CURRICULUM VITAE

- Luigi Boccuto, born in Catanzaro, Italy. Mobile phone: +1(864)344-4331; e-mail: lboccut@clemson.edu.

- 1996: secondary school diploma *summa cum laude* at the Liceo Classico "P.Galluppi" of Catanzaro.

- From 1999 to 2002 internal student at the Institute of Medical Genetics, Catholic University of Sacred Heart (UCSC), Rome, Italy.

- 2002: Medical degree, *summa cum laude*, at the Catholic University of Sacred Heart (UCSC), Rome, Italy, with the graduation thesis "Clinical-biological criteria for the identification of individuals carrying mutations of the *MSH6* gene, responsible for hereditary non-polyposis colorectal cancers".

- From 2002 to 2003 internal doctor at the Institute of Medical Genetics at UCSC.

- Since March 2003: resident in Medical Genetics at UCSC, Rome, Italy, vote of 90/100.

- June 2003: First Certificate in English by Cambridge University (Place of entry: Rome, Italy).

- October 2003: "Gianmichele Laccetti" award, for the best Italian thesis in genetic oncology, academic year 2002-03.

- 2006: Post-doc degree of specialization, *summa cum laude*, at Specialization School of Medical Genetics, Catholic University of Sacred Heart (UCSC), Rome, Italy, with the graduation thesis "Reversibility of epigenetic silencing in cancer cell lines".

- January 2007- June 2010: Research Scholar at the Greenwood Genetic Center, Greenwood, SC.

- July 2010 – December 2013: Staff Scientist at the Greenwood Genetic Center, Greenwood, SC.

- January 2014 – today: Assistant Research Scientist at the Greenwood Genetic Center, Greenwood, SC.

- April 2016 – today: Clinical Assistant Professor at the Clemson University School of Health Research, Clemson, SC.

- September 2017 – October 2019: Chief Scientific Officer at STALICLA, SA, Geneva, Switzerland.

- March 2018 – today: Research Assistant Professor of Pediatrics in the Department of Pediatrics with the University of South Carolina School of Medicine, Columbia, SC.

- May 2020 – today: Medical Collaborator and Genetist Consultant at Dante Labs srl, L'Aquila, Italy.

- Co-author of the following book chapters:
    - **Boccuto L**, Neri G: Cardiomiopatie e cardiopatie aritmogene ereditarie. In Genetica umana e medica, Neri G, Genuardi M. Elsevier Srl, 2007: pp. 343-354.
    - **Boccuto L**, Neri G: Cardiomiopatie primarie ereditarie. In Genetica umana e medica seconda edizione, Neri G, Genuardi M. Elsevier Srl, 2010: pp. 337-354.
    - **Boccuto L**, Neri G: Cardiomiopatie primarie ereditarie. In Genetica umana e medica terza edizione, Neri G, Genuardi M. Elsevier Srl, 2014: pp. 337-354.
    - Schwartz CE, **Boccuto L**: Genetics of X-linked intellectual disability. In: Sala C, Verpelli C, eds., Neuronal and Synaptic Dysfunction in Autism Spectrum Disorder and Intellectual Disability. San Diego: Academic Press, 2016: pp. 25-41.
    - Phelan K, **Boccuto L**, Sarasua S: Phelan-McDermid syndrome: clinical aspects. In: Sala C, Verpelli C, eds., Neuronal and Synaptic Dysfunction in Autism Spectrum Disorder and Intellectual Disability. San Diego: Academic Press, 2016: pp. 347-364.
    - Gennarelli M, **Boccuto L**: Autism and major psychosis. In Genetica umana e medica quarta edizione, Neri G, Genuardi M, Elsevier Srl, ISBN 978-88-214-4392-3, 2017: 323-334.
    - Phelan K, Rogers RC, **Boccuto L**: Phelan-McDermid Syndrome. 2005 May 11 [Updated 2018 Jun 7]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1198/

- Regional Editor for North America of the journal "Reviews on Recent Clinical Trials" since April 2017.

- Member of the scientific advisory board of the 5th Italian edition of the Merck Manual of Diagnosis and Therapy (translated from the 18th English version) issued by Raffaello Cortina Editore Srl, Milan, 2008. ISBN: 9788847007079.

- Member of the scientific advisory board of the 6th Italian edition of the Merck Manual of Diagnosis and Therapy (translated from the 19th English version) issued by Raffaello Cortina Editore Srl, Milan, 2016. ISBN: 9780911910193.

- Reviewer since 2010 for:

    - BMC Medical Genetics;
    - PLOS One;
    - Clinical Genetics;

- Research in Developmental Disabilities;
- European Journal of Human Genetics;
- Research in Autism Spectrum Disorder;
- Annals of Neurology;
- European Journal of Gastroenterology and Hepatology;
- Universal Journal of Clinical Medicine;
- Metabolic Brain Disease;
- Reviews on Recent Clinical Trials;
- The Application of Clinical Genetics;
- Horizon Research Publishing;
- JSM Invitro Fertilization;
- International Journal of Molecular Science;
- Computer in Biology and Medicine;
- Frontiers Oncology;
- Journal of Clinical Medicine.

- Member of the American Society of Human Genetics since 2013.

- External grant reviewer for Telethon Foundation since 2017.

- Member of the Scientific Committee of the Italian Phelan-McDermid Foundation since 2017.

- Working experience in molecular genetics of inherited colorectal cancer, in particular on sequence analysis of *DAPK* and *MSH6* genes and hyper-methylation of promoter of *MLH1* gene, and on familiar breast cancer. Working experience on genetic screening of candidate genes for autism and intellectual disabilities (*SHANK3, EGR2, SLC9A6, ARC, RPL10, GJB6, CRYL1, SLC3A2, SLC7A5, SLC7A8, ST3GAL5, CATSPER2, QPRT, IB2, ACR, RABL2B, SMS, SLC6A8, SULT4A1, ARHGAP8, ALG12*), overgrowth/malformation syndromes (*AKT1*, *AKT2*, *AKT3*, *PIK3CA*, *PIK3R2*, *PDPK1, PIK3CB, PIK3CD, PIK3R1, PIK3R3, PIK3R5*), and metabolic assays on cell lines from patients with ID, ASDs, schizophrenia, and overgrowth. Good practice with molecular genetics techniques, cell cultures, and metabolic arrays.

- Working experience in clinical genetics, especially on overgrowth, intellectual disability (ID), and autism spectrum disorders (ASDs).

- Didactic experience as tutor of summer interns at the Greenwood Genetic Center since 2009, during which period he has directly supervised 10 official interns and 13 volunteer students.

- Lecturer at the Greenwood Genetic Center Teacher Course since 2014 and at the Greenwood Genetic Center Summer High School Camp since 2018.

- Member of the thesis committees for two Ph.D. candidates at the Healthcare Genetics Program of Clemson University.

AWARDS

1. Named one of 2017 Leading Physicians of the World in the field of Genetics as Top Research Scientist.

2. Executive Award 2017 as Best Research Scientist in South Carolina.

3. Best poster at the 2016 Phelan-McDermid Syndrome International Family Conference, July 19-23, 2016, Orlando, FL for "*New approaches in the characterization of genotype/phenotype correlation in Phelan-McDermid syndrome*."

