Ludovico Graziani

ORCID: https://orcid.org/0000-0002-7217-7388

Sito web: https://www.webofscience.com/wos/author/record/KMY-8263-2024

ESPERIENZA LAVORATIVA

Dirigente Medico • Genetica Clinica

Azienda Ospedaliero Universitaria di Sassari [01/05/2025]

Città: Sassari | **Paese:** Italia

Medico Specialista in formazione • Genetica Medica

Policlinico Tor Vergata [02/2022 – 02/2025]

Città: Roma | **Paese:** Italia

Medico Specialista in formazione • Genetica Medica

Ospedale Pediatrico Bambino Gesù [01/2021 – 02/2022]

Città: Roma | **Paese:** Italia

Medico (LP)

Ausl Romagna • Ospedale Santa Maria delle Croci [11/2020 – 12/2020]

Città: Ravenna | **Paese:** Italia

ISTRUZIONE E FORMAZIONE

Specializzazione Medica • Genetica Medica

Università degli Studi di Tor Vergata [01/2021 – 18/02/2025]

Città: Roma | **Paese:** Italia | **Voto finale:** 70 cum laude

Laurea Magistrale in Medicina e Chirurgia (LM 41)

Università Cattolica del Sacro Cuore [09/2014 – 06/2020]

Città: Roma | **Paese:** Italia | **Voto finale:** 110 summa cum laude

Corso di Alta Formazione in Genetica riproduttiva e prenatale

Università degli Studi di Padova [03/2023 – 12/2023]

Città: Padova | **Paese:** Italia

Diploma di Liceo Scientifico

Liceo Scientifico Statale Fulcieri Paulucci di Calboli [09/2009 – 06/2014]

Città: Forlì | **Paese:** Italia | **Voto finale:** 100

COMPETENZE LINGUISTICHE

Lingua madre: italiano

Altre lingue:

inglese

ASCOLTO C1 **LETTURA** C1 **SCRITTURA** C1

PRODUZIONE ORALE C1 **INTERAZIONE ORALE** C1

Livelli: A1 e A2: Livello elementare B1 e B2: Livello intermedio C1 e C2: Livello avanzato

PUBBLICAZIONI

[2024]
A Novel COL4A5 Pathogenic Variant Joins the Dots in a Family with a Synchronous Diagnosis of Alport Syndrome and Polycystic Kidney Disease.

Graziani L, Minotti C, Carriero ML,...Terracciano A, Novelli A, Novelli G., Genes. 2024; 15(5):597

[2024]
Prenatal identification of a pathogenic maternal FGFR1 variant in two consecutive pregnancies with fetal forebrain malformations.

Graziani L, Nuovo S,... Mappa I, Novelli G. J Matern Fetal Neonatal Med. 2024 Dec;37(1):2344718.

[2024]
Genetic Variability of SOX10-Related Disorders within an Italian Family: Straddling the Line between Kallmann and Waardenburg Syndrome.

Graziani L, Carriero ML, Pozzi F,... Lauro D, Novelli G. Mol Syndromol 2024. Aug;15(4):339-346.

[2024]
Case report: A new de novo 6q21q22.1 interstitial deletion case in a girl with cerebellar vermis hypoplasia and developmental delay and literature review. . doi: 10.3389/fgene.2023.1315291.

Minotti C, Graziani L, Sallicandro E,... Loddo S, Novelli A.Front Genet. 2024 Feb 6;14:1315291

[2024]
Descending necrotizing mediastinitis caused by retro-pharyngeal Eggerthia catenaformis infection.

Graziani A, Tamburini MV, Congestrì F, Graziani L,... Spaggiari R. Germs. 2023 Sep 30;13(3):273-276

[2024]
Co-Inheritance of Pathogenic Variants in PKD1 and PKD2 Genes Determined by Parental Segregation and De Novo Origin: A Case Report.

Graziani L, Zampatti S, Carriero ML,... Giardina E, Novelli G. Genes (Basel). 2023 Aug 6;14(8):1589

[2024]
Clinical variability of Shashi-Pena syndrome: A novel ASXL2 variant associated with overgrowth and minor neurodevelopmental features.

