

**FORMATO EUROPEO
PER IL CURRICULUM
VITAE**



Informazioni personali

Nome e Cognome	Sandro Banfi
Telefono	081 5667585
E-mail	sandro.banfi@unicampania.it
Cittadinanza	Italiana
Data di nascita	3 giugno 1964
Sesso	M
Codice Fiscale	BNFSDR64H03F839D

Esperienza professionale

POSIZIONE ATTUALE	Professore Ordinario, Genetica Medica Università degli Studi della Campania “Luigi Vanvitelli”. Dirigente medico UOSD Genetica Medica. Università degli Studi della Campania “Luigi Vanvitelli” AZIENDA OSPEDALIERA UNIVERSITARIA DAI di SANITÀ PUBBLICA e SERVIZI di LABORATORIO
1991-1994	Post-doc Department of Pediatrics, Baylor College of Medicine, Houston
Dal 1995	Ricercatore Telethon Institute for Genetics and Medicine (T.I.G.E.M.)
Dal 2005	Coordinatore del Programma di Dottorato di Ricerca Internazionale in Genetica umana Open University
Dal 2014	Docente del Corso di Dottorato in Scienze Biomolecolari Università degli Studi della Campania “Luigi Vanvitelli”
Dal 2004	Docente Scuola Superiore Europea di Medicina Molecolare (SEMM)
2011-2012	Ricercatore Universitario, Genetica Medica Seconda Università degli Studi di Napoli.

2012-2018	Professore associato, Genetica Medica						
Dal 2018	Dirigente medico UOSD Genetica Medica. Università degli Studi della Campania “Luigi Vanvitelli” AZIENDA OSPEDALIERA UNIVERSITARIA DAI di SANITÀ PUBBLICA e SERVIZI di LABORATORIO						
Istruzione e formazione							
1993	Specializzazione in Neurologia (<i>summa cum laude</i>) Clinica Neurologica, Università di Napoli						
1989	Abilitazione professionale Il Facoltà di Medicina e Chirurgia Università di Napoli						
1989	Laurea in Medicina (<i>summa cum laude</i> e menzione alla carriera) Il Facoltà di Medicina e Chirurgia Università di Napoli						
Capacità e competenze personali							
Madrelingua(e)	ITALIANO						
Altra(e) lingua(e)	INGLESE						
Autovalutazione							
Livello europeo (*)							
INGLESE							
		Comprensione		Parlato		Scritto	
		Ascolto	Lettura	Interazione orale	Produzione orale		
C1	Livello avanzato	C1	Livello avanzato	C1	Livello avanzato	C1	Livello avanzato

(*) [Quadro comune europeo di riferimento per le lingue](#)

ATTIVITA' SCIENTIFICA

Attività di revisore per riviste internazionali

Attività svolta per le seguenti riviste scientifiche:
 Journal of Medical Genetics,
 Clinical Genetics,
 European Journal of Human Genetics,
 Experimental Eye Research,
 Genesis,
 Genome Biology,
 Genome Research,
 Genomics,
 Gene,
 Human Genetics,
 Human Molecular Genetics,
 Human Mutation,
 Trends in Biochemical Sciences,
 Investigative Ophthalmology & Visual Science (IOVS),
 Brain,
 British Journal of Ophthalmology,
 BMC Bioinformatics,
 BMC Genomics,
 BMC Molecular Biology,
 BMC Developmental Biology,

BMC Medical Genetics,
Molecular Therapy,
Molecular Vision,
PLoS ONE,
PLoS Genetics,
American Journal of Medical Genetics,
RNA Biology,
Scientific Reports,
Pharmacological Research,
International Journal of Biochemistry and Cell Biology

Attività di revisore per Enti di supporto della ricerca

Attività svolta per le seguenti agenzie di finanziamento:
Ministero Universita' e Ricerca,
The Israel Science Foundation,
EMBO,
The Wellcome Trust,
French National Research Program on Vision,
Fight for Sight-British Eye Research Foundation
MRC,
Swiss National Science Foundation,
Fondation Voir et Entendre,
Health Research Board,
CNRS,
INSERM,
The Netherlands Organisation for Health Research and Development (ZonMw),
Special Trustees of Moorfields Eye Hospital, United Kingdom,
National Science Centre,
Poland,
Academy of Finland,
Sparks Charity,
United Kingdom,
Fondazione Mariani

Affiliazioni a societa' scientifiche

American Society of Human Genetics (ASHG)
Association for Research in Vision and Ophthalmology (ARVO)
European Society of Human Genetics (ESHG)
Societa' Italiana di Genetica Umana (SIGU)
Bioinformatic Italian Society (BITS)

Membro di comitati organizzativi (nazionali ed internazionali)

Co-organizzatore del Convegno "The Biology and Development of the Eye in Health and Disease", 170 Convegno IGB, Capri, Capri, 9-12 Ottobre, 2004.
Co-organizzatore del Workshop "Next Generation Sequencing in sensory disorders" 11 Febbraio 2011, Leuven, Belgio.
Co-organizzatore del Workshop "Next Generation Sequencing – Application cases and bioinformatics development", German-Italian Dialogue 2012, 17-19 July, Naples, Italy.

Membro di consigli scientifici

Dal 2006, Membro del consiglio scientifico di Retina Italia Onlus.
Nel 2017, Membro del Scientific Advisory Board, Institut de la Vision, Paris, France
Dal 2007, membro dell'European Retinal Disease Consortium (ERDC, <http://www.erdc.info/>), consorzio composto da 18 gruppi internazionali per la definizione delle basi molecolari delle distrofie retiniche ereditarie.

Premi Scientifici e onorificenze

2009, "Board of Director Awards" dalla Fondazione statunitense FFB (Foundation Fighting Blindness).
2017, Visionary of the Quarter, European Visionary of the Quarter, European Vision Institute
1999. Vincitore della borsa di studio associata alla Scuola di Specializzazione in Neurologia, Universita' degli Studi di Napoli.

Brevetti

Inventore del brevetto dal titolo "mir-204 and mir-211 and uses thereof", depositato come PCT l'11Marzo

2014, pubblicazione WO201414005. Concesso in Europa, Cina, Stati Uniti.
Inventore della domanda di brevetto dal titolo "mir-181 inhibitors and uses thereof", depositata il 20 Aprile 2018.

Fondi di ricerca con peer review

Grant Nazionali (selezionati)

1. Systematic identification and characterization of novel candidate genes for eye diseases." Finanziato dalla Fondazione Telethon (2000-2003)
2. "Study of the molecular basis of developmental eye defects - microphthalmia, anophthalmia and coloboma (MAC)." Finanziato dalla Fondazione Telethon (2003-2006).
3. "Sviluppo di una procedura basata sul "next generation sequencing" per l'identificazione delle cause molecolari delle retinitis pigmentose autosomiche recessive". Finanziato da Retina Italia onlus (2010-2011).
4. "Genetic bases of birth defects" Finanziato dal Ministero Italiano della Salute (2008-2010).
5. "microRNA-regulated gene networks in the retina." Finanziato dalla Fondazione Telethon (2011-2016)
6. "Retinitis Pigmentosa: an integrated application of novel strategies towards diagnosis and treatment". Finanziato da Fondazione Roma (2015-2019).
7. "Systematic search for microRNAs that play a role in photoreceptor degeneration". Finanziato dalla Fondazione Telethon (2017-2018)
8. "Toward new methods for early diagnosis and screening of genetic ocular diseases in childhood". Finanziato dal Ministero italiano della Istruzione, Universita' e Ricerca (Progetto PRIN) (2017-2020).

