CURRICULUM VITAE		
INFORMAZIONI PERSONALI		
Nome		Maria Iascone
Indirizzo lavoro		ASST Papa Giovanni XXIII Bergamo

DATI DI SINTESI	
Laurea:	Scienze Biologiche
Disciplina medica:	Genetica Medica
Tipo di incarico attuale:	Responsabile f.f. SSD Laboratorio di Genetica Medica
Totale pubblicazioni:	153 https://pubmed.ncbi.nlm.nih.gov/?term=Iascone+m%5BAuthor%5D&sort=date
H-index:	22 https://www.scopus.com/authid/detail.uri?authorId=26426101900
ORCID	https://orcid.org/0000-0002-4707-212X
Scopus Author ID	26426101900
Researcher ID	AAA-4599-2019
Casistica trattata:	malattie rare con base genetica

ESPERIENZA PROFESSIONALE E CLINICA

Date (da – a)	**FEBBRAIO 2023 – AD OGGI**
Nome e indirizzo del datore di lavoro	USSD Laboratorio di Genetica Medica – Azienda Ospedaliera Papa Giovanni XXIII (già Azienda Ospedaliera Ospedali Riuniti di Bergamo)
Tipo di azienda o settore	Azienda sanitaria ospedaliera
Tipo di impiego	Dirigente Biologo
Principali mansioni e responsabilità	*Responsabile f.f. SSD Laboratorio di genetica Medica*
Date (da – a)	**GENNAIO 2006 – AD OGGI**
Nome e indirizzo del datore di lavoro	USSD Laboratorio di Genetica Medica – Azienda Ospedaliera Papa Giovanni XXIII (già Azienda Ospedaliera Ospedali Riuniti di Bergamo)
Tipo di azienda o settore	Azienda sanitaria ospedaliera
Tipo di impiego	Dirigente Biologo
Principali mansioni e responsabilità	*Responsabile Sezione di Genetica Molecolare*
Date (da – a)	**DICEMBRE 1999 – DICEMBRE 2005**
Nome e indirizzo del datore di lavoro	USC Anatomia Patologica – Azienda Ospedaliera Ospedali Riuniti di Bergamo
Tipo di azienda o settore	Azienda sanitaria ospedaliera
Tipo di impiego	Dirigente Biologo
Principali mansioni e responsabilità	*Responsabile Sezione di Genetica Molecolare*
Date (da – a)	**MARZO 1993 – NOVEMBRE 1999**
Nome e indirizzo del datore di lavoro	Istituto di Fisiologia clinica del CNR di Pisa Sez. di Massa c/o Ospedale G. Pasquinucci Via Aurelia Sud – Loc. Montepepe 54100 Massa Carrara
Tipo di azienda o settore	Istituto di ricerca
Tipo di impiego	Ricercatore a tempo determinato
Principali mansioni e responsabilità	*Responsabile Laboratorio di Biologia Molecolare Cardiovascolare*

ATTIVITÀ SVOLTE ULTIMI 5 ANNI

2018-2023

- Organizzazione e gestione del personale dipendente e non dipendente afferente alla sezione di Genetica Molecolare
- Formazione e Aggiornamento del personale
- Introduzione nuove tecnologie e metodologie Genomiche in diagnostica: Sequenziamento di Nuova Generazione (NGS); automazione dei protocolli di Genomica e dell'estrazione di acidi nucleici
- Gestione del budget economico /centro di costo assegnato alla Sezione
- Organizzazione e gestione dell'attività diagnostica di Genetica Molecolare
- Gestione dei rapporti con i clinici intra- ed extra-ospedale e fuori regione per il miglioramento dei percorsi di diagnostica genetico-molecolare (discussione indicazione alla prescrizione dei test genetici, scelta del test da eseguire, discussione dei risultati e supporto alla comunicazione degli stessi ai pazienti)
- Validazione e firma di tutti i referti di NGS prodotti dalla Sezione
- Validazione di tutti i restanti referti prodotti dalla Sezione
- *Fundraising* per il finanziamento delle borse di studio del personale non dipendente e per la messa a punto delle nuove tecnologie da introdurre in diagnostica
- Gestione delle collaborazioni con altri centri nazionali e internazionali