Minotti C., Graziani L.,... Novelli A., Digilio M.C. Mol Syndromol. 2024-09-04 - First online date

[2024]
WDFY3 Haploinsufficiency Is Associated With Autosomal Dominant Neurodevelopmental Disorders and Macrocephaly

Graziani L, Carriero ML, Ferradini V, Conte C, Bengala M, Sangiuolo FC, Novelli G. Clin Genet. 2024 Nov 29. doi: 10.1111/cge.14665. PMID: 39614649.

[2023]
A likely pathogenic ACTG1 variant in a child showing partial phenotypic overlap with Baraitser-Winter syndrome.

Graziani L, Cinnirella G, Ferradini V,... Novelli G. Am J Med Genet A. 2023 Jun;191(6):1565-1569

[2022]
Identifying phenotypic expansions for congenital diaphragmatic hernia plus (CDH+) using DECIPHER data.

Hardcastle A, Berry AM,... Graziani L,... Scott DA. Am J Med Genet A. 2022 Oct;188(10):2958-2968

[2022]
Lumbar spinal canal stenosis: An early sign of amyloid transthyretin related amyloidosis.

Graziani A, Cenni P, Lisi M, Domenicali M, Graziani L. Kardiol Pol. 2022;80(2):218-219

[2021]
Resolution of pulmonary artery thrombosis in patients with moderate COVID-19 disease.

Zanframundo G,... Graziani L. J Community Hosp Intern Med Perspect. 2021 Jun 21;11(4):470-472.

[2021]
Pulmonary artery thrombosis in COVID-19 patients.

Graziani A, Domenicali M, Zanframundo G,... Graziani L. Pulmonology. 2021 May-Jun;27(3):261-263

[2021]
Multimodal assessment in polymicrobial infective endocarditis with silent spondylodiscitis and cerebral embolism.

A Graziani, ED Giudice, S Casolari, M Lisi, L Graziani. Rev Colomb Card. 2021 Nov;28(5):483-488

[2020]
Pulmonary artery thrombosis in home patient with a mild COVID-19 disease.

Graziani A, Domenicali M, Zanframundo G,... Graziani L. Pneumologia 2020 Dec;69(2):103-106

[2020]
Scintigraphy showing the possible progression of transthyretin cardiac amyloidosis.

Graziani A, Del Giudice E, Lisi M, Domenicali M, Graziani L. Kardiol Pol. 2020 Mar 25;78(3):253-254

[2019]
Imaging in the Diagnosis of Transthyretin Cardiac Amyloidosis. AJMHS

Graziani A, Del Giudice E,... Graziani L. Albanian Journal of Medical and Health Sciences 2019; 50

CONFERENZE E SEMINARI

[17/11/2021 – 19/11/2021]
XXIV Congresso Nazionale SIGU

[28/11/2022 – 28/11/2022]
MULTIDISCIPLINARY TEAM MEETINGS of Reference Centre for Marfan Syndrome and Related Disorders. From "the Patient at the center" to "the Patient ... into the Centre"

[07/09/2022 – 09/09/2022]
XXV Congresso Nazionale SIGU

[04/10/2023 – 06/10/2023]
XXVI Congresso Nazionale SIGU

[11/05/2022 – 11/05/2022]
Approccio al Bambino Affetto da Malattia Genetica Rara

[15/04/2021 – 15/04/2021]
Approccio al Bambino Affetto da Malattia Genetica Rara

[06/03/2024 – 06/03/2024]

Approccio al Bambino Affetto da Malattia Genetica Rara

[08/04/2024 – 10/04/2024]
Human Genome Meeting 2024

[01/06/2024 – 04/06/2024] Berlino
European Society of Human Genetics 2024 Conference

[02/10/2024 – 04/10/2024]
XXVII Congresso Nazionale SIGU

INVITED PEER REVIEW

npj Genomic Medicine

Human Genetics

Human Genomics

Plos One

Scientific Reports

Molecular Syndromology

BMC Pediatrics

BMJ Case Reports

Journal of International Medical Research

Discover Medicine

Journal of Rare Diseases

European Journal of Medical Research

Journal of Public Health and Emergency

Quantitative Imaging in Medicine and Surgery

PATENTE DI GUIDA

Patente di guida: B