4. Best poster at the meeting "Synaptopathies in Neurodevelopmental Disorders: SHANK Mutations as a window into Synaptic Function", Washington, DC, USA, November 13-14, 2014, for *"Genotype-phenotype characterization of the neurobehavioral presentation in the Phelan-McDermid syndrome: SHANK3 and beyond"*.

5. Young Investigator Award at the Second International Phelan-McDermid Syndrome Symposium, Orlando, Florida, USA July 25-26 2012, for *"The role of SHANK3 and other 22q13.33 genes in PMS patients without chromosomal rearrangements"*.

6. Best poster at the First International Phelan-McDermid Syndrome Symposium, New York, New York, USA 3-4 March 2011, for *"Genotype-Phenotype Correlation Study in Phelan-McDermid Syndrome"*.

7. "Gianmichele Laccetti" 2003 award, for the best Italian thesis in genetic oncology, academic year 2002-03.

PATENTS AND GRANTS

- Co-inventor of the patent "Determination of Decreased Metabolism of Tryptophan in the Diagnosis of Autism Spectrum Disorders", attorney docket No. GGC-4-P, Customer number 22827, patent number US 9,164,106 B2, date October 20th, 2015.

- Co-P.I. of the R21 grant "Reduced NADH Production in the Presence of Tryptophan as a Biomarker of Autism Spectrum Disorders" funded from the Eunice Kennedy Shriver National Institute Of Child Health & Human Development and NICHD (R21-HD072473-01), 2012-2014.

- P.I. of the "Investigation of aberrant tryptophan metabolism as a biochemical basis for Autism Spectrum Disorders (ASDs)" grant in collaboration con Clemson University, funded by Self Regional Healthcare (SRHC), 2014-2017.

- Co-P.I. of the "Analysis of iPS cells and differentiated neuronal cells from patients with Autism Spectrum Disorders" grant in collaboration con Clemson University, funded by Self Regional Healthcare (SRHC), 2014-2017.

- Co-investigator of the Simon Center for Social Brain Support Seed Grant "Characterization of the role of the SHANK3 region in Autism Spectrum Disorder in the Phelan-McDermid syndrome" funded by the Simons Foundation Autism Research Initiative (SFARI), 2013-2016.

- Co-P.I. of the "Intersection of epigenetic regulation and mitochondrial function in autism" SC EPSCoR/IDeA Stimulus Research Grant, number 4-2017, 2018-2020.

- Co-P.I. of the "Development of an Autism Spectrum Disorder Screening Test based upon Metabolic Profiling of Fresh Blood Samples", STTR grant from NIH, number R41MH115642, 2018-2020.

BOOKS AND BOOK CHAPTERS

- Neri G, **Boccuto L**, Stevenson R: Overgrowth syndromes. A clinical guide. Oxford Press, 2019.

CHAPTERS

- **Boccuto L**, Neri G, Stevenson R: Overview on Overgrowth and Overgrowth Syndromes. In Overgrowth syndromes. A clinical guide. Oxford Press, 2019: pp. 1-17.

- **Boccuto L**, Neri G: Cardiomiopatie e cardiopatie aritmogene ereditarie. In Genetica umana e medica, Neri G, Genuardi M. Elsevier Srl, 2007: pp. 343-354.

- **Boccuto L**, Neri G: Cardiomiopatie primarie ereditarie. In Genetica umana e medica seconda edizione, Neri G, Genuardi M. Elsevier Srl, 2010: pp. 337-354.

- **Boccuto L**, Neri G: Cardiomiopatie primarie ereditarie. In Genetica umana e medica terza edizione, Neri G, Genuardi M. Elsevier Srl, 2014: pp. 337-354.

- Schwartz CE, **Boccuto L**: Genetics of X-linked intellectual disability. In: Sala C, Verpelli C, eds., Neuronal and Synaptic Dysfunction in Autism Spectrum Disorder and Intellectual Disability. San Diego: Academic Press, 2016: pp. 25-41.

- Phelan K, **Boccuto L**, Sarasua S: Phelan-McDermid syndrome: clinical aspects. In: Sala C, Verpelli C, eds., Neuronal and Synaptic Dysfunction in Autism Spectrum Disorder and Intellectual Disability. San Diego: Academic Press, 2016: pp. 347-364.

- Gennarelli M, **Boccuto L**: Autism and major psychosis. In Genetica umana e medica quarta edizione, Neri G, Genuardi M, Elsevier Srl, ISBN 978-88-214-4392-3, 2017: 323-334.

- **Boccuto L**, Neri G, Stevenson R: Overview on overgrowth and overgrowth syndromes. In Overgrowth syndromes. A clinical guide, Neri G, Boccuto L, Stevenson R, Oxford Press, 2019.

- Phelan K, Rogers C, **Boccuto L**: Deletion 22q13 syndrome: Phelan-McDermid syndrome. In Management of Genetic Syndromes. Third Edition, edited by Cassidy S and Allanson J, John Wiley & Sons, 2019.

PUBBLICATIONS – h-index: 19 – i10-index: 28 (total citations: 1408)

1. *"Potential role of fecal gluten immunogenic peptides to assess dietary compliance in celiac patients."*
   Larussa T, **Boccuto L**, Luzza F, Abenavoli L.
   Minerva Gastroenterol Dietol. 2020 May 13. doi: 10.23736/S1121-421X.20.02710-5.

2. *"SARS-CoV-2 Pandemic: Review of the Literature and Proposal for Safe Autopsy Practice."*
   Aquila I, Sacco MA, Abenavoli L, Malara N, Arena V, Grassi S, Ausania F, **Boccuto L**, Ricci C, Gratteri S, Oliva A, Ricci P.
   Arch Pathol Lab Med. 2020 May 8. doi: 10.5858/arpa.2020-0165-SA.

3. *"Epidemiology of Coronavirus Disease Outbreak: The Italian Trends."*
   Abenavoli L, Cinaglia P, Luzza F, Gentile I, **Boccuto L**.
   Rev Recent Clin Trials. 2020 Apr 7. doi: 10.2174/1574887115999200407143449.

4. *"Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder."*
   Cascio L, Chen CF, Pauly R, Srikanth S, Jones K, Skinner CD, Stevenson RE, Schwartz CE, **Boccuto L**.
   Mol Genet Genomic Med. 2020 Jan;8(1):e1036. doi: 10.1002/mgg3.1036.

5. *"Gut Microbiota and Obesity: A Role for Probiotics."*
   Abenavoli L, Scarpellini E, Colica C, **Boccuto L**, Salehi B, Sharifi-Rad J, Aiello V, Romano B, De Lorenzo A, Izzo AA, Capasso R.
   Nutrients. 2019 Nov 7;11(11). pii: E2690. doi: 10.3390/nu11112690.

