

**EUROPEAN CURRICULUM
VITAE FORMAT**



Personal information

First and Last Name Sandro Banfi
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Citizenship Italiana
Birth Date 3 giugno 1964
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Work Experience

CURRENT POSITION	Full Professor of Medical Genetics University of Campania “Luigi Vanvitelli” Naples, Italy
	Associate Investigator, Telethon Institute of Genetics and Medicine TIGEM Pozzuoli (NA), Naples
1991-1994	Post-doc Department of Pediatrics, Baylor College of Medicine, Houston
From 1995	Researcher Telethon Institute of Genetics and Medicine (T.I.G.E.M.)
From 2005	Coordinator of the UK Open University PhD Program in Human Genetics
From 2014	Lecturer, PhD Program in Biomolecular Sciences, University of Campania “Luigi Vanvitelli”
From 2004	Lecturer, European School of Molecular Medicine (SEMM)
2011-2012	Assistant Professor in Medical Genetics,

	Second University of Napoli (now University of Campania "Luigi Vanvitelli")
2012-2019	Associate Professor in Medical Genetics
Education and training	
1993	Residency in Neurology (<i>summa cum laude</i>) Institute of Neurology, University of Naples
1989	License School of Medicine, University of Naples
1989	Medical Degree (<i>summa cum laude</i>) School of Medicine, University of Naples
Personal Skills and competences	
Mother tongue	ITALIAN
Other languages	ENGLISH
• Reading skills	Excellent
• Writing skills	Excellent
• Verbal skills	Excellent
Pagina 2/15 - Curriculum vitae di BANFI Sandro	Per ulteriori informazioni: www.cedefop.eu.int/transparency www.europa.eu.int/comm/education/index_it.html www.eurescv-search.com

SCIENTIFIC ACTIVITIES

Reviewer activities for International scientific journals

Activity carried out for the following journals:
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

Reviewer activities for Research Grant Agencies

Activity carried out for the following agencies:
Italian Ministry of Research and University,
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

Affiliations to scientific societies	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)
Scientific Meeting organization	Co-organizer of the workshop "The Biology and Development of the Eye in Health and Disease", 170 Convegno IGB, Capri, Capri, 9-12 Ottobre, 2004. Co-organizer of the Workshop "Next Generation Sequencing in sensory disorders" 11 Febbraio 2011, Leuven, Belgio. Co-organizer of the Workshop "Next Generation Sequencing – Application cases and bioinform development", German-Italian Dialogue 2012, 17-19 July, Naples, Italy. Member of the Scientific Committee, XXII Meeting of the Italian Society of Human Genetics (SIGU), Rome, November 13-16, 2019
Membership to scientific councils	From 2006, Member of the Scientific Council of Retina Italia Onlus. 2017, Member of the Scientific Advisory Board, Institut de la Vision, Paris, France From 2007, member of the European Retinal Disease Consortium (ERDC, http://www.erdc.info/), consortium composed of 18 international research groups working on the definition of the molecular basis of inherited retinal diseases.
Awards and Honors	2009, "Board of Director Awards" of the US Foundation Fighting Blindness (FFB). 2017, Visionary of the Quarter, European Visionary of the Quarter, European Vision Institute
Patents	Inventor, patent entitled "mir-204 and mir-211 and uses thereof", deposited as PCT on March 11 2014, publication WO201414005. Granted in Europe, China, United States. Inventor, patent request entitled "mir-181 inhibitors and uses thereof", deposited on April 20, 2018.

Research support (with peer review)	<p>Selected National Grants</p> <ol style="list-style-type: none"> 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)." Funded by 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". Funded by Retina Italia onlus (2010-2011). 4. "Genetic bases of birth defects" Funded by Ministero Italiano della Salute (2008-2010). 5. "microRNA-regulated gene networks in the retina." Funded by Fondazione Telethon (2011-2016) 6. "Retinitis Pigmentosa: an integrated application of novel strategies towards diagnosis and treatment". Funded by Fondazione Roma (2015-2019). 7. "Systematic search for microRNAs that play a role in photoreceptor degeneration". Funded by Fondazione Telethon (2017-2018) 8. "Toward new methods for early diagnosis and screening of genetic ocular diseases in childhood". Funded by Ministero italiano della Istruzione, Universita' e Ricerca (Progetto PRIN) (2017-2020).
	<p>Selected International grants</p> <ol style="list-style-type: none"> 1. "EUROPEAN RETINAL RESEARCH TRAINING NETWORK (RETNET)". Funded by European Union (2004-2007). 2. "Functional genomics of the retina in health and disease (Evi-Genoret)". Funded by European Union (2005-2009). 3. "Novel tool for high-throughput characterization of genomic elements regulating gene expression in chordates. (TRANSCODE)" Funded by European Union (2005-2008). 4. Technological innovation of high throughput molecular diagnostics of clinically and molecularly heterogeneous genetic disorders. (Techene)". Funded by European Union (2009-2012). 5. "EyeTN - Beyond the Genome; training the next generation of ophthalmic researchers." Funded by European Union (2012-2016). 6. "MicroRNA miR-204, a new potential therapeutic tool for inherited retinal dystrophies". Funded by the US Foundation Fighting Blindness (FFB) (2016-2019). 7. "StarT- European Training Network to Diagnose, Understand and Treat Stargardt Disease, a Frequent Inherited Blinding Disorder". Funded by the European Union (2018-2022). 8. "AAV-Sponge-mediated modulation of microRNA-181a/b: a potential therapeutic approach for Inherited Retinal Disease". Funded by the US Foundation Fighting Blindness (FFB) (2019-2022).
Scientific publications	<p>Author of over 130 peer-reviewed scientific publications that reach overall the following bibliometric values:</p> <ul style="list-style-type: none"> -Total Impact Factor = 1001.983 -Average Impact Factor medio = 7.89 -Total Number of citations = 10897 (Scopus); 10174 (WoS); 14632 (Google Scholar) -Average Number of citations/article = 80.87 (Scopus); 77.075 (WoS); 104.5 (Google Scholar) -H-Index = 50 (Google Scholar); 45 (Scopus); 44 (WoS)

LIST OF SCIENTIFIC PUBLICATIONS

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, Sandkuyl 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, Marchitiello 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, Marchitiello 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, Marchitiello 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, Marchitiello 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, Marchitiello A, Moccetti 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, Surace C, 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, Zivello 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.

50. 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.
51. 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.
52. 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.
53. 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
54. 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).
55. 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).
56. 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.
57. 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).
58. 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, Gallotti L, Filla A, Casali C. Ataxia with oculomotor apraxia type 2: a clinical, pathologic, and genetic study. *Neurology* 66(8):1207-10 (2006).
59. 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, Sainan 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.
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61. 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.
62. 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.
63. 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.
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65. Trifunovic D, Karali M, Campogampiero D, Ponzin 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.

66. 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.
67. 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.
68. Maselli V, di Bernardo D, Banfi S. CoGemiR: A Comparative Genomics microRNA database. *BMC Genomics* 2008, Oct 6:9:457.
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