CURRICULUM VITAE

- Boccuto Luigi, nato a Catanzaro, il 23/08/1978, residente a Roma.

- Ha conseguito il diploma di maturità classica nel 1996, presso il liceo "P.Galluppi" di Catanzaro, con votazione di 60/60simi e lettera di lode.

- Ha conseguito la laurea in Medicina e Chirurgia nel 2002, presso l'Università Cattolica del Sacro Cuore di Roma, con votazione di 110/110 e lode.

- Frequenta dal 1999 l'Istituto di Genetica Medica dell'Università Cattolica del Sacro Cuore, in qualità di studente interno (dal 1999 al 2002) ed in seguito di medico frequentante (dal 2002 al 2003).

- Nel Marzo del 2003 ha vinto il concorso per la Scuola di Specializzazione in Genetica Medica, classificandosi al primo posto della graduatoria con il punteggio di 90/100.

- Nel Giugno 2003 ha conseguito il First Certificate in English presso la Cambridge University (Place of entry: Roma).

- In data 11 Ottobre 2003 ha vinto a Perugia il premio di laurea "Gianmichele Laccetti", riservato ai laureati in Medicina e Chirurgia, Biologia, Farmacia, Chimica e Tecnologie Farmaceutiche di tutte le Università Italiane degli anni 2002/2003 e 2003/2004 con una Tesi di Laurea in campo Oncologico, sia sperimentale che clinico.

- Da Gennaio 2007 a Giugno 2010 ha lavorato come research scholar presso il Greenwood Genetic Center, Greenwood, USA.

- Da Luglio 2010 al Dicembre 2013 lavora come Staff Scientist presso il Greenwood Genetic Center, Greenwood, USA.

- Da Gennaio 2014 a oggi lavora come Assistant Research Scientist presso il Greenwood Genetic Center, Greenwood, USA.

- Da Aprile 2016 a oggi lavora come Clinical Assistant Professor presso la Clemson University School of Health Research, Clemson, SC.

- Da Settembre 2017 a Ottobre 2019 ha lavorato come Chief Scientific Officer presso STALICLA, SA, Ginevra, Svizzera.

- Dal Marzo 2018 a oggi lavora come Research Assistant Professor of Pediatrics nel Department of Pediatrics presso la University of South Carolina School of Medicine, Columbia, SC.

- Co-autore dei seguenti capitoli:
    - **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/

- Editore regionale per il Nord America per il "Reviews on Recent Clinical Trials" da Aprile 2017.

- Membro del comitato scientifico della 5ª edizione italiana del Merck Manual of Diagnosis and Therapy (tradotta dalla 18ª edizione inglese) pubblicato da Raffaello Cortina Editore Srl, Milano, 2008. ISBN: 9788847007079.

- Membro del comitato scientifico della 6ª edizione italiana del Merck Manual of Diagnosis and Therapy (tradotta dalla 19ª edizione inglese) pubblicato da Raffaello Cortina Editore Srl, Milano, 2016. ISBN: 9780911910193.

- Revisore dal 2010 per:
    - 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.

- Membro dell'American Society of Human Genetics dal 2013.

- Revisore esterno per la Fondazione Telethon dal 2017.

- Esperienza nella genetica dei tumori ereditari, in particolare del cancro colorettale e del seno; ha effettuato indagini molecolari sui geni *DAPK* e *MSH6* e studi sulla metilazione del promotore del gene *MLH1*. Esperienza nello screening di geni candidati per autismo e ritardo mentale (*SHANK3, EGR2, SLC9A6, ARC, RPL10, GJB6, CRYL1, SLC3A2, SLC7A5, SLC7A8, ST3GAL5, CATSPER2, QPRT, IB2, ACR, RABL2B, SMS, SLC6A8, SULT4A1, ARHGAP8, ALG12*), condizioni caratterizzate da eccesso di crescita e malformazioni (*AKT1*, *AKT2*, *AKT3*, *PIK3CA, PIK3R2*, *PDPK1*, *PIK3CB, PIK3CD, PIK3R1, PIK3R3, PIK3R5*), e indagini metaboliche sul linee cellulari di pazienti affetti da autismo, ritardo mentale ed eccesso di crescita. Ottima conoscenza di tecniche di genetica molecolare, culture cellulari, array genetici e metabolici.

- Esperienza in genetica clinica, in particolare in condizioni caratterizzate da ritardo mentale, autismo, eccesso di crescita.