6. *"The Skin in Celiac Disease Patients: The Other Side of the Coin."*

Abenavoli L, Dastoli S, Bennardo L, **Boccuto L**, Passante M, Silvestri M, Proietti I, Potenza C, Luzza F, Nisticò SP.
Medicina (Kaunas). 2019 Sep 9;55(9). pii: E578. doi: 10.3390/medicina55090578. Review.

7. *"Chemical Effect of Bisphenol A on Non-Alcoholic Fatty Liver Disease."*
Dallio M, Diano N, Masarone M, Gravina AG, Patanè V, Romeo M, Di Sarno R, Errico S, Nicolucci C, Abenavoli L, Scarpellini E, **Boccuto L**, Persico M, Loguercio C, Federico A.
Int J Environ Res Public Health. 2019 Aug 28;16(17). pii: E3134. doi: 10.3390/ijerph16173134. Review.

8. *"Diet and Non-Alcoholic Fatty Liver Disease: The Mediterranean Way."*
Abenavoli L, **Boccuto L**, Federico A, Dallio M, Loguercio C, Di Renzo L, De Lorenzo A.
Int J Environ Res Public Health. 2019 Aug 21;16(17). pii: E3011. doi: 10.3390/ijerph16173011. Review.

9. *"Constitutive activation of the PI3K-AKT pathway and cardiovascular abnormalities in an individual with Kosaki overgrowth syndrome."*
Zarate YA, **Boccuto L**, Srikanth S, Pauly R, Ocal E, Balmakund T, Hinkle K, Stefans V, Schaefer GB, Collins RT 2nd.
Am J Med Genet A. 2019 Jun;179(6):1047-1052.

10. *"Nonalcoholic fatty liver disease in obese adolescents: the role of genetic polymorphisms."*
Abenavoli L, **Boccuto L**.
Hepatobiliary Surg Nutr. 2019 Apr;8(2):179-180.

11. *"Genome-scale network model of metabolism and histone acetylation reveals metabolic dependencies of histone deacetylase inhibitors."*
Shen F, **Boccuto L**, Pauly R, Srikanth S, Chandrasekaran S.
Genome Biol. 2019 Mar 1;20(1):49.

12. *"Variability in Phelan-McDermid syndrome: The impact of the PNPLA3 p.I148M polymorphism."*
Boccuto L, Abenavoli L, Cascio L, Srikanth S, DuPont B, Mitz AR, Rogers RC, Phelan K.
Clin Genet. 2018 Dec;94(6):590-591.

13. *"Obeticholic Acid: A New Era in the Treatment of Nonalcoholic Fatty Liver Disease."*
Abenavoli L, Falalyeyeva T, **Boccuto L**, Tsyryuk O, Kobyliak N.
Pharmaceuticals (Basel). 2018 Oct 11;11(4). pii: E104.

14. *"Is possible to detect nonalcoholic fatty liver disease by a new index including single nucleotide polymorphisms (SNPs)?"*
Abenavoli L, **Boccuto L**.
Ann Transl Med. 2018 Sep;6(18):366.

15. *"Beneficial effects of probiotic combination with omega-3 fatty acids in NAFLD: a randomized clinical study."*
Kobyliak N, Abenavoli L, Falalyeyeva T, Mykhalchyshyn G, **Boccuto L**, Kononenko L, Kyriienko D, Komisarenko I, Dynnyk O.
Minerva Med. 2018 Sep 13.

16. *"Health benefits of Mediterranean diet in nonalcoholic fatty liver disease."*
Abenavoli L, Di Renzo L, **Boccuto L**, Alwardat N, Gratteri S, De Lorenzo A.
Expert Rev Gastroenterol Hepatol. 2018 Jul 23.

17. *"Phelan-McDermid Syndrome".*
Phelan K, Rogers RC, **Boccuto L**.
2005 May 11 [Updated 2018 Jun 7]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1198/

18. *"Hepatitis C virus-induced hepatocellular carcinoma: a narrative review".*
Ružić M, Pellicano R, Fabri M, Luzza F, **Boccuto L**, Brkić S, Abenavoli L.
Panminerva Med. 2018 Jun 1.

19. *"The role of nanotechnology in food safety."*
Colica C. Aiello V, **Boccuto L**, Kobyliak N, Strongoli MC, Vecchio I, Abenavoli L.
Minerva Biotecnologica 2018 June;30(2):69-73.

20. *"Adiponectin in hepatology."*
Abenavoli L, **Boccuto L**, Masarone M, Pellicano R, Persico M.
Minerva Biotecnologica 2018 March;30(1):36-40.

21. *"Role of genetics and metabolism in non-alcoholic fatty liver disease."*
Abenavoli L, Pellicano R, **Boccuto L**.
Panminerva Med. 2018 Feb 13. doi: 10.23736/S0031-0808.18.03418-3.

22. *"Identification of 22q13 genes most likely to contribute to Phelan McDermid syndrome".*
Mitz AR, Philyaw TJ, **Boccuto L**, Shcheglovitov A, Sarasua SM, Kaufman WE, Thurm A.
Eur J Hum Genet. 2018 Mar;26(3):293-302.

23. "*Publisher Correction: Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome."*
Li C, Brazill JM, Liu S, Bello C, Zhu Y, Morimoto M, Cascio L, Pauly R, Diaz-Perez Z, Malicdan MCV, Wang H, **Boccuto L**, Schwartz CE, Gahl WA, Boerkoel CF, Zhai RG.
Nat Commun. 2018 Jan 18;9(1):337.

24. *"Spermine Synthase Deficiency Causes Lysosomal Dysfunction and Oxidative Stress through Polyamine Oxidation in Snyder-Robinson Syndrome".*

Li C, Brazill JM, Liu S, Bello C, Zhu Y, Cascio L, Pauly R, Goheen M, Weech A, Malicdan MCV, Wang H, **Boccuto L**, Schwartz CE, Gahl WA, Boerkoel CF, Zhai RG.
Nat Commun. 2017 Nov 2;8(1):1257.

25. *"Polyphenols treatment in patients with nonalcoholic fatty liver disease."*
Abenavoli L, Milic N, Luzza F, **Boccuto L**, De Lorenzo A.
J Transl Int Med. Sep 30, 2017; 5(3): 144-147.

26. *"The impact of genetic polymorphisms on liver diseases: entering the era of personalized medicine."*
Boccuto L, Abenavoli L.
Eur J Gastroenterol Hepatol. 2017 Sep;29(9):1102-1103.

27. *"New serum markers for detection of early hepatocellular carcinoma."*
Abenavoli L, **Boccuto L**.
Panminerva Med. 2017 Jul 13.

28. *"Genetic and epigenetic profile of patients with alcoholic liver disease".*
Boccuto L, Abenavoli L.
Ann Hepatol. 2017 Aug 1;16(4):490-500.

29. *"Adiponectin serum level changes and its dynamic relationship with hepatitis C during viral clearance."*
Abenavoli L, **Boccuto L**.
Virulence. 2017 Apr 4:1-3. doi: 10.1080/21505594.2017.1315498. [Epub ahead of print]

30. "*Looking toward the Future: An Inquiry into the Acceptance of a Hypothetical Laboratory-based Screening Test for Autism Spectrum Disorder.*"
DeLuca JM, Sarasua SM, **Boccuto L**.
Journal of Pediatric Research, submitted for publication.