AREE DI INTERESSE

Malattie genetiche ultra-rare
Malattie Cardiovascolari Ereditarie
Epatopatie Pediatriche Ereditarie
Malattie metaboliche
Innovazioni Tecnologiche del Sequenziamento
Introduzione nella pratica clinica del sequenziamento genomico (whole-exome sequencing, whole-genome sequencing)

ISTRUZIONE

Date (da – a)	Novembre 1995 – Novembre 1999
Nome e tipo di istituto di istruzione o formazione	**Scuola di Specializzazione in Genetica Medica**
	Facoltà di Medicina e Chirurgia – Università degli Studi di Genova
Qualifica conseguita	Specialista in Genetica Medica
Date (da – a)	Gennaio 1994 – Dicembre 1995
Nome e tipo di istituto di istruzione o formazione	**Scuola di Perfezionamento**
	Accademia Nazionale dei Lincei
Date (da – a)	Novembre 1986 – Marzo 1993
Nome e tipo di istituto di istruzione o formazione	**Corso di Laurea in Scienze Biologiche**
	Facoltà di Matematica, Fisica e Scienze Naturali – Università degli Studi di Pisa
Qualifica conseguita	Biologo

PROGETTI DI RICERCA E TRIALS (ULTIMI ANNI)

2023-2026	**Co- Investigator** Progetto di ricerca dal Titolo "Drop-by-Drop: Deciphering the molecular signature in Kleefstra syndrome: proof of principle working model for chromatinopathies" finanziato da Fondazione Regionale per la Ricerca Biomedica (FRRB) Regione Lombardia
2021-2023	**Co- Investigator** Progetto di ricerca dal Titolo ""Sequenziamento del genoma del neonato: fattibilità ed implicazioni cliniche, etiche, psicologiche ed economiche" presentato da Fondazione Telethon (capofila) in partenariato con ASST Papa Giovanni XXIII di Bergamo e UNIAMO Federazione Italiana Malattie Rare onlus con prot. R1.2020.0005132
2021-2023	**Principal Investigator** Progetto di ricerca dal Titolo "Responsible Implementation of Newborns Genome Sequencing: a technical and interpretative feasibility study" GSP21003-Telethon
2018-2019	**Principal Investigator** Progetto di Ricerca: "GENE - Genomic analysis Evaluation NEtwork-Studio prospettico multicentrico costo-efficacia del sequenziamento dell'intero esoma (WES) come primo test genetico in pazienti pediatrici con sospetta malattia genetica -Progetti di innovazione in ambito sanitario e socio sanitario Regione Lombardia, bando ex decreto n. 2713 del 28/02/2018
2017-2019	**Principal Investigator** Progetto di Ricerca: "RARE: Rapid Analysis for Rapid carE -Valutazione dell'utilità clinica del sequenziamento dell'intero esoma (WES) per la diagnosi urgente di malattie genetiche rare in pazienti in età pediatrica in condizioni critiche ricoverati in terapia intensiva neonatale e pediatrica - *PG23 / FROM 2017 Call for Independent Research*.
2011-2013	**Co-Investigator** Progetto di Ricerca "Massively parallel sequencing: integration of genetic data in clinical practice". Progetto Fondazione Cariplo, Ref. 2011-1481
2010-2012	**Co-Investigator** Progetto di Ricerca "Implementation of Genetic diagnosis in clinical setting for hypoplastic left heart syndrome". Regione Lombardia, decreto n. 13465 del 22 dicembre 2010

ELENCO DELLE PUBBLICAZIONI (5 ANNI)

1. The clinical and molecular spectrum of the KDM6B-related neurodevelopmental disorder.
   Rots D, Jakub TE, Keung C, Jackson A, Banka S, Pfundt R, de Vries BBA, van Jaarsveld RH, Hopman SMJ, van