- Esperienza didattica come tutor di studenti universitari per il Summer Intern Program del Greenwood Genetic Center dal 2009, period durante il quale ha svolto la funzione di supervisore per 10 interni ufficiali e 13 volontari.

- Insegnante e speaker per il Greenwood Genetic Center Teacher Course dal 2014 e per il Greenwood Genetic Center Summer High School Camp dal 2018.

- Membro del comitato di valutazione per la tesi di dottorato (Ph.D.) per due candidati presso l'Healthcare Genetics Program of Clemson University.

PREMI E RICONOSCIMENTI

1. 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."

2. 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".

3. 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"*.

LIBRI E CAPITOLI

- 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.

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- 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.

BREVETTI E GRANT

- Co-inventore del brevetto "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, data 20 Ottobre 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.

PUBBLICAZIONI – h-index: 19 – i10-index: 25 (citazioni totali: 1218)

1. *"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.

2. *"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.

3. *"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.

4. *"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.

5. *"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.

6. *"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.

7. *"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.

8. *"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.

9. *"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.

10. *"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.

11. *"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.

12. *"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.

13. *"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.

14. *"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/

15. *"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.

16. *"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.

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

18. *"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.

19. *"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.

20. *"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.
Nature Communications, accepted for publication.

21. *"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.

22. *"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.

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

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

25. *"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.

26. "*A Pilot Study of Perspectives of Laboratory Screening for Autism.*"
DeLuca JM, Sarasua SM, **Boccuto L**.
IJNS, submitted for publication.

27. "*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.

28. "*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. Doi: 10.1172/jci.insight.87623.

29. "*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.

30. "*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.

31. "*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.

32. "*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.

33. *"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. doi: 10.1038/gim.2013.144. Epub 2013 Oct 17.

34. *"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. 2014 Jan 15;23(2):418-33. doi: 10.1093/hmg/ddt434.

35. *"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.

36. *"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.

37. *"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.

38. *"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.

39. *"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.

40. *"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.

41. *"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.

42. *"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.

43. *"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**, Neri G, Genuardi M.
Cancer Biomark. 2006;2(1-2):11-27. Review. Doi: 10.3233/CBM-2006-21-203

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

45. *"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.

PRESENTAZIONI & POSTER

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

15. *"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.

16. *"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.

17. *"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 at the Gordon Research Conference "Polyamine Metabolism in Disease and Polyamine-Targeted Therapies", Waterville Valley, NH, USA, June 25-30, 2017.

18. *"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.

19. *"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.

20. *"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.

21. *"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.

22. *"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.

23. *"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.

24. *"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.

25. "*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.

26. "*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.

27. "*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.

28. "*Metabolic approach to autism spectrum disorder (ASD).*"
L. Boccuto.

Seminar at the David H. Murdock Research Institute. Kannapolis, NC, USA, November 3, 2016.

29. "*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.

30. "*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.

31. "*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.

32. "*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.

33. "*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.

34. *"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.

35. *"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.

36. *"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.

37. "*Generalized and segmental overgrowth.*"
G. Neri, **L. Boccuto**.
Platform presentation at the South Carolina Genetics Conclave, Greenwood, SC, USA, March 18, 2016.

38. "*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.

39. *"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.

40. *"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.

41. *"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.

42. *"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.

43. *"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 3nd International Florence Conference on Phenotype MicroArray
Analysis of Cells, Florence, Italy, September 10-12, 2015.

44. *"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 3nd International Florence Conference on Phenotype MicroArray Analysis of Cells, Florence, Italy, September 10-12, 2015.

45. *"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.

46. *"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.

47. *"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.

48. *"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.

49. *"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.

50. "*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.

51. *"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.

52. *"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.

53. *"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.

54. *"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.

55. *"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.

56. *"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

57. *"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, November 9-12, 2011.

58. *"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.

59. *"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.

60. *"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.

61. *"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.

62. *"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.

63. *"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.

64. *"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.

65. *"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.

66. *"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.

67. *"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.

68. *"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.

69. *"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.

70. *"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.

71. *"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.

72. *"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.

73. *"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.

74. *"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.

75. *"Nuove frontiere della genetica"*

L. Boccuto.

Platform presentation at the Progetto "Gutenberg e oltre", Catanzaro, Italy, November 26-30, 2002.

76. *"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.

77. *"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.