31. *"Concomitant 11p15.4-p15.5 duplication and terminal 22q13.33 deletion in a patient with features of Beckwith-Wiedemann syndrome."*
Peterson JF, Bick DP, Geddes GC, McCarrier J, Grignon JW Jr, Chirempes B, Broeckel U, Abidi F, Rogers RC, **Boccuto L**, DuPont B, vanTuinen P.
Am J Med Genet A. 2016 Dec;170(12):3348-3351.

32. *"PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution."*
Mirzaa G, Timms E, Conti V, Boyle EA, Girisha KM, Martin B, Kircher M, Olds C, Juusola J, Collins S, Park K, Carter M, Glass I, Krägeloh-Mann I, Chitayat D, Parikh AS, Bradshaw R, Torti E, Braddock S, Burke L, Ghedia S, M, Stewart F, Prasad C, Napier M, Saitta S, Straussberg R, Gabbett M, O'Connor BC, Keegan CE, Yin LJ, Meeng Lai AH, Martin N, McKinnonM, Addor M-C, **Boccuto L**, Schwartz CE, Lanoel A, Conway RL, Devriendt K,

Tatton-Brown K, Pierpont ME, Painter M, Lisa Worgan L, James Reggin J, Raoul Hennekam R, Tsuchiya K, Pritchard CC, Aracena M, Gripp KW, Cordisco M, Van Esch H, Garavelli L, Curry C, Goriely A, Kayserilli H, Shendure J, Graham J Jr., Guerrini R, and Dobyns WB.
JCI Insight. 2016;1(9):e87623.

33. *"Autism spectrum disorder in Phelan-McDermid syndrome: initial characterization and genotype-phenotype correlations."*
Oberman LM, **Boccuto L**, Cascio L, Sarasua S, Kaufmann WE.
Orphanet J Rare Dis. 2015 Aug 27;10(1):105.

34. *"Letter to the editor regarding Disciglio et al.: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome."*
Phelan K, **Boccuto L**, Rogers RC, Sarasua SM, McDermid HE.
Am J Med Genet A. 2015 Jul;167(7):1679-80.

35. *"Controlling false discoveries in high-dimensional situations: boosting with stability selection."*
Hofner B, **Boccuto L**, Göker M.
BMC Bioinformatics. 2015 May 6;16(1):144.

36. *"Clinical and genomic evaluation of 201 patients with Phelan-McDermid syndrome."*
Sarasua SM, **Boccuto L**, Sharp JL, Dwivedi A, Chen CF, Rollins JD, Rogers RC, Phelan K, Dupont BR.
Hum Genet. 2014 Jul;133(7):847-59. doi: 10.1007/s00439-014-1423-7. Epub 2014 Jan 31.

37. "22q13.2q13.32 genomic regions associated with severity of speech delay, developmental delay, and physical features in Phelan-McDermid syndrome."
Sarasua SM, Dwivedi A, **Boccuto L**, Chen CF, Sharp JL, Rollins JD, Collins JS, Rogers RC, Phelan K, DuPont BR.
Genet Med. 2014 Apr;16(4):318-28. Epub 2013 Oct 17.

38. "A Mutation in a Ganglioside Biosynthetic Enzyme, ST3GAL5, Results in Salt & Pepper Syndrome, a Neurocutaneous Disorder with Altered Glycolipid and Glycoprotein Glycosylation."
Boccuto L, Aoki K, Flanagan-Steet H, Chen C-F, Fan X, Bartel F, Petukh M, Pittman AR, Saul R, Chaubey A, Alexov E, Tiemeyer M, Steet R, Schwartz CE.
Hum Mol Genet. 2013 Sep 10.

39. "Decreased tryptophan metabolism in patients with autism spectrum disorders."
Boccuto L, Chen CF, Pittman AR, Skinner CD, McCartney HJ, Jones K, Bochner BR, Stevenson RE, Schwartz CE.
Mol Autism. 2013 Jun 3;4(1):16.

40. *"Prevalence of SHANK3 variants in patients with different subtypes of autism spectrum disorders."*
Boccuto L, Lauri M, Sarasua SM, Skinner CD, Buccella D, Dwivedi A, Orteschi D, Collins JS, Zollino M, Visconti P, Dupont B, Tiziano D, Schroer RJ, Neri G, Stevenson RE, Gurrieri F, Schwartz CE.
Eur J Hum Genet. 2013 Mar;21(3):310-6.

41. *"Program and abstracts for the 2011 Meeting of the Society for Glycobiology."*
Hollingsworth MT, Hart GW, Paulson JC, ... Aoki K, **Boccuto L**, Zhang Q, ... Ruhaak RL, Miyamoto S, Lebrilla CB.
Glycobiology 2011 Nov; 21(11):1454-531.

42. *"Association between deletion size and important phenotypes expands the genomic region of interest in Phelan-McDermid syndrome (22q13 deletion syndrome)."*
Sarasua SM, Dwivedi A, **Boccuto L**, Rollins JD, Chen CF, Rogers RC, Phelan K, DuPont BR, Collins JS.
J Med Genet. 2011 Nov;48(11):761-6.

43. *"A new syndrome with multiple capillary malformations, intractable seizures, and brain and limb anomalies."*
Carter MT, Geraghty MT, De La Cruz L, Reichard RR, **Boccuto L**, Schwartz CE, Clericuzio CL.
Am J Med Genet A. 2011 Feb;155(2):301-6.

44. *"Further evidence that the rs1858830 C variant in the promoter region of the MET gene is associated with autistic disorder."*
Jackson PB, **Boccuto L**, Skinner C, Collins JS, Neri G, Gurrieri F, Schwartz CE.
Autism Res. 2009 Aug;2(4):232-6.

45. *"Clinical experience in the evaluation of 30 patients with a prior diagnosis of FG syndrome."*
Lyons MJ, Graham JM Jr, Neri G, Hunter AG, Clark RD, Rogers RC, Moscarda M, **Boccuto L**, Simensen R, Dodd J, Robertson S, DuPont BR, Friez MJ, Schwartz CE, Stevenson RE.
J Med Genet. 2009 Jan;46(1):9-13.

46. *"Different mechanisms cause imprinting defects at the IGF2/H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumour."*
Cerrato F, Sparago A, Verde G, De Crescenzo A, Citro V, Cubellis MV, Rinaldi MM, **Boccuto L**, Neri G, Magnani C, D'Angelo P, Collini P, Perotti D, Sebastio G, Maher ER, Riccio A.
Hum Mol Genet. 2008 May 15;17(10):1427-35.

47. *"The Use of Microsatellite Instability, Immunohistochemistry and Other Variables in Determining the Clinical Significance of MLH1 and MSH2 Unclassified Variants in Lynch Syndrome."*
Lucci-Cordisco E, **Boccuto L**, G. Neri G, Genuardi M.

Cancer Biomark. 2006;2(1-2):11-27. Review.

