Curriculum Vitae et Studiorum

Giovanni Battista Ferrero, MD-PhD

Date of birth : June 22, 1964

Place of birth: Torino, Italy

Citizenship: Italian

Address : Thalassemia and Hemoglobinopathies Reference Center
San Luigi Gonzaga University Hospital
Regione Gonzole 10, 10043 Orbassano (Torino), Italy
Email: giovannibattista.ferrero@unito.it

CURRENT POSITION:

2020 – present : Head, Thalassemia and Hemoglobinopathies Reference Center,
San Luigi Gonzaga University Hospital

2020-present: Associate Professor of Pediatrics, Department of Clinical and Biological Sciences
School of Medicine, University of Torino

2014-present**:** Coordinator, Experimental Pediatrics Program
PhD School in Biomedical Sciences and Oncology, University of Torino

PREVIOUS POSITIONS:

2014-2020: Head, Pediatric Genetics Unit, Regina Margherita Children's Hospital, Torino

2015-2020: Associate Professor of Pediatrics, Department of Public Health and Paediatrics,
School of Medicine, University of Torino

2000-2015: Assistant Professor, Department of Paediatrics, School of Medicine, University of Torino

1995-2000: Staff Physician Regina Margherita Children's Hospital, Torino

EDUCATION:

1989: M.D. degree, summa cum laude, University of Torino

1993: PhD degree in Pediatrics, University of Naples / Baylor College of Medicine, Houston,Texas

1994: Residency in Paediatrics, University of Torino

RESEARCH TRAINING

1988-1991: Predoctoral training, Department of Biomedical Sciences, School of Medicine, University of Torino

Supervisor: Prof. Irma Dianzani, Research project: Molecular basis of phenylketonuria in Italy.

1991-1993: PhD student Dep. of Human Genetics, Baylor College of Medicine, Houston, Texas.

Supervisor: Prof. Andrea Ballabio. Research project: Positional cloning of disease genes

1994: Visiting Research Associate, Dept. of Human Genetics, Baylor College of Medicine, Houston, Texas.

Supervisor Prof. Brett Casey. Research project: Positional cloning of ZIC3 gene

RESEARCH FIELD

Prof. Ferrero is a Pediatrician and a Clinical Geneticist committed to the clinical and molecular characterization of rare diseases. During the doctoral training at Baylor College of Medicine, Houston (Texas), he contributed to the discovery of several disease genes, exploiting Positional cloning techniques.

Afterwards he developed a comprehensive approach to pediatric rare and complex diseases in the Clinical Genetics Unit of Regina Margherita Children's Hospital in Torino. Taking advantage of an international network he has been able to describe new molecular mechanisms and genotype/pehenotype correlations in malformative syndromes, such as Beckwith-Wiedemann Syndrome and Noonan Syndrome. The large cohorts of patients followed-up have allowed to define specific strategies to improve the clinical approach to rare disease in childhood.

In the last few years he has investigated the genetics of intellectual disabilities and autism, describing rare causes of neurodevelopmental defects, collaborating with the Autism Sequencing Consortium, an international collaborative study coordinated by the Icahn School of Medicine at Mount Sinai, New York.

Recently, he has been appointed as Head of the Thalassemia and Hemoglobinopathies Reference Center at the San Luigi Gonzaga University Hospital, where he coordinates a number of Clinical studies exploring new therapeutic approach for these common mendelian disorders.

The intersection between genetics, genomics and clinical care of patients affected by rare diseases can be considered the main focus of his entire career.

SCIENTIFIC AWARDS

1997: "Federico Marsico" Award of the Italian Society of Paediatric Cardiology

2007: Best Platform Presentation Award of the Italian Society of Pediatric Genetics (SIMGEPED)

2009: Poster Award of the European Society of Human Genetics

2011: Best Platform Presentation Award of the Italian Society of Human Genetics (SIGU)

2018: European Society of Human Genetics Award for the most cited Paper of the European Journal of Human Genetics in year 2016: (Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome. Eur J Hum Genet. 2016 Feb;24(2):183-90.

GRANTS AWARDED

1996. Fondazione Telethon: Molecular genetics of X-linked heterotaxy, as P.I.

1999. Compagnia di San Paolo, Torino: Approccio molecolare alle malformazioni congenite: i difetti di lateralizzazione e le craniostenosi, as P.I.