Binsbergen E, Valenzuela I, Hempel M, Bierhals T, Kortüm F, Lecoquierre F, Goldenberg A, Hertz JM, Andersen CB, Kibæk M, Prijoles EJ, Stevenson RE, Everman DB, Patterson WG, Meng L, Gijavanekar C, De Dios K, Lakhani S, Levy T, Wagner M, Wieczorek D, Benke PJ, Lopez Garcia MS, Perrier R, Sousa SB, Almeida PM, Simões MJ, Isidor B, Deb W, Schmanski AA, Abdul-Rahman O, Philippe C, Bruel AL, Faivre L, Vitobello A, Thauvin C, Smits JJ, Garavelli L, Caraffi SG, Peluso F, Davis-Keppen L, Platt D, Royer E, Leeuwen L, Sinnema M, Stegmann APA, Stumpel CTRM, Tiller GE, Bosch DGM, Potgieter ST, Joss S, Splitt M, Holden S, Prapa M, Foulds N, Douzgou S, Puura K, Waltes R, Chiocchetti AG, Freitag CM, Satterstrom FK, De Rubeis S, Buxbaum J, Gelb BD, Branko A, Kushima I, Howe J, Scherer SW, Arado A, Baldo C, Patat O, Bénédicte D, Lopergolo D, Santorelli FM, Haack TB, Dufke A, Bertrand M, Falb RJ, Rieß A, Krieg P, Spranger S, Bedeschi MF, **Iascone** M, Josephi-Taylor S, Roscioli T, Buckley MF, Liebelt J, Dagli AI, Aten E, Hurst ACE, Hicks A, Suri M, Aliu E, Naik S, Sidlow R, Coursimault J, Nicolas G, Küpper H, Petit F, Ibrahim V, Top D, Di Cara F; Genomics England Research Consortium; Louie RJ, Stolerman E, Brunner HG, Vissers LELM, Kramer JM, Kleefstra T.
Am J Hum Genet. 2023 May 13:S0002-9297(23)00132-5. doi: 10.1016/j.ajhg.2023.04.008. Online ahead of print.
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2. *ARF1*-related disorder: phenotypic and molecular spectrum.
de Sainte Agathe JM, Pode-Shakked B, Naudion S, Michaud V, Arveiler B, Fergelot P, Delmas J, Keren B, Poirsier C, Alkuraya FS, Tabarki B, Bend E, Davis K, Bebin M, Thompson ML, Bryant EM, Wagner M, Hannibal I, Lenberg J, Krenn M, Wigby KM, Friedman JR, **Iascone** M, Cereda A, Miao T, LeGuern E, Argilli E, Sherr E, Caluseriu O, Tidwell T, Bayrak-Toydemir P, Hagedorn C, Brugger M, Vill K, Morneau-Jacob FD, Chung W, Weaver KN, Owens JW, Husami A, Chaudhari BP, Stone BS, Burns K, Li R, de Lange IM, Biehler M, Ginglinger E, Gérard B, Stottmann RW, Trimouille A.
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PMID: 37185208

3. De novo variants in CNOT9 cause a neurodevelopmental disorder with or without epilepsy.
von Wintzingerode L, Ben-Zeev B, Cesario C, Chan KM, Depienne C, Elpeleg O, **Iascone** M, Kelley WV, Nassogne MC, Niceta M, Pezzani L, Rahner N, Revencu N, Bekheirnia MR, Santiago-Sim T, Tartaglia M, Thompson ML, Trivisano M, Hentschel J, Sticht H, Abou Jamra R, Oppermann H.
Genet Med. 2023 Apr 20;25(7):100859. doi: 10.1016/j.gim.2023.100859. Online ahead of print.
PMID: 37092538