48. *"Diagnostic strategies in overgrowth syndromes."*
Boccuto L, Lapunzina P, Gurrieri F, Neri G.
Ital J Pediatr 2006; 32: 81-100.

49. *"Two PMS2 Mutations in a Turcot Syndrome Family with Small Bowel Cancers"*
Agostini M, Tibiletti MG, Lucci-Cordisco E, Chiaravalli A, Morreau H, Furlan D, **Boccuto L**, Pucciarelli S, Capella C, Boiocchi M, Viel A.
Am J Gastroenterol 2005; 100: 1886-1891.

PLATFORM PRESENTATIONS AND POSTERS

1. *"Phelan-McDermid syndrome: history, achievements and news."*
L. Boccuto.
Oral presentation at the Phelan-McDermid Syndrome Australian Conference, Sydney, Australia, January 20-22, 2020.

2. *"PNPLA3 polymorphisms: from Phelan-McDermid syndrome to liver disease."*
L. Boccuto.
Oral presentation at the UMG Seminars in Molecular Medicine. University of magna Graecia, Catanzaro, Italy, December 18, 2019.

3. *"Genotype-phenotype correlation in Phelan-McDermid syndrome"*
L. Jain L, S. Srikanth, M. Wetsel, T. Fasolino, L. Oberman, R. Steet, **L. Boccuto**, S. Sarasua.
Oral presentation at the 37th annual meeting of the SOUTHEASTERN REGIONAL GENETICS GROUP (SERGG), Asheville, NC, July 18-20, 2019.

4. *"Germline PIK3CA Variants in Overgrowth Syndromes."*
J. Gass, **L. Boccuto**, R. Louie, S. Srikanth, R. Pauly, R. Stevenson.
Poster presentation at the 2019 ACMG Annual Clinical Genetics Meeting, Seattle, WA, April 2-6, 2019.

5. *"Personalized medicine in autism: new approaches to an old problem."*
L. Boccuto.
Oral presentation at the Human Genetics Symposium, Bob Jones University, Greenville, SC, USA, March 29, 2019.

6. *"Genetics and Environmental Aspects of Autism Spectrum Disorder: New Models and New Theories"*
L. Boccuto.
Oral presentation at the Converge Autism Summit, Greenville, SC, March 1-2, 2019.

7. *"PNPLA3 polymorphisms: from Phelan-McDermid syndrome to liver disease."*
   L. Boccuto.
   Oral presentation at the UMG Seminars in Molecular Medicine. University of magna Graecia, Catanzaro, Italy, December 18, 2018.

8. *"Metabolomic approaches to autism spectrum disorder (ASD)."*
   L. Boccuto.
   Seminar at the Southern Wesleyan University, Central, SC. October 23, 2018.

9. "*The pharmacogenetic effect of a SNP in the PNPLA3 gene in patients with Phelan-McDermid syndrome*."
   L. Abenavoli, L. Cascio, S. Srikanth, B. DuPont, A.R. Mitz, R.C. Rogers, K. Phelan, **L. Boccuto**.
   Poster presentation at the Phelan-McDermid Syndrome International Conference, Madrid, Spain, September 21-23, 2018.

10. "*The impact of the PNPLA3 gene in Phelan-McDermid syndrome: from liver function to pharmacogenetics*."
    L. Boccuto, L. Abenavoli, L. Cascio, S. Srikanth, B. DuPont, A.R. Mitz, R.C. Rogers, K. Phelan.
    Poster presentation at the Phelan-McDermid Syndrome Foundation 2018 International Family Conference and Phelan-McPosium, Grapevine, TX, USA, July 18-22, 2018.

11. "*Metabolic characterization of patients with Phelan-McDermid syndrome: the role of the PNPLA3 gene*."
    L. Boccuto.
    Oral presentation at the Annual Conference of the Italian Association for the Phelan-McDermid syndrome, Bologna, Italy, April 14, 2018.

12. "*Genetic and Environmental Aspects of Autism Spectrum Disorder (ASD): New Models and New Theories*."
    L. Boccuto.
    Oral presentation at the Human Genetics Symposium, Bob Jones University, Greenville, SC, USA, March 8, 2018.

13. "*Development of an Autism Spectrum Disorder Screening Test Based Upon Metabolic Profiling of Fresh Blood Samples*."
    Champaigne K, **L. Boccuto**.
    Oral presentation at the SCAND meeting, Greenwood, SC, USA, March 21, 2018.

14. *"The new frontiers of genomic medicine: the lesson of Phelan-McDermid syndrome and the PNPLA3 gene".*
    L. Boccuto

Oral presentation at the UMG Seminars in Molecular Medicine. University of magna Graecia, Catanzaro, Italy, December 18, 2017.

15. *"Metabolic investigation of segmental overgrowth: new insights in pathogenic mechanisms and treatments."*
L. Boccuto.
Biolog Webinar, October 26, 2017.

16. *"Novel treatment approaches for autism spectrum disorder: an in vitro model."*
L. Boccuto, L. Cascio, K. Champaigne, K. Jones, R. Pauly, S. Srikanth, C.F. Chen, S. Dunn, S. Sorrow, R. Cubillan, C. Skinner, CE Schwartz.
Poster presentation at the American Society of Human Genetics, Orlando, FL, USA, October 17-21, 2017.

17. *"Metabolic approaches for treatment of autism spectrum disorder."*
L. Boccuto, L. Cascio, K. Champaigne, K. Jones, R. Pauly, S. Srikanth, C.F. Chen, S. Dunn, S. Sorrow, R. Cubillan, C. Skinner, CE Schwartz.
Poster presentation at the SCAND Symposium, Columbia, SC, USA, October 9, 2017.

18. *"Clinical, molecular and biochemical aspects of Snyder-Robinson syndrome (spermine synthase deficiency)."*
C.E. Schwartz, L. Cascio, K. Jones, J. Norris, C. Skinner, C.-F. Chen, R.E. Stevenson, **L. Boccuto**.
Poster presentation at the Gordon Research Conference "Polyamine Metabolism in Disease and Polyamine-Targeted Therapies", Waterville Valley, NH, USA, June 25-30, 2017.

19. *"Metabolomic approaches to the study of neurodevelopmental disorders."*
L. Boccuto, L. Cascio, S. Srikanth, K. Jones, C.F. Chen, R. Pauly, C. Skinner, S. Sorrow, C. Schwartz.
Oral presentation at 3rd annual USC Neuroscience Community Retreat, Columbia, SC, USA, May 22, 2017.

20. *"Vascular anomalies associated with the PI3K-AKT pathway"*
L. Boccuto.
Oral presentation at 9th Annual Vascular Anomalies Symposium: New Insights from Research in Vascular Anomalies. Bon Secour St. Francis Hospital, Charleston, SC, USA, May 19, 2017.

21. *"Tryptophan and autism: a novel approach to an old problem."*
L. Boccuto.
Oral presentation at the Simons Foundation, New York, NY, USA, March 3, 2017.

22. *"Phelan-McDermid Syndrome: genotype-phenotype correlation and new perspectives on research and treatment."*
L. Boccuto.

Oral presentation at the Simons Foundation, New York, NY, USA, March 3, 2017.