1999. MURST: Approccio genetico e biomolecolare ai difetti di lateralizzazione dell'uomo, as head of Research Unit

2000. MURST: Basi metaboliche e molecolari dei difetti congeniti, as head of Research Unit

2007. Regione Piemonte, Ricerca Sanitaria Finalizzata: Patogensi molecolare dela Sindrome di Noonan: un modello di predisposizione genetica all'insorgenza di tumori ematologici, as PI

2007. PRIN : Genomic Imprinting defects in growth disorders and cancer, as head of Research Unit

2008 - Regione Piemonte, Ricerca Sanitaria Finalizzata: Selezione e follow-up di pazienti affetti da Sindrome di Beckwith-Wiedemann ed analisi molecolare della regione 11p15 con tecnica MS-MLPA, as PI

2008 - Fondazione Cassa Risparmio di Torino, Progetto Alfieri: Analisi genomica con CGH-Array in pazienti affetti da Ritardo mentale sindromico come strumento per l'individuazione di geni coinvolti nello sviluppo del sistema nervoso centrale dell'uomo, as PI

2009 PRIN: Novel approaches to the study of teh growth disorders associated with genomic imprinting, as head of Research Unit

2015, PRIN: Molecular genetics and new directions for clinical management of growth disorders associated with genomic imprinting, as as head of Research Unit

2018, Compagnia di San Paolo, Torino: Sviluppo della banca dati Nazionale della Sindrome di Noonan e delle RASopatie correlate, as PI

PROFESSIONAL MEMBERSHIPS

Member of the Societa' Italiana di Pediatria (SIP)

Member of the Società Italiana Genetica Umana (SIGU)

Member and founder of the Società Italiana Malattie Genetiche Pediatriche e Disabilità Congenite (SIMGEPED)

Member of the European Society of Human Genetics (ESHG)

Associazione Italiana Sindrome di Noonan e RASopatie, Member of the Scientific Committee

Associazione Italiana Sindrome di Beckwith Wiedemann, Member of the Scientific Committee

SELECTION OF RELEVANT PUBLICATIONS

Author of 150 publications on research journals. Citation metrics (Scopus): Total citations: 5259, H-Index: 38

1: Barbero U, Fornari F, Gagliardi M, Fava A, Giorgi M, Alunni G, Gaglioti CM, Piga A, **Ferrero GB**, Longo F.

Myocardial longitudinal strain as the first heraldof cardiac impairment in very early iron overload state: an echocardiography and biosusceptometry study on beta-thalassemia patients. **Am J Cardiovasc Dis**. 2021

Oct 25;11(5):555-563.

2: COVID-19 Host Genetics Initiative (including **Ferrero GB**)

Mapping the human genetic architecture of COVID-19. **Nature**. 2021 Dec;600(7889):472-477.

3: Bryant L, Li D, Cox SG, …, **Ferrero GB**, …, Bhoj EJ.

Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentifiedneurodegenerative disorder in 46 patients. **Sci Adv**. 2020 Dec 2;6(49):eabc9207.

4: Libraro A, D'Ascanio V, Cappa M, Chiarito M, Digilio MC, Einaudi S, Grandone A, Maghnie M, Mazzanti L, Mussa A, Patti G, Scarano E, Spinuzza A, Vannelli S, Wasniewska MG, **Ferrero GB**, Faienza MF.

Growth in Children With Noonan Syndromeand Effects of Growth Hormone Treatment on Adult Height. **Front Endocrinol**(Lausanne). 2021 Dec 22;12:761171.

5: Longo F, Piolatto A, **Ferrero GB**, Piga A.

Ineffective Erythropoiesis in β-Thalassaemia: Key Steps and Therapeutic Options by Drugs. **Int J Mol Sci**, 2021 Jul 5; 22 (13) : 7229

6: Piolatto A, Berchialla P, Allegra S, De Francia S, Ferrero GB, Piga A, Longo F.

Pharmacological and clinical evaluation of deferasirox formulations for treatment tailoring. **Sci Rep** 2021 Jun 15; 11 (1) : 12581

7: Carli D, De Pellegrin M, Franceschi L, Zinali F, Paonessa M, Spolaore S, Cardaropoli S, Cravino M, Marcucci L, Andreacchio A, Resta N, Ferrero GB, Mussa A

Evolution over Time of Leg Length Discrepancy in Patients with Syndromic and Isolated Lateralized Overgrowth. **J Pediatr** 2021 Jul; 234: 123-127.