Grant Internazionali (selezionati)

1. "EUROPEAN RETINAL RESEARCH TRAINING NETWORK (RETNET)". Finanziato dalla Unione Europea (2004-2007).
2. "Functional genomics of the retina in health and disease (Evi-Genoret)". Finanziato dalla Unione Europea (2005-2009).
3. "Novel tool for high-throughput characterization of genomic elements regulating gene expression in chordates. (TRANSCODE)" Finanziato dalla Unione Europea (2005-2008).
4. Technological innovation of high throughput molecular diagnostics of clinically and molecularly heterogeneous genetic disorders. (Techene)". Finanziato dalla Unione Europea (2009-2012).
5. "EyeTN - Beyond the Genome; training the next generation of ophthalmic researchers." Finanziato dalla Unione Europea (2012-2016).
6. "MicroRNA miR-204, a new potential therapeutic tool for inherited retinal dystrophies". Finanziato dalla Fondazione statunitense Foundation Fighting Blindness (FFB) (2016-2019).
7. "StarT- European Training Network to Diagnose, Understand and Treat Stargardt Disease, a Frequent Inherited Blinding Disorder". Finanziato dalla Unione Europea (2018-2022)

Pubblicazioni scientifiche

Autore di oltre 130 pubblicazioni scientifiche internazionali peer-reviewed che presentano nel loro complesso i seguenti valori bibliometrici:

- Impact Factor totale = 862.8
- Impact Factor medio = 8.6
- Numero totale di citazioni = 11936 (Google Scholar); 8998 (Scopus); 8530 (WoS)
- Numero medio di citazioni/articolo = 81.74 (Google Scholar); 74.36 (Scopus); 74.17 (WoS)
- H-Index complessivo = 53 (Google Scholar); 47 (Scopus)

ELENCO DELLE PUBBLICAZIONI SCIENTIFICHE

1. S. Banfi and H.Y. Zoghbi. Molecular genetics of hereditary ataxias. In Bailliere's Clinical Neurology, eds TH Brandt et al: Bailliere Tindall, London, volume 3, number 2, pp. 281-95.
2. Filla A, De Michele G, Marconi R, Santorelli F, Trombetta L, Banfi S, Campanella G:Effects of thyrotropin-releasing hormone on heart rate in inherited ataxias. Med Sci Res 17: 569-570, 1989.
3. Cavalcanti F, Cocozza S, Filla A, De Michele G, Pianese L, Porcellini A, Monticelli A, Pandolfo M, Banfi S, Varrone S et al: Friedreich's disease. A linkage study in southern and central Italy. Acta Neurol 14: 519-523, 1992
4. Banfi S., Ledbetter SA, Chinault AC, Zoghbi HY: An easy and rapid method for the detection of chimeric yeast artificial chromosome clones. Nucleic Acids Res 20: 1814, 1992.
5. Filla A, De Michele G, Orefice G, Santorelli F, Trombetta L, Banfi S, Squitieri F, Napolitano G, Puma D, Campanella G: A double-blind cross-over trial of amantadine hydrochloride in Friedreich's ataxia. Can J Neurol Sci

20: 52-55, 1993.

6. Filla A, De Michele G, Santorelli F, Banfi S, Campanella G, Marconi R, Rossi F, Cavalcanti F: Epidemiological survey of hereditary ataxias and spastic paraplegias in Molise, Italy. In *Handbook of cerebellar diseases*, R. Lechtenberg ed., 407-413, Marcel Dekker, New York, 1993.
7. Orr HT, Chung M-Y, Banfi S, Kwiatkowski TJ, Jr., Servadio A, Beaudet AL, McCall AE, Duvick LA, Ranum LPW, Zoghbi HY: Expansion of an unstable trinucleotide (CAG) repeat in Spinocerebellar ataxia type 1. *Nature Genetics* 4:221, 1993.
8. Kwiatkowski TJ, Orr HT, Banfi S, McCall AE, Jodice C, Persichetti F, Novelletto A, LeBorgne-Demarquoy F, Duvick LA, Frontali M, et al: The gene for autosomal dominant spinocerebellar ataxia (SCA1) maps centromeric to D6S89 and shows no recombination, in nine large kindreds, with a dinucleotide repeat at the AM10 locus. *Am J Hum Genet* 53: 391-400, 1993.
9. Banfi S, Chung M-Y, Kwiatkowski TJ, Jr., Ranum LPW, McCall AE, Chinault AC, Orr HT, Zoghbi HY: Mapping and cloning of the critical region for the spinocerebellar ataxia type 1 gene in a yeast artificial chromosome contig spanning 1.2Mb. *Genomics* 18:627, 1993.
10. Banfi S, Servadio A, Chung M-Y, Kwiatkowski TJ, Jr., McCall AE, Duvick LA, Shen Y, Roth EJ, Orr HT, Zoghbi HY: Identification and characterization of the gene causing type 1 spinocerebellar ataxia. *Nature Genetics* 7:513, 1994.
11. Ranum LPW, Chung M-Y, Banfi S, Bryer A, Schut LJ, Ramesar R, Duvick LA, McCall AE, Subramony SH, Goldfarb L, Gomez C, Sandkuijl LA, Orr HT, Zoghbi HY: Molecular and clinical correlations in spinocerebellar ataxia type 1 (SCA1): Evidence for familial effects on the age of onset. *Am. J. Hum. Genet.* 55: 244-252, 1994.
12. Filla A, De Michele G, Banfi S, Santoro L, Perretti A, Cavalcanti F, Pianese L, Castaldo I, Barbieri F, Campanella G, and Cocozza S: Has spinocerebellar ataxia type 2 a distinct phenotype? Genetic and clinical study of an Italian family. *Neurology*, 45: 793-796, 1995.
13. Banfi S and Zoghbi HY: Detection of chimerism in YAC clones. In *YAC protocols* ed D. Markie Humana Press, Totowa, New Jersey, volume 54, 115-121, 1995.
14. Banfi S, Servadio A, Chung M-y, Capozzoli F, Duvick L, Elde R, Zoghbi HY, Orr HT: Cloning and developmental expression analysis of the murine homolog of the spinocerebellar ataxia type 1 gene (Sca1). *Human Molecular Genetics* 5:33, 1996.
15. Banfi S*, Borsani G*, Rossi E, Bernard L, Guffanti A, Rubboli F, Marchitiello A, Giglio S, Coluccia E, Zollo M, Zuffardi O, Ballabio A: Identification and mapping of human cDNAs homologous to Drosophila mutant genes through EST database searching. *Nature Genetics* 13:167-74, 1996. *The first two authors contributed equally to the work.
16. Guffanti A, Banfi S, Simon G, Ballabio A, Borsani G: DRES search engine: of flies, men and ESTs. *TIG*, 13(2): 79-80, 1997.
17. Rubboli F, Bulfone A, Bogni S, Marchitiello A, Zollo M, Borsani G, Ballabio A, Banfi S: A mammalian homolog of the Drosophila retinal degeneration B gene: implications for the evolution of phototransduction mechanisms. *Genes Funct.* 1997 Jun;1(3):205-13.
18. Banfi S, Borsani G, Bulfone A, Ballabio A: Drosophila-related expressed sequences. *Hum Mol Genet* 1997;6(10):1745-53.
19. Jackson FR, Banfi S, Guffanti A, Rossi E: A novel zinc finger-containing RNA-binding protein conserved from fruitflies to humans. *Genomics* 1997 May 1; 41(3):444-52.
20. Banfi S, Guffanti A, Borsani G: How to get the best of dbEST. *Trends Genet* 1998 Feb;14(2): 80-1.
21. Bione S, Sala C, Manzini C, Arrigo G, Zuffardi O, Banfi S, Borsani G, Jonveaux P, Philippe C, Zuccotti M, Ballabio A, Toniolo D: A human homologue of the Drosophila melanogaster diaphanous gene is disrupted in a patient with premature ovarian failure: evidence for conserved function in oogenesis and implications for human sterility. *Am J Hum Genet* 1998 Mar; 62(3):533-41.
22. Matilla A, Roberson ED, Banfi S, Morales J, Armstrong DL, Burright EN, Orr HT, Sweatt JD, Zoghbi HY, Matzuk MM: Mice lacking ataxin-1 display learning deficits and decreased hippocampal paired-pulse facilitation. *J Neurosci* 1998 Jul 15;18(14):5508-16.
23. de Conciliis L, Marchitiello A, Wapenaar MC, Borsani G, Giglio S, Mariani M, Consalez GG, Zuffardi O, Franco B, Ballabio A, Banfi S: Characterization of Cxorf5 (71-7A), a novel human cDNA mapping to Xp22 and encoding a protein containing coiled-coil alpha-helical domains. *Genomics* 1998 Jul 15;51(2):243-50.