23. *"Biomarkers in ASD: A SCAND collaborative project."*
L. Boccuto.
Oral presentation at the SC Autism and Neurodevelopmental Disorders Consortium (SCAND) meeting, Charleston, SC, USA, March 28, 2017.

24. *"Phelan-McDermid syndrome: molecular aspects and future perspectives in research and treatment."*
L. Boccuto.
Oral presentation at the Human Genetic Symposium "Genetic Syndromes: Between Diagnosis & Solutions!", Bob Jones University, Greenville, SC, USA, March 30, 2017.

25. *"Molecular characterization of activating mutations in Pi3K-AKT pathway genes: novel insights on pathogenesis and treatment."*
L. Boccuto, L. Cascio, K. Jones, C.F. Chen, C. Skinner, R. Pauly, R.E. Stevenson, C.E. Schwartz.
Poster at the 2017 Keystone Symposia Conference "PI3K Pathways in Immunology, Growth Disorders and Cancer", Santa Fe, NM, USA, January 19-23, 2017.

26. "*Tryptophan and autism: a novel approach to an old problem*."
L. Boccuto.
Lecture at the Clemson College of Behavioral, Social, and Health Science, Clemson, SC, USA, November 21, 2016.

27. "*Autism: Developing a new approach to a complex neurobehavioral disorder*."
L. Boccuto.
Platform presentation at the South Carolina Genetics Conclave, Greenwood, SC, USA, November 18, 2016.

28. "*Identifying Potentially Haploinsufficient Genes in 22q13 Deletion Syndrome*"
A. Mitz, T. Philyaw, **L. Boccuto**, A. Shcheglovitov, A. Thurm.
Poster at the Society for Neuroscience 2016 Meeting, San Diego, CA, USA, November 12-16, 2016.

29. "*Metabolic approach to autism spectrum disorder (ASD).*"
L. Boccuto.
Seminar at the David H. Murdock Research Institute, Kannapolis, NC, USA, November 3, 2016.

30. "*Investigation of aberrant tryptophan metabolism as a biochemical basis for Autism Spectrum Disorders (ASDs)*".
L. Boccuto, K.D. Champaigne, L. Cascio, D. Dean, C.F. Chen, C.E. Schwartz.

Poster at the Clemson University School of Health Research (CUSHR) Fall Meeting, Clemson, SC, USA, October 28, 2016.

31. "*Parents' Perspectives of Medical Screening for Autism: Would They Say "Yes" to the Test?*"
J. DeLuca, S. Sarasua, **L. Boccuto**.
International Society for Neonatal Screening (ISNS) 9th International Symposium, The Hague, The Netherlands, September 11–14, 2016.

32. "*Non-typical Gorlin syndrome: expanding the phenotype spectrum of PTCH1.*"
L. Boccuto, N. Di Donato, E. Blue, E.G.Bend, L. Cascio, S. Dunn, K. Jones, C. Skinner, G. Neri, D.A. Nickerson, M. Bamshad, C.E. Schwartz.
Poster at the 37th Annual David W. Smith Workshop on Malformations and Morphogenesis. Lake Arrowhead, CA, USA, September 9-13, 2016.

33. "*Metabolomic approaches to Autism Spectrum Disorder (ASD).*"
L. Boccuto.
Platform Presentation at the Research Committee Meeting of the "Els for Autism" Foundation, Jupiter, FL, USA, August 31, 2016.

34. "*Metabolomic characterization of neurobehavioral subgroups in Fragile x syndrome*."
L. Boccuto.
Platform presentation at "Advances and Innovations for Collaborative Research in Fragile X, Autism, Sex Chromosome Variations: Working Toward Patient-Centered Outcomes." Atlanta, GA, USA, August 11-13, 2016.

35. *"New approaches in the characterization of genotype/phenotype correlation in Phelan-McDermid syndrome."*
L. Boccuto, L. Cascio, L. Oberman, R. Dixon, K. Jones, J. Stallworth, S. Sarasua, S. Dunn, M. Darmer, C. Rogers, C. Skinner, K. Phelan, B. DuPont, C.E. Schwartz, W. Kaufmann.
Poster at the 2016 Phelan-McDermid Syndrome International Family Conference. Orlando, FL, USA, July 19-23, 2016.

36. *"Family perspectives of medical screening for autism."*
J. DeLuca, S. Sarasua, **L. Boccuto**.
Poster at the Southeastern Regional Genetics Group (SERGG) Annual Meeting. Ponte Vedra Beach, FL, USA, July 14-16, 2016.

37. *"Potential approaches to metabolic treatment in ASD."*
L. Boccuto.
Platform presentation at the South Carolina Consortium on Autism and Neurodevelopmental Disorders (SCCAND) Meeting. Columbia, SC, USA, April 22, 2016.

38. "*Generalized and segmental overgrowth.*"
G. Neri, **L. Boccuto**.

Platform presentation at the South Carolina Genetics Conclave. Greenwood, SC, USA, March 18, 2016.

39. "*Alcoholic Liver Disease (ALD): have a glass of good genes!*"
L. Boccuto.
Seminar at the Greenwood Genetic Center Statewide Case Conference. Greenwood, SC, USA, February 24, 2016.

40. *"Biolog phenotype metabolic microarrays: sorting out variable phenotypes associated with single genes."*
L. Boccuto, L. Cascio, K. Jones, C. Skinner, C.-F. Chen, C.E. Schwartz.
Poster at the 17th International Fragile X and other Early-Onset Cognitive Disorders Workshop, Strasbourg, France, September 27-30, 2015.

41. *"Phenotype microarray analysis may provide insight for potential therapeutic approaches in human disorders."*
C. Schwartz, L. Cascio, J. Norris, K. Jones, C.-F. Chen, **L. Boccuto**.
Platform presentation at the 17th International Fragile X and other Early-Onset Cognitive Disorders Workshop, Strasbourg, France, September 27-30, 2015.

42. *"Phenotype microarray: A novel methodology to access metabolic pathways in human disorders."*
C. Schwartz, C.-F. Chen, **L. Boccuto**.
Platform presentation at the 2nd International Symposium on Profiling (ISPROF 2015), Costa de Caparica, Portugal, September 21-24, 2015.

43. *"Novel approaches to the genotype/phenotype characterization in the Phelan-Mcdermid syndrome."*
L. Boccuto.
Seminar at Neuroscience Institute, National Council of Research, Milan, Italy, September 18, 2015.

44. *"Biolog phenotype metabolic microarrays: finding the link between genotype and phenotype"*
L. Boccuto, L. Cascio, K. Jones, C. Skinner, C.-F. Chen, C.E. Schwartz.
Platform presentation at the 3rd International Florence Conference on Phenotype MicroArray Analysis of Cells, Florence, Italy, September 10-12, 2015.

45. *"Application of state-of-the-art machine learning techniques to the PM data on autism-spectrum disorders – boosting with false discovery control."*
B. Hofner, **L. Boccuto**, M. Göker.
Platform presentation at the 3rd International Florence Conference on Phenotype MicroArray Analysis of Cells, Florence, Italy, September 10-12, 2015.

