . Hennekam, R. C. (2015). Phenotype and genotype in 103 patients with tricho-rhino-phalangeal syndrome. European Journal of Medical Genetics, 58(5), 279-292. doi:10.1016/j.ejmg.2015.03.002

24. Mari, F., Marozza, A., Mencarelli, M. A., Lo Rizzo, C., Fallerini, C., Dosa, L., . . . Renieri, A. (2015). Coffin-siris and nicolaides-baraitser syndromes are a common well recognizable cause of intellectual disability. Brain and Development, 37(5), 527-536. doi:10.1016/j.braindev.2014.08.009
25. Mauri, L., Franzoni, A., Scarcello, M., Sala, S., Garavelli, L., Modugno, A., . . . Penco, S. (2015). SOX2, OTX2 and PAX6 analysis in subjects with anophthalmia and microphthalmia. European Journal of Medical Genetics, 58(2), 66-70. doi:10.1016/j.ejmg.2014.12.005
26. Orivoli, S., Pavlidis, E., Cantalupo, G., Pezzella, M., Zara, F., Garavelli, L., . . . Piccolo, B. (2015). Xp1122 microduplications including HUWE1: Case report and literature review. Neuropediatrics, 47(1), 51-56. doi:10.1055/s-0035-1566233
27. Riccardi, F., Rivolta, G. F., Uliana, V., Grati, F. R., La Starza, R., Marcato, L., . . . Martorana, D. (2015). Cryptic 13q34 and 4q35.2 deletions in an italian family. Cytogenetic and Genome Research, 147(1), 24-30. doi:10.1159/000442068
28. Volpi, N., Coppa, G. V., Zampini, L., MacCari, F., Galeotti, F., Garavelli, L., . . . Gabrielli, O. (2015). Plasmatic and urinary glycosaminoglycan profile in a patient affected by multiple sulfatase deficiency. Clinical Chemistry and Laboratory Medicine, 53(7), e157-e160. doi:10.1515/cclm-2014-0997
29. Wenger, T. L., Harr, M., Ricciardi, S., Bhoj, E., Santani, A., Adam, M. P., . . . Zackai, E. H. (2015). Erratum to "CHARGE-like presentation, craniosynostosis and mild mowat-wilson syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases" american journal of medical genetics part A. 164:2557-2566, 2014. American Journal of Medical Genetics, Part A, 167(7), 1682-1683. doi:10.1002/ajmg.a.36860
30. Alders, M., Al-Gazali, L., Cordeiro, I., Dallapiccola, B., Garavelli, L., Tuysuz, B., . . . Hennekam, R. C. (2014). Hennekam syndrome can be caused by FAT4 mutations and be allelic to van maldergem syndrome. Human Genetics, 133(9), 1161-1167. doi:10.1007/s00439-014-1456-y
31. Forlino, A., Vetro, A., Garavelli, L., Ciccone, R., London, E., Stratakis, C. A., & Zuffardi, O. (2014). PRKACB and carney complex. New England Journal of Medicine, 370(11), 1065-1067. doi:10.1056/NEJMc1309730
32. Garavelli, L., Santoro, L., Iori, A., Gargano, G., Braibanti, S., Pedori, S., . . . Gabrielli, O. (2014). Multiple sulfatase deficiency with neonatal manifestation. Italian Journal of Pediatrics, 40, 86. doi:10.1186/s13052-014-0086-2
33. Gardeitchik, T., Mohamed, M., Fischer, B., Lammens, M., Lefeber, D., Lace, B., . . . Morava, E. (2014). Clinical and biochemical features guiding the diagnostics in neurometabolic cutis laxa. European Journal of Human Genetics, 22(7), 888-895. doi:10.1038/ejhg.2013.154
34. Gargano, G., Guidotti, I., Balestri, E., Vagnarelli, F., Rosato, S., Comitini, G., . . . Garavelli, L. (2014). Hydrops fetalis in a preterm newborn heterozygous for the c.4A>G SHOC2 mutation. American Journal of Medical Genetics, Part A, 164(4), 1015-1020. doi:10.1002/ajmg.a.36376
35. Iughetti, L., Predieri, B., Bruzzi, P., Predieri, F., Vellani, G., Madeo, S. F., . . . Bozzola, M. (2014). Ten-year longitudinal study of thyroid function in children with down's syndrome. Hormone Research in Paediatrics, 82(2), 113-121. doi:10.1159/000362450
36. Micale, L., Augello, B., Maffeo, C., Selicorni, A., Zucchetti, F., Fusco, C., . . . Merla, G. (2014). Molecular analysis, pathogenic mechanisms, and readthrough therapy on a large cohort of kabuki syndrome patients. Human Mutation, 35(7), 841-850. doi:10.1002/humu.22547
37. Pizzamiglio, M. R., Piccardi, L., Bianchini, F., Canzano, L., Palermo, L., Fusco, F., . . . Ursini, M. V. (2014). Incontinentia pigmenti: Learning disabilities are a fundamental hallmark of the disease. PLoS ONE, 9(1) doi:10.1371/journal.pone.0087771
38. Sousa, S. B., Hennekam, R. C., Abdul-Rahman, O., Alders, M., Azzarello-Burri, S., Bottani, A., . . . Hennekam, R. C. (2014). Phenotype and genotype in nicolaides-baraitser syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 166(3), 302-314. doi:10.1002/ajmg.c.31409
39. Wenger, T. L., Harr, M., Ricciardi, S., Bhoj, E., Santani, A., Adam, M. P., . . . Zackai, E. H. (2014). CHARGE-like presentation, craniosynostosis and mild mowat-wilson syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. American Journal of Medical Genetics, Part A, 164(10), 2557-2566. doi:10.1002/ajmg.a.36696
40. Cordelli, D. M., Garavelli, L., Savasta, S., Guerra, A., Pellicciari, A., Giordano, L., . . . Franzoni, E. (2013). Epilepsy in mowat-wilson syndrome: Delineation of the electroclinical phenotype. American Journal of Medical Genetics, Part A, 161(2), 273-284. doi:10.1002/ajmg.a.35717
41. Cordelli, D. M., Pellicciari, A., Kiriazopulos, D., Franzoni, E., & Garavelli, L. (2013). Epilepsy in mowat-wilson syndrome: Is it a matter of GABA? Epilepsia, 54(7), 1331-1332. doi:10.1111/epi.12204