24. Borsani G, Ballabio A, Banfi S: A practical guide to orient yourself in the labyrinth of genome databases. *Hum Mol Genet* 1998;7(10):1641-8.
25. Bulfone A, Gattuso C, Marchitello A, Pardini C, Boncinelli E, Borsani G, Banfi S, Ballabio A: The embryonic expression pattern of 40 murine cDNAs homologous to *Drosophila* mutant genes (Dres): a comparative and topographic approach to predict gene function. *Hum Mol Genet*, 1998 Dec;7(13):1997-2006.
26. Borsani G, De Grandi G, Ballabio A, Bulfone A, Bernard L, Banfi S, Gattuso C, Mariani M, Dixon M, Donnai D, Metcalfe K, Winter R, Robertson M, Axton R, Brown A, van Heyningen V and Hanson I: EYA4, a novel vertebrate gene related to *Drosophila* eyes absent. *Hum Mol Genet*, 1999; 8(1):11-23
27. Volta M, Bulfone A, Gattuso C, Rossi E, Mariani M, Consales GG, Zuffardi O, Ballabio A, Banfi S, Franco B: Identification and characterization of CDS2, a mammalian homolog of the *Drosophila* CDP-diacylglycerol synthase gene. *Genomics*, 1999 Jan 1;55(1):68-77.
28. Volorio S, Simon G, Repetto M, Banfi S, Borsani G, Ballabio A, Zollo M: Sequencing analysis of forty-eight human IMAGE cDNA clones similar to *Drosophila* mutant proteins. *DNA Sequence*, 1998 9(5-6) : 307-315.
29. Banfi S, Bassi MT, Andolfi G, Marchitello A, Zanotta S, Ballabio A, Casari G, Franco B. Identification and characterization of AFG3L2, a novel paraplegin-related gene. *Genomics*, 1999 59(1): 51-58.
30. Barbieri AM, Lupo G, Bulfone A, Andreazzoli M, Mariani M, Fougerousse F, Consalez GG, Borsani G, Beckmann JS, Barsacchi G, Ballabio A, Banfi S. A novel homeobox gene, vax2, controls the patterning of the eye dorso-ventral axis. *Proceedings of National Academy of Sciences USA* 96(19): 10729-10734, 1999.
31. Piccini M, Vitelli F, Seri M, Galietta LJV, Moran O, Bulfone A, Banfi S, Pober B, Renieri A. KCNE1 like gene is deleted in AMME contiguous gene syndrome: identification and characterization of the human and mouse homologs. *Genomics* , 1999 60(3) 251-257.
32. Bulfone A, Menguzzato E, Broccoli V, Marchitello A, Gattuso C, Mariani M, Consalez GG, Martinez S, Ballabio A, Banfi S. Barhl1, a gene belonging to a new subfamily of mammalian homeobox genes, is expressed in migrating neurons of the CNS. *Human Molecular Genetics*, 2000; 9(9):1443-1452.
33. Coppola M, Pizzigoni A, Banfi S, Bassi MT, Casari G, Incerti B. Identification and Characterization of YME1L1, a Novel Paraplegin-Related Gene. *Genomics*, 2000; 66(1): 48-54.
34. Buanne P, Corrente G, Micheli L, Palena A, Lavia P, Spadafora C, Krishnappa Lakshmana M, Rinaldi A, Banfi S, Quarto M, Bulfone A, Tirone F. Cloning of PC3B, a Novel Member of the PC3/BTG/TOB Family of Growth Inhibitory Genes, Highly Expressed in the Olfactory Epithelium. *Genomics*, 2000; 68(3): 253-263
35. Liu Y, Lupo G, Marchitello A, Gestri G, He RQ, Banfi S, Barsacchi G, Expression of the Xvax2 gene demarcates presumptive ventral telencephalon and specific visual structures in *Xenopus laevis*. *Mechanisms of Development*, 2001; 100(1): 115-118
36. Bermingham NA, Hassan BA, Wang VY, Fernandez M, Banfi S, Bellen HJ, Fritzsch B, Zoghbi HY. Proprioceptor pathway development is dependent on MATH1. *Neuron*, 2001; 30:411-422.
37. Barbieri AM, Broccoli V, Bovolenta P, Alfano G, Marchitello A, Mocchetti C, Crippa L, Bulfone A, Marigo V, Ballabio A and Banfi S. Vax2 inactivation in mouse determines alteration of the eye dorsal-ventral axis, misrouting of the optic fibers and eye coloboma. *Development*, 2002, 129: 805-813.
38. Saglio G, Storlazzi CT, Giugliano E, SuraceC, Anelli L, Rege-Cambrin G, Zagaria A, Jimenez Velasco A, Heiniger A, Scaravaglio P, Torres Gomez A, Roman Gomez J, Archidiacono N, Banfi S, and Rocchi M. A 76-kb duplon maps close to the BCR gene on chromosome 22 and the ABL gene on chromosome 9: Possible involvement in the genesis of the Philadelphia chromosome translocation. *Proceedings of National Academy of Sciences USA*, 2002, 99(15): 9882-7.
39. Conte I, Lestingi M, den Hollander A, Miano M, Alfano G, Circolo D, Pugliese M, Testa F, Simonelli F, Rinaldi E, Baiget M, Banfi S, Ciccodicola A. Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. *Gene*, 2002, 297(1-2):33-8.
40. Reymond A, Marigo V, Yaylaoglu MB, Leoni A, Ucla C, Scamuffa N, Cacciopoli C, Dermitzakis ET, Lyle R, Banfi S, Eichele G, Antonarakis SE and Ballabio A. Human chromosome 21 gene expression atlas in the mouse. *Nature*, 2002, 420: 582-6.
41. Conte I, Lestingi M, den Hollander A, Alfano G, Ziviello C, Pugliese M, Circolo D, Cacciopoli C, Ciccodicola A, Banfi S. Identification and characterization of the retinitis pigmentosa 1-like1 gene (RP1L1): a novel candidate for retinal degenerations. *European Journal of Human Genetics*, 2003, 11: 155-162.
42. Ferrante M.I., Barra A., Truong J-P., Banfi S., Disteche C.M., Franco B. Characterization of the OFD1/Ofd1