46. *"Functional studies of the AKT/PIK3CA/MTOR pathway and treatment approaches in conditions with segmental overgrowth."*
L. Boccuto.
Seminar at Catholic University of Sacred Heart, Rome, Italy, September 8, 2015.

47. *"Characterization of metabolic profiles in genetic conditions with intellectual disability and behavioral disorders."*
L. Boccuto.
Seminar at Catholic University of Sacred Heart, Rome, Italy, September 8, 2015.

48. *"Pi3K-AKT pathway: from isolated cancer to syndromes. Metabolic characterization and new therapeutic perspectives."*
L. Boccuto, L. Cascio, C.-F. Chen, X. Lei, K. Jones, C. Skinner, R. Stevenson, C.E. Schwartz.
Platform presentation at the 36th David W. Smith Annual Workshop on Malformations and Morphogenesis, St. Michaels, Maryland, USA, August 14-19, 2015.

49. *"Snyder-Robinson Syndrome: Molecular and Biochemical Aspect."*
C. Schwartz, **L. Boccuto**.
Platform presentation at the 2015 Snyder-Robinson syndrome Conference, Washington, DC, USA, July 16, 2015.

50. *"Functional studies and potential therapeutic approaches in disorders of the Pi3K-AKT pathway"*
L. Boccuto, L. Cascio, D. Dyment, K. Jones, J. Norris, C.-F. Chen, C. Skinner, L. Basel-Vanagaite, R. Stevenson, M. Innes, C. Schwartz.
Platform presentation at the 35th David W. Smith Annual Workshop on Malformations and Morphogenesis, Madison, Wisconsin, USA, July 25-30, 2014.

51. "*Metabolic profile of PMS cell lines"*
L. Boccuto
Platform presentation at the International Phelan-McDermid Syndrome Foundation Meeting, Orlando, Florida, USA, July 23-26, 2014.

52. *"Decreased tryptophan metabolism: the biochemical fingerprints of autism spectrum disorders"*
L. Boccuto, C.-F. Chen, A. Pittman, C. Skinner, H. McCartney, K. Jones, B. Bochner, R. Stevenson, C. Schwartz.
Poster at the American Society of Human Genetics (ASHG), Boston, Massachusetts, October 22-26, 2013.

53. *"Novel somatic mutations in Pi3K-AKT pathway genes in patients with segmental overgrowth and novel approaches for functional studies"*
L. Boccuto, M. DeGraff, J. Norris, K. Jones, L. Seaver, L. Cascio, C.-F. Chen, H. Dorman, C. Skinner, R. Saul, A. Hunter, W. Foulkes, K. Brockmann, S. Yang, R. Stevenson, C. Schwartz.

Platform presentation at the 34th David W. Smith Annual Workshop on Malformations and Morphogenesis, Mont-Tremblant, Quebec, Canada, August 9-14, 2013.

54. *"Phenotype Microarray: A Novel Methodology To Access Metabolic Pathways In Human Disorders"*
C. Schwartz, **L. Boccuto**, C.-F. Chen, K. Jones, A. Pittman, M. DeGraff, L. Cascio, C. Skinner.
Platform presentation at the 34th David W. Smith Annual Workshop on Malformations and Morphogenesis, Mont-Tremblant, Quebec, Canada, August 9-14, 2013.

55. *"The role of SHANK3 and other 22q13.33 genes in PMS patients without chromosomal rearrangements"*
L. Boccuto
Platform presentation at the Second International Phelan-McDermid Syndrome Symposium, Orlando, Florida, USA, July 25-26, 2012.

56. *"Severity of Speech Delay, Developmental Delay, and Physical features in Phelan-McDermid Syndrome are associated with 22q13.2q13.32 Genomic Regions"*
S.M. Sarasua, A. Dwivedi, **L. Boccuto**, C-F Chen, J.D. Rollins, R.C. Rogers,
M.C. Phelan, and B.R. DuPont.
Poster at the Second International Phelan-McDermid Syndrome Symposium, Orlando, Florida, USA, July 25-26, 2012.

57. *"22q13.2q13.32 Associated with Speech Delay, Developmental Delay, and Physical Features in Phelan-McDermid Syndrome"*
S.M. Sarasua, A. Dwivedi, **L. Boccuto**, C-F Chen, J.D. Rollins, R.C. Rogers, M.C. Phelan, and B.R. DuPont.
Poster at the Southeast Regional Newborn Screening and Genetics Collaborative (SERC) and 30th Annual Meeting of the Southeastern Regional Genetics Group (SERGG), Ponte Vedra Beach, Florida, USA, July 19-21, 2012.

58. *"A Missense Mutation in ST3GAL5 Results in a Severe Intellectual Disability Syndrome Associated with Altered Glycosphingolipid and O-Linked Glycan Expression"*
K. Aoki, **L. Boccuto**, Q. Zhang, H. Wang, F. Bartel, X. Fan, R. Saul, A. Chaubey, X. Yang, R. Steet, C. Schwartz, M. Tiemeyer.
Platform presentation at the Annual Conference of the Society for Glycobiology, Westin Seattle, Seattle, WA,USA, November 9-12, 2011.

59. *"A homozygous mutation in the ganglioside biosynthetic enzyme, ST3GAL5, results in a severe autosomal recessive neurocutaneous condition and altered glycosphingolipids and)-linked glycan expression"*
C. Schwartz, **L. Boccuto**, Q. Zhang, F. Bartel, K, Aoki, X. Fan, R. Saul, A. Chaubey, H. Wang, R. Steet, M. Tiemeyer, X. Yong.
Platform presentation at the 61st Meeting of the American Society of Human Genetics (ASHG), Montreal, Canada, October 11-15, 2011.

60. *"Metabolic abnormalities in patients with autism spectrum disorders"*
L. Boccuto, C.F. Chen, H. McCartney, C.D. Skinner, R.E. Stevenson, Charles E. Schwartz.
Poster at the "Autism 2010 Geneva Center for Autism International Symposium", Toronto, Canada, November 2-5, 2010.

61. *"Genotype-Phenotype Correlation Study in Phelan-McDermid Syndrome"*
S. Sarasua, J. Collins, A. Dwivedi, **L. Boccuto**, C. Rogers, C. Phelan, B. DuPont
Poster at the First International Phelan-McDermid Syndrome Symposium, New York, New York, USA, March 3-4, 2011.

62. *"The role of SHANK3 variants in patients with clinical manifestation of 22q13.3 deletion syndrome"*
L. Boccuto, M. Lauri, K. Phelan, M.E. Grimaldi, R.C. Rogers, D. Battaglia, C. D. Skinner, G. Neri, B. Dupont, F. Gurrieri, M. Zollino, C.E. Schwartz.
Poster at the 30th David W. Smith Annual Workshop on Malformations and Morphogenesis, Philadelphia, Pennsylvania, USA, August 5-9, 2009.