8: Carli D, Giorgio E, Pantaleoni F, Bruselles A, Barresi S, Riberi E, Licciardi F, Gazzin A, Baldassarre G, Pizzi S, Niceta M, Radio FC, Molinatto C, Montin D, Calvo PL, Ciolfi A, Fleischer N, **Ferrero GB**, Brusco A, Tartaglia M.

NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. **Hum Mutat**. 2019 Jun;40(6):721-728.

9: Brioude F, Kalish JM, Mussa A, Foster AC, Bliek J, **Ferrero GB,** Boonen SE, ColeT, Baker R, Bertoletti M, Cocchi G, Coze C, De Pellegrin M, Hussain K, Ibrahim A,Kilby MD, Krajewska-Walasek M, Kratz CP, Ladusans EJ, Lapunzina P, Le Bouc Y,Maas SM, Macdonald F, Õunap K, Peruzzi L, Rossignol S, Russo S, Shipster C, Skórka A, Tatton-Brown K, Tenorio J, Tortora C, Grønskov K, Netchine I, Hennekam RC, Prawitt D, Tümer Z, Eggermann T, Mackay DJG, Riccio A, Maher ER.

Expert consensus document: Clinical and molecular diagnosis, screening and management ofBeckwith-Wiedemann syndrome: an international consensus statement**. Nat Rev Endocrinol**. 2018 Apr;14(4):229-249.

10: Mussa A, Molinatto C, Cerrato F, Palumbo O, Carella M, Baldassarre G, Carli D, Peris C, Riccio A, **Ferrero GB.** Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. **Pediatrics.** 2017 Jul;140(1). pii: e20164311. doi:10.1542/peds.2016-4311. Epub 2017 Jun 20.

11: Di Gregorio E, Riberi E, Belligni EF, Biamino E, Spielmann M, Ala U, Calcia A,Bagnasco I, Carli D, Gai G, A, Naretto VG, Sirchia F, Sorasio L, Ungari S, Zonta A, Zacchetti G, Talarico F, Pappi P, Cavalieri S, Giorgio E, Mancini C, Ferrero M, Brussino A, Savin E, Gandione M, Pelle A, Giachino DF, De Marchi M, Restagno G, Provero P,Cirillo Silengo M, Grosso E, Buxbaum JD, Pasini B, De Rubeis S, Brusco A, **Ferrero GB**.

Copy number variants analysis in a cohort of isolated and syndromicdevelopmental delay/intellectual disability reveals novel genomic disorders,position effects and candidate disease genes. **Clin Genet**. 2017 Oct;92(4):415-422.

12: Pannone L, Bocchinfuso G, Flex E, Rossi C, Baldassarre G, Lissewski C,Pantaleoni F, Consoli F, Lepri F, Magliozzi M, Anselmi M, Delle Vigne S, Sorge G,Karaer K, Cuturilo G, Sartorio A, Tinschert S, Accadia M, Digilio MC, Zampino G, De Luca A, Cavé H, Zenker M, Gelb BD, Dallapiccola B, Stella L, **Ferrero GB**,Martinelli S, Tartaglia M.

Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. **Hum Mutat.** 2017Apr;38(4):451-459.

13: Mussa A, Molinatto C, Baldassarre G, Riberi E, Russo S, Larizza L, Riccio A, **Ferrero GB**.

Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review andMeta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol. **J Pediatr**. 2016 Sep;176:142-149.

14: Giorgio E, Ciolfi A, Biamino E, Caputo V, Di Gregorio E, Belligni EF, Calcia A, Gaidolfi E, Bruselles A, Mancini C, Cavalieri S, Molinatto C, Cirillo Silengo M, **Ferrero GB**, Tartaglia M, Brusco A.

Whole exome sequencing is necessary toclarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. **Am J Med Genet A**. 2016 Jul;170(7):1772-9.

15: Mussa A, Russo S, De Crescenzo A, Freschi A, Calzari L, Maitz S, Macchiaiolo M, Molinatto C, Baldassarre G, Mariani M, Tarani L, Bedeschi MF, Milani D, Melis D, Bartuli A, Cubellis MV, Selicorni A, Cirillo Silengo M, Larizza L, Riccio A**, Ferrero GB**

(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome**. Eur J Hum Genet**. 2016 Feb;24(2):183-90.