- genes on the human and mouse sex chromosomes and exclusion of Ofd1 for the Xpl mouse mutant. *Genomics*, 2003 Jun;81(6):560-9.
43. Banfi S and Ballabio A. Disease-related genes: identification, *Nature encyclopedia of Human Genome*, editor D. Cooper, vol. 2: 38-41. 2003 MacMillan publisher.
 44. Lavorgna G, Lestingi M, Zivello C, Testa F, Simonelli F, Manitto MP, Brancato R, Ferrari M, Rinaldi E, Ciccodicola A, Banfi S. Identification and characterization of C1orf36, a transcript highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. *Biochem Biophys Res Commun*, 2003, 308(3):414-21.
 45. Simonelli F, Cennamo G, Zivello C, Testa F, De Crecchio G, Nesti A, Manitto MP, Ciccodicola A, Banfi S, Brancato R, Rinaldi E. Clinical features of X linked juvenile retinoschisis associated with new mutations in the XLRS1 gene in Italian families. *Br J Ophthalmol*, 2003, 87(9):1130-1134.
 46. Bulfone A, Cacciopoli C, Pardini C, Faedo A, Martinez S, Banfi S. Pcp4l1, a novel gene encoding a Pcp4-like polypeptide, is expressed in specific domains of the developing brain. *Gene Expression Patterns*, 2004, 4: 297-301.
 47. Criscuolo C, Banfi S, Orio M, Gasparini P, Monticelli A, Scarano V, Santorelli FM, Perretti A, Santoro L, De Michele G, and Filla A. A novel mutation in SACS gene in a family from southern Italy. *Neurology*, 2004, 62: 100-2
 48. Criscuolo C., Mancini P., Sacca F., De Michele G., Monticelli A., Santoro L., Scarano V., Banfi S., and Filla A.. Ataxia with oculomotor apraxia type 1 in Southern Italy: Late onset and variable phenotype. *Neurology*, 2004 63: 2173-2175.
 49. Vitiello C., D'Adamo P., Gentile F., Vingolo E.M., Gasparini P., and Banfi S. A Novel GJA1 Mutation Causes Oculodentodigital Dysplasia Without Syndactyly. *American Journal of Medical Genetics, A*. 2005 133(1):58-60.
 50. Alfano G, Vitiello C, Cacciopoli C, Caramico T, Carola A, Szego MJ, McInnes RR, Auricchio A, Banfi S. Natural antisense transcripts (NATs) associated with genes involved in eye development. *Hum Mol Genet*. 2005 Apr 1;14(7):913-23.
 51. Coppola G, Criscuolo C, De Michele G, Striano S, Barbieri F, Striano P, Perretti A, Santoro L, Brescia Morra V, Sacca F, Scarano V, D'Adamo AP, Banfi S, Gasparini P, Santorelli FM, Lehesjoki AE, Filla A. Autosomal recessive progressive myoclonus epilepsy with ataxia and mental retardation. *J Neurol*. 2005 Aug;252(8):897-900.
 52. Zivello C, Simonelli F, Testa F, Anastasi M, Bianchi Marzoli S, Falsini B, Ghiglione D, Macaluso C, Manitto MP, Garrè C, Ciccodicola A, Rinaldi E, and Banfi S. Molecular genetics of autosomal dominant retinitis pigmentosa (ADRP): a comprehensive study of 43 Italian families. *Journal of Medical Genetics*, 2005 Jul;42(7):e47
 53. Criscuolo C, Mancini P, Menchise V, Sacca F, De Michele G, Banfi S, Filla A. Very late onset in ataxia oculomotor apraxia type I. *Ann. Neurol*. 57, 777 (2005).
 54. Criscuolo C, Sacca F, De Michele G, Mancini P, Combarros O, Infante J, Garcia A, Banfi S, Filla A and Berciano J. Novel mutation of SACS gene in a Spanish family with autosomal recessive spastic ataxia. *Mov. Disord.* 20, 1358-61 (2005).
 55. Sud R, Jones CM, Banfi S and Dawson SJ. Transcriptional regulation by Barhl1 and Brn-3c in organ of corti derived cell lines. *Brain Res Mol Brain Res*. 2005 141(2):174-80.
 56. Antonini D, Rossi B, Han R, Minichiello A, Di Palma T, Corrado M, Banfi S, Zannini M, Brissette JL, and Missiro C. An autoregulatory loop directs the tissue-specific expression of p63 through a long-range evolutionary conserved enhancer. *Molecular and Cellular Biology*, 26: 3308-18 (2006).
 57. Criscuolo C, Chessa L, Di Giandomenico S, Mancini P, Sacca F, Grieco GS, Piane M, Barbieri F, De Michele G, Banfi S, Pierelli F, Rizzato N, Santorelli FM, Galloste L, Filla A, Casali C. Ataxia with oculomotor apraxia type 2: a clinical, pathologic, and genetic study. *Neurology* 66(8):1207-10 (2006).
 58. Cremers FP, Kimberling WJ, Kulm M, de Brouwer A, van Wijk E, Te Brinke H, Cremers CW, Hoefsloot LH, Banfi S, Simonelli F, Fleischhauer JC, Berger W, Kelley PM, Haralambous E, Bitner-Glindzicz M, Webster AR, Saihan Z, Debaere E, Leroy BP, Silvestri G, McKay G, Koenekoop RK, Millan JM, Rosenberg T, Joensuu T, Sankila EM, Weil D, Weston MD, Wissinger B, Kremer H. Development of a genotyping Microarray for usher syndrome. *J Med Genet*. 2007 Feb;44(2):153-60. Epub 2006 Sep 8.
 59. Karali M, Peluso I, Marigo V, Banfi S. Identification and characterization of microRNAs expressed in the mouse eye. *Invest Ophthalmol Vis Sci*, 48(2): 509-15 (2007).
 60. Testa F, Zivello C, Rinaldi M, Rossi S, Di Iorio V, Interlandi E, Ciccodicola A, Banfi S, Simonelli F. Clinical phenotype of an Italian family with a new mutation in the PRPF8 gene. *Eur J Ophthalmol*. 2006 Sep-Oct;16(5):779-81.

61. Simonelli F, Zivello C, Testa F, Rossi S, Fazzi E, Bianchi PE, Fossarello M, Signorini S, Bertone C, Galantuomo S, Brancati F, Valente EM, Ciccodicola A, Rinaldi E, Auricchio A, Banfi S. "Clinical and molecular genetics of Leber's congenital amaurosis (LCA): a multicenter study of Italian patients, *Invest Ophthalmol Vis Sci*, 48(9): 4284-90, 2007.
62. Costa V, Conte I, Zivello C, Casamassimi A, Alfano G, Banfi S, Ciccodicola A. Identification and expression analysis of novel Jakmip1 transcripts. *Gene*, 402(1-2):1-8, 2007. Epub 2007 Jul 14.
63. Criscuolo C, Mancini P, Ammendola S, Cicala D, Banfi S, De Michele G, Filla A. Screening for POLG1 mutations in a Southern Italian ataxia population. *J Neurol*. 255(3):454-5, 2008. Epub 2007 Dec 19.
64. Trifunovic D, Karali M, Campogampiero D, Ponzi D, Banfi S, Marigo V. A high-resolution RNA expression atlas of Retinitis Pigmentosa genes in the human and mouse retinas. *Invest Ophthalmol Vis Sci* 49(6):2330-6 (2008). Epub 2008 Feb 15.
65. Maguire AM, Simonelli F, Pierce EA, Pugh EN Jr, Mingozi F, Bennicelli J, Banfi S, Marshall KA, Testa F, Surace EM, Rossi S, Lyubarsky A, Arruda VA, Konkle B, Stone E, Sun J, Jacobs J, Dell'Osso L, Hertle R, Ma J, Redmond TM, Zhu-X, Hauck B, Zelenaia O, Shindler KS, Maguire MG, Fraser Wright J, Volpe NJ, Wellman McDonnell J, Auricchio A, High KA, Bennett J. Vision in a Safety Study of Gene Transfer for Leber Congenital Amaurosis. *New England Journal of Medicine*, 358(21):2240-8 (2008). Epub 2008 Apr 27.
66. Ginocchio VM, De Brasi D, Genesio R, Ciccone R, Gimelli S, Fimiani F, de Berardinis T, Nitsch L, Banfi S, Magli A, Della Casa R. Sonic Hedgehog deletion and distal trisomy 3p in a patient with microphthalmia and microcephaly, lacking cerebral anomalies typical of holoprosencephaly. *Eur J Med Genet*. 2008 Nov-Dec;51(6):658-65. Epub 2008 Aug 13.
67. Maselli V, di Bernardo D, Banfi S. CoGemiR: A Comparative Genomics microRNA database. *BMC Genomics* 2008, Oct 6;9:457.
68. Gennarino VA, Sardiello M, Avellino R, Meola N, Maselli V, Anand S, Cutillo L, Ballabio A, Banfi S. MicroRNA target prediction by expression analysis of host genes. *Genome Res*. 2009 Mar;19(3):481-90. Epub 2008 Dec 16
69. den Hollander A, McGee TL, Zivello C, Banfi S, Dryja TP, Gonzalez-Fernandez F, Ghosh D, Berson E. A homozygous missense mutation in the IRBP gene (RBP3) associated with autosomal recessive retinitis pigmentosa. *Invest Ophthalmol Vis Sci*. 2009 50(4):1864-72. Epub 2008 Dec 13.
70. Criscuolo C, Filla A, Coppola G, Rinaldi C, Carbone R, Pinto S, Wang Q, de Leva MF, Salvatore E, Banfi S, Brunetti A, Quarantelli M, Geschwind DH, Pappatà S, De Michele G. Two novel CYP7B1 mutations in Italian families with SPG5: a clinical and genetic study. *J Neurol*. 2009, 256(8):1252-7. Epub 2009 Apr 12.
71. Sardiello M, Palmieri M, di Ronza A, Medina DL, Valenza M, Gennarino VA, Di Malta C, Donaudy F, Embrione V, Polishchuk RS, Banfi S, Parenti G, Cattaneo E and Ballabio A. A gene network regulating lysosomal biogenesis and function. *Science*, 2009 325(5939):473-7. Epub 2009 Jun 25.
72. Grillo G, Turi A, Licciulli F, MignoneF, Liuni S, Banfi S, Gennarino VA, Horner DS, Pavesi G, Picardi E and Pesole G. UTRdb and UTRsite (release 2010): a collection of sequences and regulatory motifs of the untranslated regions of eukaryotic mRNAs. *Nucleic Acids Res*. 2010, 38(Database issue):D75-80. Epub 2009 Oct 30.
73. Maguire AM, High KA, Auricchio A, Wright, F, Pierce EA, Testa F, Mingozi F, Bennicelli J, Ying G, Rossi S, Fulton A, Marshall KA, Banfi S, Chung D, Morgan JIW, Hauck B, Zelenaia O, Zhu X, Raffini L, Coppieters F, De Baere E, Shindler KS, Volpe NJ, Surace EM, Acerra C, Lyubarsky A, Redmond TM, Stone E, Sun J, McDonnell JW, Leroy BP, Simonelli F, Bennett J. Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial. *Lancet* 2009, 374(9701):1597-605. Epub 2009 Oct 23.
74. Simonelli F, Maguire AM, Testa F, Pierce EA, Mingozi F, Bennicelli JL, Rossi F, Marshall K, Banfi S, Surace EM, Sun J, Redmond TM, Zhu X, ShindlerKS, Ying G, Zivello C, Acerra C, Wright JF, McDonnell JW, High KA, Bennett J, Auricchio A. Gene therapy for Leber's congenital amaurosis is safe and effective through 1.5 years after vector administration. *Mol Ther*. 2010 Mar;18(3):643-50. Epub 2009 Dec 1.
75. Meola N, Gennarino VA, Banfi S. microRNAs and genetic diseases. *PathoGenetics* 2009, 2:7 (4 November 2009).
76. Licastro D, Gennarino VA, Petrera F, Sanges R, Banfi S and Stupka. Promiscuity of enhancer, coding and non-coding transcription functions in ultraconserved elements. *BMC Genomics*. 2010 Mar 4;11(1):151.
77. De Cegli, R Romito A, Iacobacci S, Mao L, Lauria, M Fedele AO, Klose J, Borel C, Descombes P, Antonarakis SE, di Bernardo D, Banfi S, Ballabio A and Cobellis G. A mouse embryonic stem cell bank for inducible overexpression of human chromosome 21 genes. *Genome Biology* 2010, 11:R64