4. FDXR-associated disease: a challenging differential diagnosis with inflammatory peripheral neuropathy.
Masnada S, Previtali R, Erba P, Beretta E, Camporesi A, Chiapparini L, Doneda C, **Iascone** M, Sartorio MUA, Spaccini L, Veggiotti P, Osio M, Tonduti D, Moroni I.
Neurol Sci. 2023 Apr 12:1-7. doi: 10.1007/s10072-023-06790-0. Online ahead of print.
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5. Prenatal Clinical Findings in *RASA1*-Related Capillary Malformation-Arteriovenous Malformation Syndrome.
Coccia E, Valeri L, Zuntini R, Caraffi SG, Peluso F, Pagliai L, Vezzani A, Pietrangiolillo Z, Leo F, Melli N, Fiorini V, Greco A, Lepri FR, Pisaneschi E, Marozza A, Carli D, Mussa A, Radio FC, Conti B, **Iascone** M, Gargano G, Novelli A, Tartaglia M, Zuffardi O, Bedeschi MF, Garavelli L.
Genes (Basel). 2023 Feb 22;14(3):549. doi: 10.3390/genes14030549.
PMID: 36980822 Free PMC article.

6. The crucial role of titin in fetal development: recurrent miscarriages and bone, heart and muscle anomalies characterise the severe end of titinopathies spectrum.
Di Feo MF, Lillback V, Jokela M, McEntagart M, Homfray T, Giorgio E, Casalis Cavalchini GC, Brusco A, **Iascone** M, Spaccini L, D'Oria P, Savarese M, Udd B.
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7. Menkes disease complicated by concurrent ACY1 deficiency: A case report.
Mauri A, Saielli LA, Alfei E, **Iascone** M, Marchetti D, Cattaneo E, Di Lauro A, Antonelli L, Alberti L, Bonaventura E, Veggiotti P, Spaccini L, Cereda C.
Front Genet. 2023 Mar 2;14:1077625. doi: 10.3389/fgene.2023.1077625. eCollection 2023.
PMID: 36936426 Free PMC article.

8. Chung-Jansen syndrome can mimic Cornelia de Lange syndrome: Another player among chromatinopathies?
Conti B, Rinaldi B, Rimoldi M, Villa R, **Iascone** M, Gangi S, Porro M, Ajmone PF, Colli AM, Mosca F, Bedeschi MF.
Am J Med Genet A. 2023 Jun;191(6):1586-1592. doi: 10.1002/ajmg.a.63164. Epub 2023 Feb 26.
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9. Rock around DYRK1A: Ethnic diversity, clinical challenges.
Moroni A, Pezzani L, Alfei E, Scatigno A, Cereda A, Marzaroli M, Guuva C, Gabbiadini S, Pezzoli L, Marchetti D, Spaccini L, **Iascone** M.
Am J Med Genet A. 2023 May;191(5):1459-1464. doi: 10.1002/ajmg.a.63140. Epub 2023 Feb 11.
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10. *DSP*-Related Cardiomyopathy as a Distinct Clinical Entity? Emerging Evidence from an Italian Cohort.
Di Lorenzo F, Marchionni E, Ferradini V, Latini A, Pezzoli L, Martino A, Romeo F, Iorio A, Bianchi S, **Iascone** M, Calò L,