16: Baldassarre G, Mussa A, Banaudi E, Rossi C, Tartaglia M, Silengo M, **FerreroGB.**

Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly)missense mutation**. Am J Med Genet** A. 2014 Dec;164A(12):3120-5.

17: Mussa A, Russo S, De Crescenzo A, Chiesa N, Molinatto C, Selicorni A,Richiardi L, Larizza L, Silengo MC, Riccio A, **Ferrero GB**.

Prevalence of Beckwith-Wiedemann syndrome in North West of Italy. **Am J Med Genet A**. 2013 Oct;161A(10):2481-6.

18: Timeus F, Crescenzio N, Baldassarre G, Doria A, Vallero S, Foglia L, Pagliano S, Rossi C, Silengo MC, Ramenghi U, Fagioli F, Cordero di Montezemolo L, **Ferrero GB.**

Functional evaluation of circulating hematopoietic progenitors in Noonan syndrome. **Oncol Rep** 2013;30(2):553-9.

19: **Ferrero GB**, Picco G, Baldassarre G, Flex E, Isella C, Cantarella D, Corà D, Chiesa N, Crescenzio N, Timeus F, Merla G, Mazzanti L, Zampino G, Rossi C,Silengo M, Tartaglia M, Medico E.

Transcriptional hallmarks of Noonan syndromeand Noonan-like syndrome with loose anagen hair. **Hum Mutat**. 2012 Apr;33(4):703-9.

20: Chiesa N, De Crescenzo A, Mishra K, Perone L, Carella M, Palumbo O, Mussa A, Sparago A, Cerrato F, Russo S, Lapi E, Cubellis MV, Kanduri C, Cirillo Silengo M, Riccio A, **Ferrero GB.**

The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwith-Wiedemann syndrome and Silver-Russell syndrome cases**. Hum Mol Genet**. 2012 Jan 1;21(1):10-25.

21: Baldassarre G, Mussa A, Dotta A, Banaudi E, Marinosci A, Rossi C, Tartaglia M, Silengo M, **Ferrero GB**.

Prenatal features of Noonan syndrome:prevalence and prognostic value. **Prenat Diagn**. 2011;31(10):949-54.

22: Martinelli S, De Luca A, Stellacci E, Rossi C, Checquolo S, Lepri F, CaputoV, Silvano M, Buscherini F, Consoli F, Ferrara G, Digilio MC, Cavaliere ML, vanHagen JM, Zampino G, van der Burgt I, **Ferrero GB**, Mazzanti L, Screpanti I, YntemaHG, Nillesen WM, Savarirayan R, Zenker M, Dallapiccola B, Gelb BD, Tartaglia M.

Heterozygous germline mutations in the CBL tumor-suppressor gene cause a Noonan syndrome-like phenotype. **Am J Hum Genet**. 2010 Aug 13;87(2):250-7.

23: Cordeddu V, Di Schiavi E, Pennacchio LA, Ma'ayan A, Sarkozy A, Fodale V,Cecchetti S, Cardinale A, Martin J, Schackwitz W, Lipzen A, Zampino G, MazzantiL, Digilio MC, Martinelli S, Flex E, Lepri F, Bartholdi D, Kutsche K, **Ferrero GB**,Anichini C, Selicorni A, Rossi C, Tenconi R, Zenker M, Merlo D, Dallapiccola B,Iyengar R, Bazzicalupo P, Gelb BD, Tartaglia M.

Mutation of SHOC2 promotesaberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. **Nat Genet.** 2009 Sep;41(9):1022-6.

24: Sarkozy A, Carta C, Moretti S, Zampino G, Digilio MC, Pantaleoni F, SciolettiAP, Esposito G, Cordeddu V, Lepri F, Petrangeli V, Dentici ML, Mancini GM,Selicorni A, Rossi C, Mazzanti L, Marino B, **Ferrero GB**, Silengo MC, Memo L,Stanzial F, Faravelli F, Stuppia L, Puxeddu E, Gelb BD, Dallapiccola B, TartagliaM.

Germline BRAF mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: molecular diversity and associated phenotypic spectrum. **Hum Mutat**.2009 Apr;30(4):695-702.

Torino, 20 April 2022 ,