78. Bandah-Rozenfeld D, Collin RW, Banin E, Ingeborgh van den Born L, Coene KL, Siemiatkowska AM, Zelinger L, Khan MI, Lefeber DJ, Erdinest I, Testa F, Simonelli F, Voesenek K, Blokland EA, Strom TM, Klaver CC, Qamar R, Banfi S, Cremers FP, Sharon D, den Hollander AJ. Mutations in IMPG2, encoding interphotoreceptor matrix proteoglycan 2, cause autosomal-recessive retinitis pigmentosa. *Am J Hum Genet.* 2010 Aug 13;87(2):199-208. Epub 2010 Jul 30.
79. Conte I, Carrella S, Avellino R, Karali M, Marco-Ferreres R, Bovolenta P, Banfi S. miR-204 is required for lens and retinal development via Meis2 targeting. *Proc Natl Acad Sci U S A.* 2010 107(35):15491-6. Epub 2010 Aug 16.
80. Alfano G, Conte I, Caramico T, Avellino R, Arno B, Pizzo MT, Tanimoto N, Beck SC, Huber G, Dollé P, Seeliger M and Banfi S. Vax2 regulates retinoic acid distribution and cone opsin expression in the vertebrate eye. *Development.* 2011 138(2):261-71. Epub 2010 Dec 9.
81. Diez-Roux G, Banfi S, Sultan M, Geffers L, Anand S, Rozado D, Magen A, Canidio E, Pagani M, Peluso I, Lin-Marq N, Koch M, Bilio M, Cantuero I, Verde R, De Masi C, Bianchi SA, Cicchini J, Perroud E, Mehmeti S, Dagand E, Schrinner S, Nürberger A, Schmidt K, Metz K, Zwingmann C, Brieske N, Springer C, Martinez-Hernandez A, Herzog S, Grabbe F, Sieverding C, Fischer B, Schrader K, Büsing M, Schubert S, Helbig C, Alunni V, Battaini MA, Mura C, Henrichsen CN, Garcia-Lopez R, Echevarria D, Puelles E, Garcia-Calero E, Kruse S, Uhr M, Kauck C, Feng G, Milyaev N, Ong CK, Kumar L, Lam MS, Semple CA, Gyenesi A, Mundlos S., Radefol U, Lehrach H, Sarmientos P, Reymond A, Davidson DR, Dollé P, Antonarakis SE, Yaspo ML, Martinez M, Baldock RA, Eichele G, and Ballabio A. A high-resolution anatomical atlas of the transcriptome in the mouse embryo. *PLoS Biol.* 2011 9(1):e1000582.
82. Karali M, Peluso I, Gennarino VA, Bilio M, Verde R, Lago G, Dollé P and Banfi S. miRNeye: a microRNA expression atlas of the mouse eye. *BMC Genomics.* 2010 Dec 20;11:715.
83. Testa F, Surace EM, Rossi S, Marrocco E, Gargiulo A, Di Iorio V, Zivello C, Nesti A, Fecarotta S, Bacci ML, Giunti M, della Corte M, Banfi S*, Auricchio A* and Simonelli F*. Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. *Invest Ophthalmol Vis Sci.* 2011 Jul 29;52(8):5618-24. *co-corresponding authors.
84. Gennarino VA, Sardiello M, Mutarelli M, Dharmalingam G, Maselli V, Lago G and Banfi S. HOCTAR database: a unique resource for microRNA target prediction. *Gene.* 2011 Jul 1;480(1-2):51-8. Epub 2011 Mar 22.
85. Vozzi D, Aaspöllu A, Athanasakis E, Berto A, Fabretto A, Licastro D, Külm M, Testa F, Trevisi P, Vahter M, Zivello C, Martini A, Simonelli F, Banfi S, and Gasparini P. Molecular Epidemiology of Usher Syndrome in Italy. *Mol Vis.* 2011;17:1662-8. Epub 2011 Jun 22.
86. Karali M, Manfredi A, Puppo A, Marrocco E, Gargiulo A, Allocca M, Della Corte M, Rossi S, Giunti M, Bacci ML, Simonelli F, Surace EM, Banfi S and Auricchio A. microRNA-Restricted Transgene Expression in the Retina. *PLoS One.* 2011;6(7):e22166. Epub 2011 Jul 26.
87. Ozgül RK, Siemiatkowska AM, Yücel D, Myers CA, Collin RW, Zonneveld MN, Beryozkin A, Banin E, Hoyng CB, van den Born LI; European Retinal Disease Consortium*, Bose R, Shen W, Sharon D, Cremers FP, Klevering BJ, den Hollander AJ, Corbo JC. Exome sequencing and cis-regulatory mapping identify mutations in MAK, a gene encoding a regulator of ciliary length, as a cause of retinitis pigmentosa. *Am J Hum Genet.* 2011 Aug 12;89(2):253-64. *Sandro Banfi is part of this consortium.
88. Meola N, Pizzo M, Alfano G, Surace EM and Banfi S. The long non-coding RNA Vax2os1 controls the cell cycle progression of photoreceptor progenitors in the mouse retina. *RNA.* 2012 Jan;18(1):111-23. Epub 2011 Nov 29.
89. Cotugno G, Annunziata P, Karali M, Banfi S and Auricchio A. Impact of age at administration, lysosomal storage, and transgene regulatory elements on AAV2/8-mediated rat liver transduction. 1. *PLoS One.* 2012;7(3):e33286. Epub 2012 Mar 13.
90. Gennarino VA, D'Angelo G, Dharmalingam G, Fernandez S, Russolillo G, Sanges R, Mutarelli M, Belcastro V, Ballabio A, Verde A, Sardiello M and Banfi S. Identification of microRNA-regulated gene networks by expression analysis of target genes. *Genome Res.* 2012 Jun;22(6):1163-72. Epub 2012 Feb 24.
91. Neri M, Valli E, Alfano G, Bovolenta M, Spitali P, Rapezzi C, Muntoni F, Banfi S, Perini G, Gualandi F and Ferlini A. The absence of dystrophin brain isoform expression in healthy human heart ventricles explains the pathogenesis of 5' X-linked dilated cardiomyopathy. *BMC Medical Genetics* 2012, 13:20.
92. Testa F, Rossi S, Sodi A, Passerini I, Di Iorio V, Della Corte M, Banfi S, Surace EM, Menchini U, Auricchio A, Simonelli F. Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. *Invest Ophthalmol Vis Sci.* 2012 Jul 3;53(8):4409-15.

