

**FORMATO EUROPEO
PER IL CURRICULUM
VITAE**



Informazioni personali

Nome e Cognome Sandro Banfi

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Cittadinanza Italiana

Sesso M

Esperienza professionale

POSIZIONE ATTUALE Professore Ordinario, Genetica Medica
Universita' degli Studi della Campania "Luigi Vanvitelli".

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
Universita' degli Studi della Campania "Luigi Vanvitelli"

Dal 2004 Docente
Scuola Superiore Europea di Medicina Molecolare (SEMM)

2011-2012 Ricercatore Universitario, Genetica Medica
Seconda Universita' 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 (*summa cum laude*)
Clinica Neurologica, Università di Napoli

1989 Abilitazione professionale
Il Facoltà di Medicina e Chirurgia Università di Napoli

1989 Laurea in Medicina (*summa cum laude* 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	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,

	<p>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</p>
Attività di revisore per Enti di supporto della ricerca	<p>Attività svolta per le seguenti agenzie di finanziamento: Ministero Università 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</p>
Affiliazioni a società scientifiche	<p>American Society of Human Genetics (ASHG) Association for Research in Vision and Ophthalmology (ARVO) European Society of Human Genetics (ESHG) Società Italiana di Genetica Umana (SIGU) Bioinformatic Italian Society (BITS)</p>
Membro di comitati organizzativi (nazionali ed internazionali)	<p>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.</p>
Membro di consigli scientifici	<p>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.erd.c.info/), consorzio composto da 18 gruppi internazionali per la definizione delle basi molecolari delle distrofie retiniche ereditarie.</p>
Premi Scientifici e onorificenze	<p>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, Università degli Studi di Napoli.</p>
Brevetti	<p>Inventore del brevetto dal titolo "mir-204 and mir-211 and uses thereof", depositato come PCT l'11Marzo</p>
Pagina 3/13 - Curriculum vitae di BANFI Sandro	<p>Per ulteriori informazioni: www.cedefop.eu.int/transparency www.europa.eu.int/comm/education/index_it.html www.eurescv-search.com</p>

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, Università 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 = 55 (Google Scholar); 48 (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, Coccozza 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 Coccozza 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, Marchitello 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, Marchitello 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, Marchitello A, Wapenaar MC, Borsani G, Giglio S, Mariani M, Consalez GG, Zuffardi O, Franco B, Ballabio A, Banfi S: Characterization of Cxor5 (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, Galletta 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, Fritsch 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, 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 duplicon 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, Caccioppoli 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, Caccioppoli 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, Ziviello 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, Ziviello 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, Caccioppoli C, Pardini C, Faedo A, Martinez S, Banfi S. Pcp411, 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, Caccioppoli 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. Ziviello 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 Missero 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, Rizzuto N, Santorelli FM, Gallosti 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-Glindzic 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, Ziviello 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, Ziviello 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, Ziviello 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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