Novelli G, Mango R, Sangiuolo F.
Int J Mol Sci. 2023 Jan 27;24(3):2490. doi: 10.3390/ijms24032490.
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11. Newborn screening for X-linked adrenoleukodystrophy in Italy: Diagnostic algorithm and disease monitoring.
Bonaventura E, Alberti L, Lucchi S, Cappelletti L, Fazzone S, Cattaneo E, Bellini M, Izzo G, Parazzini C, Bosetti A, Di Profio E, Fiore G, Ferrario M, Mameli C, Sangiorgio A, Masnada S, Zuccotti GV, Veggiotti P, Spaccini L, **Iascone** M, Verduci E, Cereda C, Tonduti D; XALD-NBS Study Group.
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12. SBIDDS Syndrome: A New Spoke of the Epigenetic Machinery Wheel.
Aleo S, Pezzani L, Milani D, Pezzoli L, Marchisio P, **Iascone** M.
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13. Novel Insulin-Like Growth Factor 1 Gene Mutation: Broadening of the Phenotype and Implications for Insulin Resistance.
Giacomozzi C, Martin A, Fernández MC, Gutiérrez M, **Iascone** M, Domené HM, Dominici FP, Bergadá I, Cangiano B, Persani L, Pennisi PA.
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14. Genome-Wide DNA Methylation Profiling Solves Uncertainty in Classifying *NSD1* Variants.
Ferilli M, Ciolfi A, Pedace L, Niceta M, Radio FC, Pizzi S, Miele E, Cappelletti C, Mancini C, Galluccio T, Andreani M, **Iascone** M, Chiriatti L, Novelli A, Micalizzi A, Matraxia M, Menale L, Faletra F, Prontera P, Pilotta A, Bedeschi MF, Capolino R, Baban A, Seri M, Mammì C, Zampino G, Digilio MC, Dallapiccola B, Priolo M, Tartaglia M.
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15. Congenital diaphragmatic hernia in Coffin Siris syndrome: Further evidence from two cases.
Rimoldi M, Rinaldi B, Villa R, Cerasani J, Beltrami B, **Iascone** M, Silipigni R, Boito S, Gangi S, Colombo L, Porro M, Cesaretti C, Bedeschi MF.
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16. A clustering of heterozygous missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder.
Snijders Blok L, Verseput J, Rots D, Venselaar H, Innes AM, Stumpel C, Õunap K, Reinson K, Seaby EG, McKee S, Burton B, Kim K, van Hagen JM, Waisfisz Q, Joset P, Steindl K, Rauch A, Li D, Zackai EH, Sheppard SE, Keena B, Hakonarson H, Roos A, Kohlschmidt N, Cereda A, **Iascone** M, Rebessi E, Kernohan KD, Campeau PM, Millan F, Taylor JA, Lochmüller H, Higgs MR, Goula A, Bernhard B, Velasco DJ, Schmanski AA, Stark Z, Gallacher L, Pais L, Marcogliese PC, Yamamoto S, Raun N, Jakub TE, Kramer JM, den Hoed J, Fisher SE, Brunner HG, Kleefstra T.
HGG Adv. 2022 Nov 1;4(1):100157. doi: 10.1016/j.xhgg.2022.100157. eCollection 2023 Jan 12.
PMID: 36408368 Free PMC article.

17. Dominant ARF3 variants disrupt Golgi integrity and cause a neurodevelopmental disorder recapitulated in zebrafish.
Fasano G, Muto V, Radio FC, Venditti M, Mosaddeghzadeh N, Coppola S, Paradisi G, Zara E, Bazgir F, Ziegler A, Chillemi G, Bertuccini L, Tinari A, Vetro A, Pantaleoni F, Pizzi S, Conti LA, Petrini S, Bruselles A, Prandi IG, Mancini C, Chandramouli B, Barth M, Bris C, Milani D, Selicorni A, Macchiaiolo M, Gonfiantini MV, Bartuli A, Mariani R, Curry CJ, Guerrini R, Slavotinek A, **Iascone** M, Dallapiccola B, Ahmadian MR, Lauri A, Tartaglia M.
Nat Commun. 2022 Nov 11;13(1):6841. doi: 10.1038/s41467-022-34354-x.
PMID: 36369169 Free PMC article.