93. Licastro D, Mutarelli M, Peluso I, Neveling K, Wieskamp N, Rispoli R, Vozzi D, Athanasakis E, D'Eustachio A, Pizzo M, D'Amico F, Zivello C, Simonelli F, Fabretto A, Scheffer H, Gasparini P §, Banfi S§, Nigro V§. Molecular diagnosis of Usher syndrome: application of two different Next Generation Sequencing-based procedures. *PLoS One*. 2012;7(8):e43799. Epub 2012 Aug 29. §co-corresponding authors.
94. Estrada-Cuzcano A, Koenekoop R, Senechal A, De Baere E, de Ravel T, Banfi S, Kohl S, Ayuso C, Sharon D, Hoyng C, Hamel C, Leroy B, Zivello C, Lopez I, Bazinet A, Wissinger B, Sliesoratyte I, Avila-Fernandez A, Littink K, Vingolo E, Signorini S, Banin E, Mizrahi-Meissonnier L, Zrenner E, Kellner U, Collin RWJ, den Hollander A, Cremers F, Klevering B. BBS1 Mutations Underlie a Wide Spectrum of Phenotypes Ranging from Nonsyndromic Retinitis Pigmentosa to Bardet-Biedl Syndrome. *Arch Ophthalmol*. 2012 Nov 1;130(11):1425-32.
95. De Cegli R, Iacobacci S, Flore G, Gambardella G, Mao L, Cutillo L, Lauria M, Klose J, Illingworth E, Banfi S and di Bernardo D. Reverse engineering a mouse embryonic stem cell-specific transcriptional network reveals a new modulator of neuronal differentiation. *Nucleic Acids Res*. 2013 Jan;41(2):711-26. doi: 10.1093/nar/gks1136. Epub 2012 Nov 23.
96. Perrault I, Estrada-Cuzcano A, Lopez I, Kohl S, Li S, Testa F, Zekveld R, Wang X, Pomares E, Andorf J, Aboussair N, Banfi S, Delphin N, den Hollander A, Edelson C, Florijn R, JeanPierre M, Leowski C, Megarbane A, Villanueva C, Flores B, Munnich A, Ren H, Zobor D, Bergen A, Chen R, Cremers F, Gonzales-Duarte R, Koenekoop RK, Simonelli F, Stone E, Wissinger B, Zhang Q, Kaplan J, Rozet J-M. Union Makes Strength: A worldwide collaborative genetic and clinical study to provide a comprehensive survey of RD3 mutations and delineate the associated phenotype. *PLoS One*. 2013;8(1):e51622. Epub 2013 Jan 7.
97. Testa F, Maguire AM, Rossi S, Pierce EA, Melillo P, Marshall K, Banfi S, Surace EM, Sun J, Acerra C, Wright JF, Wellman J, High KA, Alberto A, Bennett J and Simonelli F. Three Year Follow-Up after Unilateral Subretinal Delivery of Adeno-Associated Virus in Patients with Leber Congenital Amaurosis Type 2. *Ophthalmology*. 2013 Jun;120(6):1283-91. doi: 10.1016/j.ophtha.2012.11.048. Epub 2013 Mar 6.
98. Sanges R, Hadzhiev Y, Gueroult-Bellone M, Roure A, Ferg M, Meola N, Amore G, Basu S, Brown E, De Simone M, Petrera F, Licastro D, Strähle U, Banfi S, Lemaire P, Birney E, Müller F, Stupka E. Highly conserved elements discovered in vertebrates are present in non-syntenic loci of tunicates, act as enhancers and can be transcribed during development. *Nucleic Acids Res*. 2013 Apr 1;41(6):3600-18. doi: 10.1093/nar/gkt030. Epub 2013 Feb 7.
99. Shaham O, Gueta K, Mor E, Oren-Giladi P, Grinberg D, Xie Q, Cvekl A, Shomron N, Davis N, Keydar-Prizant M, Raviv S, Pasmanik-Chor M, Bell RE, Levy C, Avellino R, Banfi S, Conte I, and Ashery-Padan R. Pax6 Regulates Gene Expression in the Vertebrate Lens through miR-204. *PLoS Genet*. 2013 Mar;9(3):e1003357. doi: 10.1371/journal.pgen.1003357. Epub 2013 Mar 14.
100. Peluso I, Conte I, Testa F, Dharmalingam G, Pizzo M, Collin RWJ, Meola N, Barbato S, Mutarelli M, Zivello C, Barbarulo AM, Nigro V, Melone MAB, the European Retinal Disease Consortium, Simonelli F, Banfi S. The ADAMTS18 gene is responsible for autosomal recessive Early Onset Severe Retinal Dystrophy. *Orphanet J Rare Dis*. 2013 Jan 28;8:16. doi: 10.1186/1750-1172-8-16.
101. Avellino R, Carrella S, Pirozzi M, Risolino M, Franco P, Stoppelli P, Verde P, Banfi S*, Conte I*. MiR-204 targeting of Ankrd13A controls both mesenchymal neural crest and lens cell migration. *PLoS One*, 2013 Apr 19;8(4):e61099. doi: 10.1371/journal.pone.0061099. *co-corresponding authors.
102. Conte I*, Banfi S*, Bovolenta P*. Noncoding RNAs in the development of sensory organs and related diseases. *Cell Mol Life Sci*. 2013 Nov;70(21):4141-55. doi: 10.1007/s00018-013-1335-z. Epub 2013 Apr 16. *co-corresponding authors.
103. Manes G, Meunier I, Avila-Fernandez A, Banfi S, Le Meur G, Zanolonghi X, Corton M, Simonelli F, Brabet P, Labesse G, Audi I, Mohand-Said S, Zeitz C, Sahel JA, Weber M, Dolifus H, Dhaenens CM, Allorge D, De Baere E, Koenekoop RK, Kohl S, Cremers FPM, Hollyfield JG, Senechal A, Hebrard M, Bocquet B, Ayuso Garcia C, and Hamel CP. Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. *Am J Hum Genet*. 2013 Sep 5;93(3):571-8. doi: 10.1016/j.ajhg.2013.07.018. Epub 2013 Aug 29.
104. Poulter JA, Al-Araimi M, Conte I, van Genderen MM, Sheridan E, Carr IM, Parry DA, Shires M, Carrella S, Bradbury J, Khan K, Lakeman P, Sergouniotis PI, Webster AR, Moore AT, Pal B, Mohamed MD, Venkataramana A, Ramprasad V, Shetty R, Saktivel M, Kumaramanickavel G, Tan A, Mackey DA, Hewitt AW, Banfi S, Ali M, Inglehearn CF & Toomes C. Recessive mutations in SLC38A8 cause foveal hypoplasia and optic nerve misrouting without albinism. *Am J Hum Genet*. 2013 Dec 5;93(6):1143-50. doi: 10.1016/j.ajhg.2013.11.002. Epub 2013 Nov 27.
105. Coppola A, Romito A, Borel C, Gehrigc C, Gagnebinc M, Falconnet E, Izzo A, Altucci L, Banfi S, Antonarakis SE, Minichiotti G and Cobellis G. Cardiomyogenesis is controlled by the miR-99a/let-7c cluster and epigenetic modifications. *Stem Cell Res*. 2013 Nov 28;12(2):323-337. doi: 10.1016/j.scr.2013.11.008. [Epub ahead of print]
106. Conte I, Merella S, Garcia Manteiga, JM, Migliore C, Lazarevic D, Carrella S, Avellino R, Marco-Ferreres R,