42. Garavelli, L., Piemontese, M. R., Cavazza, A., Rosato, S., Wischmeijer, A., Gelmini, C., . . . Superti-Furga, A. (2013). Multiple tumor types including leiomyoma and wilms tumor in a patient with gorlin syndrome due to 9q22.3 microdeletion encompassing the PTCH1 and FANC-C loci. American Journal of Medical Genetics, Part A, 161(11), 2894-2901. doi:10.1002/ajmg.a.36259
43. Garavelli, L., Simonte, G., Rosato, S., Wischmeijer, A., Albertini, E., Guareschi, E., . . . Grzeschik, K. -. (2013). Focal dermal hypoplasia (goltz-gorlin syndrome): A new case with a novel variant in the PORCN gene (c.1250T>C:P.F417S) and unusual spinal anomaly. American Journal of Medical Genetics, Part A, 161(7), 1750-1754. doi:10.1002/ajmg.a.35964
44. Gervasini, C., Russo, S., Cereda, A., Parenti, I., Masciadri, M., Azzollini, J., . . . Larizza, L. (2013). Cornelia de lange individuals with new and recurrent SMC1A mutations enhance delineation of mutation repertoire and phenotypic spectrum. American Journal of Medical Genetics, Part A, 161(11), 2909-2919. doi:10.1002/ajmg.a.36252
45. Ritelli, M., Dordoni, C., Venturini, M., Chiarelli, N., Quinzani, S., Traversa, M., . . . Colombi, M. (2013). Clinical and molecular characterization of 40 patients with classic ehlers-danlos syndrome: Identification of 18 COL5A1 and 2 COL5A2 novel mutations. Orphanet Journal of Rare Diseases, 8(1) doi:10.1186/1750-1172-8-58
46. Wischmeijer, A., Van Laer, L., Tortora, G., Bolar, N. A., Van Camp, G., Fransen, E., . . . Loeys, B. L. (2013). Thoracic aortic aneurysm in infancy in aneurysms-osteoarthritis syndrome due to a novel SMAD3 mutation: Further delineation of the phenotype. American Journal of Medical Genetics, Part A, 161(5), 1028-1035. doi:10.1002/ajmg.a.35852
47. Baban, A., Torre, M., Costanzo, S., Gimelli, S., Bianca, S., Divizia, M. T., . . . Calevo, M. G. (2012). Familial poland anomaly revisited. American Journal of Medical Genetics, Part A, 158 A(1), 140-149. doi:10.1002/ajmg.a.34370
48. Callea, M., Yavuz, I., Deroma, L., Montanari, M., Clarich, G., Maglione, M., . . . Garavelli, L. (2012). Oral manifestation of goltz-gorlin syndrome in a young girl. Head and Face Medicine, 8 doi:10.1186/1746-160X-8-S1-P8
49. Caputo, V., Cianetti, L., Niceta, M., Carta, C., Ciolfi, A., Bocchinfuso, G., . . . Tartaglia, M. (2012). A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies myhre syndrome. American Journal of Human Genetics, 90(1), 161-169. doi:10.1016/j.ajhg.2011.12.011
50. Garavelli, L., Gargano, G., Simonte, G., Rosato, S., Wischmeijer, A., Melli, N., . . . Neri, G. (2012). Simpson-golabi-behmel syndrome type 1 in a 27-week macrosomic preterm newborn: The diagnostic value of rib malformations and index nail and finger hypoplasia. American Journal of Medical Genetics, Part A, 158 A(9), 2245-2249. doi:10.1002/ajmg.a.35474
51. Malagoli, C., Crespi, C. M., Rodolfi, R., Signorelli, C., Poli, M., Zanichelli, P., . . . Vinceti, M. (2012). Maternal exposure to magnetic fields from high-voltage power lines and the risk of birth defects. Bioelectromagnetics, 33(5), 405-409. doi:10.1002/bem.21700