18. Short stature in PRMT7 Mutations: first evidence of response to growth hormone treatment.
Rodari G, Villa R, Porro M, Gangi S, **Iascone** M, Elli F, Giacchetti F, Profka E, Collini V, Dall'Antonia A, Arosio M, Mantovani G, Bedeschi MF, Giavoli C.
Eur J Hum Genet. 2023 Feb;31(2):195-201. doi: 10.1038/s41431-022-01220-9. Epub 2022 Nov 9.
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19. Delineation of a KDM2B-related neurodevelopmental disorder and its associated DNA methylation signature.
van Jaarsveld RH, Reilly J, Cornips MC, Hadders MA, Agolini E, Ahimaz P, Anyane-Yeboa K, Bellanger SA, van Binsbergen E, van den Boogaard MJ, Brischoux-Boucher E, Caylor RC, Ciolfi A, van Essen TAJ, Fontana P, Hopman S, **Iascone** M, Javier MM, Kamsteeg EJ, Kerkhof J, Kido J, Kim HG, Kleefstra T, Lonardo F, Lai A, Lev D, Levy MA, Lewis MES, Lichty A, Mannens MMAM, Matsumoto N, Maya I, McConkey H, Megarbane A, Michaud V, Miele E, Niceta M, Novelli A, Onesimo R, Pfundt R, Popp B, Prijoles E, Relator R, Redon S, Rots D, Rouault K, Saida K, Schieving J, Tartaglia M, Tenconi R, Uguen K, Verbeek N, Walsh CA, Yosovich K, Yuskaitis CJ, Zampino G, Sadikovic B, Alders M, Oegema R.
Genet Med. 2023 Jan;25(1):49-62. doi: 10.1016/j.gim.2022.09.006. Epub 2022 Nov 1.

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20. Severe Epilepsy and Movement Disorder May Be Early Symptoms of *TMEM106B*-Related Hypomyelinating Leukodystrophy.
Solazzi R, Moscatelli M, Sebastiano DR, Canafoglia L, Pezzoli L, **Iascone** M, Granata T.
Neurol Genet. 2022 Aug 29;8(5):e200022. doi: 10.1212/NXG.0000000000200022. eCollection 2022 Oct.
PMID: 36046422 Free PMC article.

21. Prenatal overgrowth and polydramnios: Would you think about Noonan syndrome?
Beltrami B, Cerasani J, Consales A, Villa R, Resta N, Loconte DC, Boito S, Caschera L, Bassi L, Colombo L, **Iascone** M, Bedeschi MF.
Clin Case Rep. 2022 Aug 22;10(8):e6256. doi: 10.1002/ccr3.6256. eCollection 2022 Aug.
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22. Mystery(n) Phenotypic Presentation in Europeans: Report of Three Further Novel Missense *RNF213* Variants Leading to Severe Syndromic Forms of Moyamoya Angiopathy and Literature Review.
Santoro C, Mirone G, Zanobio M, Ranucci G, D'Amico A, Cicala D, **Iascone** M, Bernardo P, Piccolo V, Ronchi A, Limongelli G, Carotenuto M, Nigro V, Cinalli G, Piluso G.
Int J Mol Sci. 2022 Aug 11;23(16):8952. doi: 10.3390/ijms23168952.
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23. Abnormal B-Cell Maturation and Increased Transitional B Cells in CBL Syndrome.
Saettini F, Coliva TA, Vendemini F, Galbiati M, Bugarin C, Masetti R, Moratto D, Chiarini M, Guerra F, **Iascone** M, Badolato R, Cazzaniga G, Niemeyer C, Flotho C, Biondi A.
Front Pediatr. 2022 Jul 28;10:935951. doi: 10.3389/fped.2022.935951. eCollection 2022.
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24. Atypical, Composite, or Blended Phenotypes: How Different Molecular Mechanisms Could Associate in Double-Diagnosed Patients.
Rosina E, Pezzani L, Pezzoli L, Marchetti D, Bellini M, Pilotta A, Calabrese O, Nicastro E, Cirillo F, Cereda A, Scatigno A, Milani D, **Iascone** M.
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25. Prenatal ultrasound findings associated with PIGW variants: One more piece in the FRYNS syndrome puzzle? PIGW-related prenatal findings.
Ronzoni L, Boito S, Meossi C, Cesaretti C, Rinaldi B, Agolini E, Rizzuti T, Pezzoli L, Silipigni R, Novelli A, **Iascone** M, Persico N, Natacci F.
Prenat Diagn. 2022 Nov;42(12):1493-1502. doi: 10.1002/pd.6204. Epub 2022 Jul 19.
PMID: 35788948 Review.