- Emmett W, Sanges R, Bockett N, Davidson NP, Meroni G, van Heel D, Bovolenta P, Stupka E, Banfi S. The combination of transcriptomics and informatics identifies pathways targeted by miR-204 during neurogenesis and axon guidance. *Nucleic Acids Res.* 2014;42(12):7793-806. doi: 10.1093/nar/gku498. Epub 2014 Jun 3.
107. Fernandez S, Risolino M, Mandia N, Talotta F, Soini Y, Incoronato M, Condorelli G, Banfi S, Verde P. miR-340 inhibits tumor cell proliferation and induces apoptosis by targeting multiple negative regulators of p27 in non-small cell lung cancer. *Oncogene*. 2014 Aug 25;0. doi: 10.1038/onc.2014.267. [Epub ahead of print]
108. Karali M, Banfi S. Inherited Retinal Dystrophies: The role of gene expression regulators. *Int J Biochem Cell Biol*. 2015 Feb 16. pii: S1357-2725(15)00050-3. doi: 10.1016/j.biocel.2015.02.007. [Epub ahead of print]
109. Carrella S, D'Agostino Y, Barbato S, Huber-Reggi SP, Salierno FG, Manfredi A, Neuhauss SCF, Banfi S*, Conte I*. miR-181a/b control the assembly of visual circuitry by regulating retinal axon specification and growth. *Dev Neurobiol*. 2015 Nov;75(11):1252-67. doi: 10.1002/dneu.22282. Epub 2015 Jun 11. *co-corresponding authors.
110. Di Frusco G, Schulz A, De Cegli R, Savarese M, Mutarelli M, Parenti G, Banfi S, Braulke T, Nigro V, Ballabio A. LYSOPLEX: an efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway. *Autophagy* 2015, 11:6, 928-938, doi: 10.1080/15548627.2015.1043077.
111. Trapani I, Banfi S, Simonelli F, Surace E, Auricchio A. Gene therapy of inherited retinal degenerations: prospects and challenges. *Hum Gene Ther*. 2015 Apr;26(4):193-200. doi: 10.1089/hum.2015.030.
112. Conte I, Hadfield KD, Barbato S, Carrella S, Pizzo M, Bhat RS, Carissimo A, Karali M, Porter LF, Urquhart J, Hateley S, O'Sullivan J, Manson F, Neuhauss SCF, Banfi S*, and Black GCM*. MiR-204 is responsible for inherited retinal dystrophy associated with ocular coloboma. *PNAS*, 2015 Jun 23;112(25):E3236-45. doi: 10.1073/pnas.1401464112. Epub 2015 Jun 8. *co-corresponding authors.
113. Saksens NTM, Krebs MP, Schoenmaker-Koller FE, Hicks W, Yu M, Shi L, Rowe L, Collin GB, Charette JR, Letteboer SJ, Neveling K, van Moorsel TW, Abu-Ltaif S, De Baere E, Walraedt S, Banfi S, Simonelli F, Cremers FPM, Boon CJF, Roepman R, Leroy BP, Peachey NS, Hoyng CB, Nishina PM and den Hollander AJ. Mutations in α -catenin 1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. *Nat Genet*. 2015 Dec 21. doi: 10.1038/ng.3474. [Epub ahead of print]
114. Carrella S, Barbato S, D'Agostino Y, Salierno FG, Manfredi A, Banfi S*, Conte I*. TGF- β controls miR-181/ERK regulatory network during retinal axon specification and growth. *PLoS One*. 2015 Dec 7;10(12):e0144129. doi: 10.1371/journal.pone.0144129. eCollection 2015. *co-corresponding authors.
115. Karali M, Persico M, Mutarelli M, Carissimo A, Pizzo M, Marwah V, Ambrosio C, Pinelli M, Carrella D, Ferrari S, Ponzin D, Nigro V, Di Bernardo D, Banfi S. High-resolution analysis of the human retina miRNome reveals isomiR variations and novel microRNAs. *Nucleic Acids Res*. 2016 Feb 29;44(4):1525-40. doi: 10.1093/nar/gkw039.
116. Pinelli M, Carissimo A, Cutillo L, Lai C, Mutarelli M, Moretti N, Veer Singh M, Karali M, Carrella D, Pizzo M, Russo F, Ferrari S, Ponzin D, Angelini C, Banfi S*, Di Bernardo D*. An atlas of gene expression and gene co-regulation in the human retina. *Nucleic Acids Res*. 2016 Jul 8;44(12):5773-84. doi: 10.1093/nar/gkw486.*co-corresponding authors.
117. Rossi S, De Rosa G, D'Alterio FM, Orrico A, Banfi S, Testa F, Simonelli F. Intrafamilial heterogeneity of congenital optic disc pit maculopathy. *Ophthalmic Genet*. 2017 May-Jun;38(3):267-272. doi: 10.1080/13816810.2016.1188120. Epub 2016 Jun 8.
118. Petit C, Bonnet C, Riahi Z, Chantot-Bastaraud S, Smagghe L, Letexier M, Marcaillou C, Lefevre G, Hardelin J-P, El Amraoui A, Singh-Estivale A, Mohand-Saïd S, Kohl S, Kurtenbach A, Sliesoraityte I, Zobor D, Gherbi S, Testa F, Simonelli F, Banfi S, Fakin A, Glavač D, Jarc-Vidmar M, Zupan A, Battelino S, Martorell L, Claveria, MA Catala-Mora J, Dad S, Møller L, Rodriguez Jorge J, Hawlina M, Auricchio A, Sahel JA, Marlin S, Zrenner E, and Audo I. An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. *Eur J Hum Genet*. 2016 Dec;24(12):1730-1738. doi: 10.1038/ejhg.2016.99.
119. Piccolo P, Attanasio S, Secco I, Sangermano R, Strisciuglio C, Limongelli G, Miele E, Mutarelli M, Banfi S, Nigro V, Pons T, Valencia A, Zentilin L, Campione S, Nardone G, Lynnes TC, Celestino-Soper PB, Spoonamore KG, D'Armiento FP, Giacca M, Staiano A, Vatta M, Colles C, Brunetti-Pierri N. MIB2 variants altering NOTCH signalling result in left ventricle hypertrabeculation/non-compaction and are associated with Ménétrier-like gastropathy. *Hum Mol Genet*. 2017 Jan 1;26(1):33-43. doi: 10.1093/hmg/ddw365.
120. Testa F, Filippelli M, Brunetti-Pierri R, Di Frusco G, Di Iorio V, Pizzo M, Torella A, Barillari MR, Nigro V, Brunetti-Pierri N, Simonelli F, and Banfi S. Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. *Eur J Hum Genet*. 2017 May;25(5):651-655. doi: 10.1038/ejhg.2017.23.
121. Di Iorio V, Karali M, Brunetti-Pierri R, Filippelli M, Di Frusco G, Pizzo M, Mutarelli M, Nigro V, Testa F, Banfi S*, Simonelli S*. Clinical and genetic evaluation of a cohort of pediatric patients with severe Inherited Retinal