63. *"22q13.3 deletion syndrome is a multigenic disorder, with SHANK3 as the major pathogenic gene"*
M. Zollino, M.E. Grimaldi, **L. Boccuto**, C.E. Schwartz , D. Battaglia, E. Mercuri, F. Guzzetta, G. Marangi, D. Orteschi, D. Buccella, M. Lauri, P. Visconti, G. Gobbi, F. Guerrieri, and G. Neri.
Platform presentation at the 41st European Society of Human Genetics (ESHG), Wien, Austria, May 23-26, 2009.

64. *"22q13.3 deletion syndrome is a single gene disorder caused by haploinsufficiency of SHANK3"*
M. Zollino, M.E. Grimaldi, **L. Boccuto**, C.E. Schwartz , D. Battaglia, D. Lettori, C. Veredice, E. Mercuri, F. Guzzetta, D. Orteschi, D. Buccella, M. Lauri, F. Guerrieri, and G. Neri.
Platform presentation at the 19th Meeting of Dysmorphology, Strasbourg, France, September 4-5, 2008.

65. *"A database of unclassified variants in genes involved in hereditary colorectal cancer: a tool for genetic counselling and management of colorectal cancer families"*
M. Genuardi, E. Lucci Cordisco, R. Tricarico, **L. Boccuto**, E. Muscarella.
Poster at the 10th National Congress of the Italian Society of Human Genetics (SIGU), Montecatini Terme (PT), Italy, November 14-17, 2007.

66. *"Ruolo dei geni MYH ed MLH1 nella Sindrome di Muir-Torre (MTS)"*
G. Ponti, E. Lucci Cordisco, L. Losi, **L. Boccuto**, B. Roncari, S. Maffei, M.Pedroni, C. Di Gregorio, F.D. Tiziano, M. Genuardi, G. Rossi, P. Benatti, F. Domati, L. Roncucci, M. Ponz de Leon.

Platform presentation at the 4th National Congress of the Italian Association for the Study of the Familiarity and Inheritance of the Gastrointestinal Tumors (AIFEG) – Pavia, Italy, November 10-11, 2005.

67. *"Effetti differenziali di trattamenti demetilanti su linee cellulari tumorali"*
L. Boccuto, E. Lucci Cordisco, G. Neri, M. Genuardi.
Platform presentation at the 8th National Congress of the Italian Society of Human Genetics (SIGU) – Chia Laguna (CA), Italy, September 28-30, 2005.

68. *"Proposta di protocolli diagnostici nelle sindromi da eccesso di crescita"*
L. Boccuto, P. Lapunzina, F. Gurrieri, G. Neri.
Poster at the 8th National Congress of the Italian Society of Human Genetics (SIGU) – Chia Laguna (CA), Italy, September 28-30, 2005.

69. *"Mutazioni del gene MLH1 in pazienti HNPCC (Hereditary Non Polyposis Colorectal Cancer) con fenotipo Muir-Torre (MTS)"*
E. Lucci Cordisco, G. Ponti, **L. Boccuto**, B. Roncari, F.D. Tiziano, S. Maffei, M. Pedroni, L. Losi, C. Di Gregorio, G. Neri, M. Ponz De Leon, M. Genuardi.
Poster at the 8th National Congress of the Italian Society of Human Genetics (SIGU) – Chia Laguna (CA), Italy, September 28-30, 2005.

70. *"Differential activity of demethylating agents on the MLH1 promoter"*
E. Lucci Cordisco, **L. Boccuto**, I.Zito, G. Neri, M. Genuardi.
Poster at the 37th European Society of Human Genetics (ESHG), Prague, Czech Republic, May 7-10, 2005.

71. *"Analisi mutazionale dei geni BRCA1/BRCA2 in famiglie italiane ad alto rischio di tumore della mammella e dell'ovaio"*
F. D'Amico, E. Lucci Cordisco, A.L. Putignano, I.Zito, G. Chichierchia, **L. Boccuto**, L. Nardone, D. Terribile, P. Belli, G. Pastore, S. Greggi, G. Neri, M. Genuardi.
Poster at the 7th National Congress of the Italian Society of Human Genetics (SIGU), Pisa, Italy, October 13-15, 2004.

72. *"Genetica: la nuova dimensione della medicina"*
L. Boccuto.
Platform presentation at the Scientific Seminars of the Liceo Classico "P. Galluppi", Catanzaro, Italy, April 15-16, 2004.

73. *"Evalutation of MLH1 gene expression following reactivating treatments in colorectal cancer cell lines with hypermethylated MLH1 promoter"*
M. Genuardi, I. Zito, E. Lucci Cordisco, **L. Boccuto**, G. Chichierchia, G. Neri.
Poster at the 53rd Annual Meeting of American Society of Human Genetics (ASHG), Los Angeles, California, USA, November 4-8, 2003.

74. *"Studio dell'espressione del gene MLH1 in seguito a trattamenti con agenti demetilanti e iperacetilanti in linee cellulari con promotore di MLH1 ipermetilato"*
E. Lucci Cordisco, I.Zito, **L. Boccuto**, G. Chichierchia, G. Neri, M. Genuardi.
Poster at the 6th National Congress of the Italian Society of Human Genetics (SIGU) - Verona, Italy, September 24-27, 2003.

75. *"MSH6 mutations in familial colorectal cancer: correlations with clinical and immunohistochemical data"*
M. Genuardi, E. Lucci Cordisco, **L. Boccuto**, E. Dalla Longa, E. Passerini, P. Sala, L. Bertario, P. Benatti, C. Di Gregorio, M.A. Caligo, M.G. Tibiletti, M. Ponz De Leon, P. Radice, G. Neri.
Poster at the 4th Joint Meeting of the Leeds Castle Polyposis Group and the International Collaborative Group on Hereditary NonPolyposis Colorectal Cancer, Cleveland, Ohio, USA, September 4-6, 2003.

76. *"Nuove frontiere della genetica"*
L. Boccuto.
Platform presentation at the Progetto "Gutenberg e oltre", Catanzaro, Italy, November 26-30, 2002.

77. *"Mutazioni del gene MSH6 in carcinomi colorettali familiari: correlazioni con dati clinici, molecolari e immunoistochimici"*
E. Lucci Cordisco, **L. Boccuto**, E. Dalla Longa, E. Passerini, P. Sala, L. Bertario, P. Benatti, C. Di Gregorio, M.G. Tibiletti, M.A. Caligo, M. Ponz De Leon, P. Radice, G. Neri, M. Genuardi.
Platform presentation at the 1st National Congress of the Italian Association for the Study of the Familiarity and Inheritance of the Gastrointestinal Tumors (AIFEG), Firenze, Italy, November 15-16, 2002.

78. *"Analisi immunoistochimica e mutazionale del gene MSH6 in carcinomi colorettali (CCR) familiari"*
E. Lucci Cordisco, **L. Boccuto**, E. Dalla Longa, E. Passerini, L. Bertario, P. Benatti, C. Di Gregorio, M.G. Tibiletti, M.A. Caligo, M. Ponz De Leon, P. Radice, G. Neri, M. Genuardi.
Platform presentation at the 5th National Congress of the Italian Society of Human Genetics (SIGU), Verona, Italy, September 24-27, 2002.