26. [Clinical pathway on pediatric cardiomyopathies: a genetic testing strategy proposed by the Italian Society of Pediatric Cardiology].
Girolami F, **Iascone** M, Pezzoli L, Passantino S, Limongelli G, Monda E, Rubino M, Adorisio R, Lombardi M, Ragni L, Olivotto I, Favilli S; a nome della Società Italiana di Cardiologia Pediatrica (SICP).
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27. An example of parenchymal renal sparing in the context of complex malformations due to a novel mutation in the PBX1 gene.
Ruscitti F, Cerminara M, **Iascone** M, Pezzoli L, Rosti G, Romano F, Ronchetto P, Martucciello G, Buratti S, Buffelli F, Bocciardi R, Puliti A, Divizia MT.
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28. A missense mutation in DDRGK1 gene associated to Shohat-type spondyloepimetaphyseal dysplasia: Two case reports and a review of literature.
Franceschi R, **Iascone** M, Maitz S, Marchetti D, Mariani M, Selicorni A, Soffiati M, Maines E.
Am J Med Genet A. 2022 Aug;188(8):2434-2437. doi: 10.1002/ajmg.a.62857. Epub 2022 Jun 7.
PMID: 35670300 Review.

29. Family history is key to the interpretation of exome sequencing in the prenatal context: unexpected diagnosis of Basal Cell Nevus Syndrome.
Rinaldi B, Cesaretti C, Boito S, Villa R, Guerneri S, Borzani I, Rizzuti T, Marchetti D, Conte G, Cinnante C, Triulzi F, Persico N, **Iascone** M, Natacci F.
Prenat Diagn. 2022 Jun;42(7):927-933. doi: 10.1002/pd.6171. Epub 2022 May 27.
PMID: 35584264

30. Neuroimaging appearance of hypothalamic hamartomas in monozygotic twins with Pallister-Hall syndrome: case report and review of the literature.
Consales A, Ardemani G, Cinnante CM, Catalano MR, Giavoli C, Villa R, **Iascone** M, Fontana C, Bedeschi MF,

Fumagalli M.
BMC Neurol. 2022 Mar 24;22(1):118. doi: 10.1186/s12883-022-02618-0.
PMID: 35331151 Free PMC article. Review.

31. Mucopolysaccharidosis-Plus Syndrome, a Rapidly Progressive Disease: Favorable Impact of a Very Prolonged Steroid Treatment on the Clinical Course in a Child.
Faraguna MC, Musto F, Crescitelli V, **Iascone** M, Spaccini L, Tonduti D, Fedeli T, Kullmann G, Canonico F, Cattoni A, Dell'Acqua F, Rizzari C, Gasperini S.
Genes (Basel). 2022 Feb 28;13(3):442. doi: 10.3390/genes13030442.
PMID: 35327996 Free PMC article. Review.

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Saettini F, Fazio G, Bonati MT, Moratto D, Massa V, Di Fede E, Castiglioni S, Marchetti D, Chiarini M, Sottini A, **Iascone** M, Cazzaniga G, Imberti L, Biondi A, Gervasini C, Badolato R.
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COMMISSIONI E ALTRI INCARICHI (3 ANNI)

2018 – ad oggi	Valutatore per EMQN (European Molecular Quality Network) per test genetici basati su NGS
2018-2022	Gruppo di Lavoro SIGU (Società Italiana Genetica Medica) per le "*Indicazioni per la refertazione di analisi genetiche eseguite mediante metodica Next-Generation- Sequencing (NGS)",* documento finale pubblicato 7 agosto 2022.
2020	Gruppo di Lavoro CSS (Consiglio Superiore di Sanità) - Sezione I per le scienze omiche. Pubblicazione del documento "*Trasferimento delle Tecniche Omiche nella pratica clinica* (TTO)"

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Il sottoscritto autorizza al trattamento dei dati personali, secondo quanto previsto dal Regolamento UE n. 679/2016 – GDPR General Data Protection Regulation, D.Lgs 196/2003 modificato da D.Lgs 101/2018.”

CITTA’ _____BERGAMO___

DATA____25.11.2022_____

NOME E COGNOME (FIRMA)

Maria Iascone