52. Van Houdt, J. K. J., Nowakowska, B. A., Sousa, S. B., Van Schaik, B. D. C., Seuntjens, E., Avonce, N., . . . Vermeesch, J. R. (2012). Heterozygous missense mutations in SMARCA2 cause nicolaides-baraitser syndrome. Nature Genetics, 44(4), 445-449. doi:10.1038/ng.1105
53. Zampatti, S., Castori, M., Fischer, B., Ferrari, P., Garavelli, L., Dionisi-Vici, C., . . . Brancati, F. (2012). De barsy syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 158 A(4), 927-931. doi:10.1002/ajmg.a.35231
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55. Garavelli, L., Rosato, S., Wischmeijer, A., Gelmini, C., Esposito, A., Mazzanti, L., . . . Riccio, A. (2011). 22q11.2 distal deletion syndrome: Description of a new case with truncus arteriosus type 2 and review. Molecular Syndromology, 2(1), 35-44. doi:10.1159/000334262
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64. Garavelli, L., D'Apice, M. R., Rivieri, F., Bertoli, M., Wischmeijer, A., Gelmini, C., . . . Novelli, G. (2009). Mandibuloacral dysplasia type A in childhood. American Journal of Medical Genetics, Part A, 149(10), 2258-2264. doi:10.1002/ajmg.a.33005
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66. Garavelli, L., Zollino, M., Cerruti Mainardi, P., Gurrieri, F., Rivieri, F., Soli, F., . . . Neri, G. (2009). Mowat-wilson syndrome: Facial phenotype changing with age: Study of 19 italian patients and review of the literature. American Journal of Medical Genetics, Part A, 149(3), 417-426. doi:10.1002/ajmg.a.32693
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Self-declaration affidavit (*Dichiarazione sostitutiva dell'atto di notorietà*)
(according to art.47 and art.19 of D.P.R. n. 445/2000)

I, aware of the consequences of making false statements, falsehood of acts and use of false facts, punishable by law according to art. 76 D.P.R. n. 445/2000 and art. 496 of the Italian Penal Code, under my own responsibility DECLARE that the information herein is true, correct and complete, and that, upon request, I am willing and able to provide an authenticated copy of the titles and documents mentioned.
I authorize management of my personal data as indicated according to regulation UE 2016/679 and previous D.Lgs 196/2003.

La Sottoscritta, ai sensi degli artt. 46 e 47 D.P.R. n. 445/2000, consapevole delle sanzioni penali previste dall'art. 76 D.P.R. n. 445/2000 nel caso di mendaci dichiarazioni, falsità negli atti, uso o esibizione di atti falsi o contenenti dati non più corrispondenti a verità, DICHIARA che quanto sopra riportato corrisponde a verità, e che i titoli e gli allegati sono, su richiesta, disponibili in copia fotostatica conforme agli originali.
La sottoscritta autorizza il trattamento dei dati personali ai sensi del Regolamento UE 2016/679 sul trattamento dei dati personali e del precedente D.Lgs 196/03".

Reggio Emilia, 07-01-2019

Livia Garavelli