- Dystrophies. *Genes (Basel)*. 2017 Oct 20;8(10). pii: E280. doi: 10.3390/genes8100280. *co-corresponding authors.
122. Barbato S, Marrocco E, Intartaglia D, Pizzo M, Asteriti S, Naso F, Falanga D, Bhat R, Meola N, Carissimo A, Karali M, Prosser H, Cangiano L, Surace EM, Banfi S*, and Conte I*. MiR-211 is essential for adult cone photoreceptor maintenance and visual function. *Sci Rep*. 2017 Dec 5;7(1):17004. doi: 10.1038/s41598-017-17331-z. *co-corresponding authors.
123. Tarallo A, Carissimo A, Gatto F, Nusco E, Banfi S, Toscano A, Musumeci O, Coletta M, Karali M, Acampora E, Damiano C, Minopoli N, Fecarotta S, Della Casa R, Mongini T, Vercelli L, Santoro L, Ruggiero L, Deodato F, Taurisano R, Bembi B, Dardis A, Pim Pijnappel WW, van der Ploeg AT, Parenti G. "MICRO-RNA AS BIOMARKERS IN POMPE DISEASE", *Genet Med*. 2018 Jul 12. doi: 10.1038/s41436-018-0103-8.
124. Karali M, Banfi S. Non-coding RNAs in retinal development and function. *Hum Genet*. 2018 Sep 5. doi: 10.1007/s00439-018-1931-y.
125. Van de Sompele S, Smith C, Karali M, Corton M, Van Schil K, Peelman F, Cherry T, Rosseel T, Verdin H, Derolez J, Van Laethem T, Khan KN, McKibbin M, Toomes C, Ali M, Torella A, Testa F, Jimenez B, Simonelli F, De Zaeytijd J, Van den Ende J, Leroy BP, Coppieeters F, Ayuso C, Inglehearn CF, Banfi S*, De Baere E*. *co-corresponding authors. Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. *Genet Med*. 2018 Oct 31. doi: 10.1038/s41436-018-0345-5.
126. Musacchia F, Ciolfi A, Mutarelli M, Bruselles A, Castello R, Pinelli M, Basu S, Banfi S, Casari G, Tartaglia M, Nigro V, TUDP. VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. *BMC Bioinformatics*. 2018 Dec 12;19(1):477. doi: 10.1186/s12859-018-2532-4.
127. Indrieri A, Carrella S, Romano A, Spaziano A, Marrocco E, Fernandez-Vizarra E, Barbato S, Pizzo M, Ezhova Y, Golia FM, Ciampi L, Tammaro R, Henao-Mejia J, Williams A, Flavell FA, De Leonibus E, Zeviani M, Surace EM, Banfi S*, Franco B*. *co-corresponding authors. miR-181a/b downregulation exerts a protective action on Mitochondrial Disease models. *EMBO Mol Med*. 2019 May;11(5). pii: e8734. doi: 10.15252/emmm.201708734.
128. Cappuccio G, Brunetti-Pierri R, Torella A, Pinelli M, Castello R, Casari G, Nigro V, Banfi S, Simonelli F, TUDP and Brunetti-Pierri N. Retinal dystrophy in a individual carrying a de novo missense variant of SMARCA4. *Mol Genet Genomic Med*. 2019 Apr 11:e682. doi: 10.1002/mgg3.682. [Epub ahead of print]
129. Cappuccio G, Attanasio S, Alagia M, Mutarelli M, Borzone R, Karali M, Genesio R, Mormile A, Nitsch L, Imperati F, Esposito A, Banfi S, Del Giudice E, Brunetti-Pierri N. Microdeletion of pseudogene chr14.232.a affects LRFN5 expression in cells of a patient with autism spectrum disorder. *Eur J Hum Genet*. 2019 May 31. doi: 10.1038/s41431-019-0430-5. [Epub ahead of print]
130. Karali M, Guadagnino I, Marrocco E, De Cegli R, Carissimo A, Pizzo M, Casarosa S, Conte I, Surace EM, Banfi S. AAV-miR-204 Protects from Retinal Degeneration by Attenuation of Microglia Activation and Photoreceptor Cell Death. *Mol Ther Nucleic Acids*. 2019 Nov 18;19:144-156. doi: 10.1016/j.omtn.2019.11.005. [Epub ahead of print]
131. Karali M, Testa F, Brunetti-Pierri R, Di Iorio V, Pizzo M, Paolo Melillo, Barillari MR, Torella A, Musacchia F, D'Angelo L, Banfi S*, Simonelli F*. Clinical and genetic analysis of a European cohort with pericentral retinitis pigmentosa. *Int J Mol Sci*. 2019 Dec 20;21(1). pii: E86. doi: 10.3390/ijms21010086. *co-corresponding authors
132. Khan M, Cornelis SS, del Pozo-Valero M, Whelan L, Runhart EH, Mishra K, Bults F, AlSwaiti Y, AlTabishi A, De Baere E, Banfi S, Banin E, Bauwens M, Ben-Yosef T, Boon CJF, van den Born LI, Defoort S, Devos A, Dockery A, Dudakova L, Fakin A, Farrar GJ, Ferraz Sallum JM, Fujinami K, Gilissen C, Glavač D, Gorin MB, Greenberg J, Hayashi T, Hettinga Y, Hoischen A, Hoyng CB, Hufendiek K, Jägle H, Kamakari S, Karali M, Kellner U, Klaver CCW, Kousal B, Lamey T, MacDonald IM, Matynia A, McLaren T, Mena MD, Meunier I, Miller R, Newman H, Ntzozini B, Oldak M, Pieterse M, Podhajcer OL, Puech B, Ramesar R, Rüther K, Salameh M, Vallim Salles M, Sharon D, Simonelli F, Spital G, Steehouwer M, Szaflik JP, Thompson JA, Thuillier C, Tracewska AM, van Zweeden M, Vincent AL, Zanlonghi X, Liskova P, Stöhr H, De Roach J, Ayuso C, Roberts L, Weber BHF, Dhaenens C-M, Cremers, FPM. Resolving the dark matter of ABCA4 for 1,054 Stargardt disease probands through integrated genomics and transcriptomics. *Genet Med*. 2020 Apr 20. doi: 10.1038/s41436-020-0787-4.
133. Naso F, Intartaglia D, Falanga D, Soldati C, Polishchuk E, Giamundo G, Tiberi P, Marrocco E, Scudieri P, Di Malta C, Trapani I, Nusco E, Salierro FG, Surace EM, Galietta LJV, Banfi S, Auricchio A, Ballabio A, Medina DL, Conte I. Light-responsive microRNA miR-211 targets Ezrin to modulate lysosomal biogenesis and retinal cell clearance. *EMBO J*. 2020 Mar 10:e102468. doi: 10.15252/embj.2019102468. [Epub ahead of print]
134. Bedoni N, Quinodoz M, Pinelli M, Cappuccio G, Torella A, Nigro V, Testa F, Simonelli F, TUDP (Telethon Undiagnosed Disease Program), Corton M, Lualdi S, Lanza F, Morana G, Ayuso C, Di Rocco M, Filocamo M, Banfi S, Brunetti-Pierri N, Superti-Furga A, Rivolta C. An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome affecting multiple tissues and organs. *Hum Mol Genet*. 2020 Jun

12:ddaa112. doi: 10.1093/hmg/ddaa112. Online ahead of print.

135. Barillari MR, Karali M, Di Iorio V, Contaldo M, Piccolo V, Esposito M, Costa G, Argenziano G, Serpico R, Carotenuto M, Cappuccio G, Banfi S, Melillo P, Simonelli F. Mild form of Zellweger Spectrum Disorders (ZSD) due to variants in PEX1: detailed clinical investigation in a 9-years-old female. *Mol Genet Metab Rep*. 2020 Jun 20;24:100615. doi: 10.1016/j.ymgmr.2020.100615. eCollection 2020 Sep.
136. Olivier G, Corton M, Intartaglia D, Verbakel SK, Sergouniotis PI, Le Meur G, Dhaenens CM, Naacke H, Avila-Fernández A, Hoyng CB, Klevering J, Bocquet B, Roubertie A, Sénechal A, Banfi S, Muller A, Hamel CL, Black GC, Conte I, Roosing S, Zanlonghi X, Ayuso C, Meunier I, Manes G. Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. *J Med Genet*. 2020 Aug 17:jmedgenet-2020-107150. doi: 10.1136/jmedgenet-2020-107150. Online ahead of print.
137. Runhart EH, Khan M, Cornelis SS, Roosing S, Del Pozo-Valero M, Lamey TM, Liskova P, Roberts L, Stöhr H, Klaver CCW, Hoyng CB, Cremers PFM, Dhaenens CM; ABCA4 Disease Consortium Study Group*. Association of Sex With Frequent and Mild ABCA4 Alleles in Stargardt Disease. *JAMA Ophthalmol*. 2020 Aug 20:e202990. doi: 10.1001/jamaophthalmol.2020.2990. Online ahead of print. *Sandro Banfi is part of this consortium.
138. Cappuccio G, Sayou C, Tanno PL, Tisserant E, Bruel AL, Kennani SE, Sá J, Low KJ, Dias C, Havlovicová M, Hančárová M, Eichler EE, Devillard F, Moutton S, Van-Gils J, Dubourg C, Odent S, Gerard B, Piton A, Yamamoto T, Okamoto N, Firth H, Metcalfe K, Moh A, Chapman KA, Aref-Eshghi E, Kerckhof J, Torella A, Nigro V, Perrin L, Piard J, Le Guyader G, Jouan T, Thauvin-Robinet C, Duffourd Y, George-Abraham JK, Buchanan CA, Williams D, Kini U, Wilson K; Telethon Undiagnosed Diseases Program*, Sousa SB, Hennekam RCM, Sadikovic B, Thevenon J, Govin J, Vitobello A, Brunetti-Pierri N. De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. *Genet Med*. 2020 Jul 22. doi: 10.1038/s41436-020-0898-y. Online ahead of print. *Sandro Banfi is part of this consortium.
139. Kuehlewein L, Zobor D, Andreasson SO, Ayuso C, Banfi S, Bocquet B, Bernd AS, Biskup S, Boon CJF, Downes SM, Fischer MD, Holz FG, Kellner U, Leroy BP, Meunier I, Nasser F, Rosenberg T, Rudolph G, Stingl K, Thiadens AAHJ, Wilhelm B, Wissinger B, Zrenner E, Kohl S, Weisschuh N; RD-CURE Consortium. *JAMA Ophthalmol*. 2020 Oct 15:e204206. doi: 10.1001/jamaophthalmol.2020.4206. Online ahead of print.
140. Di Iorio V, Karali M, Melillo P, Testa F*, Brunetti-Pierri R, Musacchia F, Condroyer C, Neidhardt J, Audi I, Zeitz C, Banfi S* and Simonelli F*. Spectrum of disease severity in patients with X-linked Retinitis pigmentosa due to RPGR mutations. *Invest Ophthalmol Vis Sci*. 2020 Dec 1;61(14):36. doi: 10.1167/iovs.61.14.36. *co-corresponding authors