CURRICULUM VITAE ET STUDIORUM

Andrea Ballabio M.D

Date and place of birth: 27 January, 1957, Naples, Italy

CURRENT POSITIONS: Director, Telethon Institute of Genetics and Medicine (TIGEM)

Professor of Medical Genetics, Faculty of Medicine, "Federico II" University, Naples, Italy

Visiting Professor, Dept. of Molecular and Human Genetics, Baylor College of Medicine, Jan and Dan Duncan Neurological Research Institute, Texas Children Hospital, Houston, TX

Visiting Professor, Department of Pharmacology, University of Oxford, UK

Professor, SSM School for Advanced Studies, University of Naples "Federico II", Naples, Italy.

Co-Founder CASMA Therapeutics, Boston, MA, USA

EDUCATION

Year	Institution	Degree
1981	Faculty of Medicine, University of Naples "Federico II", Italy	M.D. degree (cum laude)
1985	Department of Pediatrics, Faculty of Medicine, University of Naples "Federico II", Italy	Residency in Pediatrics (cum laude)

POST-DOCTORAL ACTIVITY

1984-85 Research Assistant at the Paediatric Research Unit, Guy's Hospital, London, England
Supervisors: Dr. M. Adinolfi, Dr. F. Giannelli

1986-87	Research Fellow at the International Institute of Genetics and Biophysics (IIGB), CNR, Naples, Italy Supervisor: Dr. M.G. Persico
1988-89	Research Fellow at the Department of Pediatrics Faculty of Medicine, University of Naples "Federico II", Italy Supervisor: Prof. G. Andria
1989	Research Associate at the Institute of Molecular Genetics, Baylor College of Medicine, Houston TX Supervisor: Prof. C.T. Caskey
1990-92	Assistant Professor, Institute for Molecular Genetics, Baylor College of Medicine, Houston TX
1992	Associate Professor of Medical Genetics at the Faculty of Medicine, Universita' Cattolica del Sacro Cuore, Rome, Italy
1992-94	Associate Professor, Institute for Molecular Genetics, Baylor College of Medicine, Houston TX
1993-94	Co-Director Human Genome Center, Baylor College of Medicine, Houston TX
1993-97	Professor in Medical Genetics, Department of Molecular Biology, University of Siena, Italy
1994-present	Director, Telethon Institute of Genetics and Medicine (TIGEM), Italy
1998-2000	Professor of Genetics, Faculty of Medicine, San Raffaele Università Vita e Salute, Milan, Italy
2000- 2003	Professor of Medical Genetics, Faculty of Medicine, II University of Naples, Italy
2003- present	Professor of Medical Genetics, Faculty of Medicine, "Federico II" University, Naples, Italy
2009-present	Visiting Professor, Dept. of Molecular and Human Genetics, Baylor College of Medicine, Jan and Dan Duncan Neurological Research Institute, Texas Children Hospital, Houston, TX
2013-present	Visiting Professor, Department of Pharmacology, University of Oxford, UK
2018-present	Co-founder CASMA Therapeutics, Boston, MA, USA
2020-present	Coordinator of graduate program in Genomic and Experimental Medicine for the SSM School of Advanced Studies

HONORS

1998	President of the European Society of Human Genetics (ESHG)

1995-2000	Council member of the Human Genome Organization (HUGO)
1998- present	Member of the European Molecular Biology Organization (EMBO)
2006	Torchbearer at the XX Torino 2006 Olympic Winter Games – Turin 2006
2007	"Knight" of the Order of Merit of Italy
2007	18th on Board of the "Luna Rossa" Challenge, America's Cup, Valencia, Spain
2007	Silver Medal of the President of Italy
2009-2012	Council Member of the European Molecular Biology Organization (EMBO)
2010-2014	Recipient of the Advanced Investigator Award of the European Research Council
2016	Winner of the Louis-Jeantet Prize for Medicine.
2016-2021	Recipient of the Advanced Investigator Award of the European Research Council
2017	Chair of the Gordon Conference on Lysosomal diseases.
2020	Member of the "Accademia Nazionale delle Scienze detta dei XL"
2020	"Grande Ufficiale" of the Italian Republic

ADVISORY BOARDS

- BIOMED 2 Ad Hoc Working Group on Human Genome Research - *1995-1996*
- l'Institut Fédératif de Recherche des Enfants Malades (IFREM) Advisory Board - *1995-present*
- The Vision of Children Advisory Board, San Diego, CA - *1994*
- German Human Genome Project Advisory Board - *1995-2000*
- Board Member of the European Society of Human Genetics (ESHG) – 1995-2000
- External Advisor for New Research Initiatives, University of Antwerp, Belgium – *1996*
- Chair of the European Commission BIOMED 2 Review Panel on Human Genome Research - *1996*
- Ingenium Pharmaceuticals Scientific Advisory Board, Martinsried, Germany, *1999-2000*
- Member of the French Scientific Commission for the Human Genome Network (French Research Ministry)- *2001*
- Member of BioMedNet reviews Advisory Committee - *2003*
- Member of the European Society of Gene Therapy Advisory Committee – *2003*
- Member of the Advisory Group of VI Programme for European Community

Research and Technology – *2001-2006*
- Fondazione Viamarconidieci Advisory Board, Naples, Italy - *1995-present*
- Commissione Nazionale Post-Genoma, Italy - *2000-present*
- Member of Xeptagen Life Biotechnology Advisory Board - *2002 - present*
- Member of Academic Council of European School of Molecular Medicine (SEMM) *2002 - present*
- Member of the Genome Canada Panel for Applications in the Competition on Applied Genomics and Proteomics Research in Human Health – *2004 and 2005*
- Member of Advisory Board of the European Society of Gene Therapy *2004-present*
- Member of the Advisory Board of the INGM – Istituto Nazionale di Genetica Molecolare di Milano – *2004-present*
- Member of the Scientific Advisory Group of PRIME – Priorities for mouse functional genomic research in Europe – *2005*
- Member of the Scientific Advisory Board of the Fondazione Piemontese for Cancer Research (IRCC), Candiolo, Torino, Italy – *2005*
- Member of the Scientific Committee of the Fondazione Chiara D'Onofrio, Roma, Italy - *2005*
- Member of the Embo Panel for the review of the Spanish research agency (CSIC) - *2005*
- Member of the Mouse, Other Model Organisms and Development Scientific Review Committee for the Wellcome Trust Sanger Institute - *2005*
- Member of the Scientific Review Committee of the Wellcome Trust Sanger Institute Strategic Plan 2006-2011 - *2005*
- Member of the Scientific Committee of the Inter-University Consortium "Scuola per l'Alta Formazione" – *2006*
- Vice Chairman and Member of the ERC (European Research Council) Panel (LS2) for the evaluation of the Advanced Grants on Genetics, Genomics and System Biology - *2008*
- Member of the Strategic Committee for Innovative Clinical Research of the Heart Care Foundation – *2008*
- Member of the Diagnostics Scientific Committee of the International Rare Diseases Research Consortium (IRDiRC) – *2012-2016*
- Member of the Scientific Advisory Board of the National Niemann-Pick Disease Foundation (NNPDF) - Fort Atkinson, WI, USA - *2013-2015*
- Member of the Scientific Advisory Board of Shire Pharmaceutics *2014-2017*.
- Member of the Committee for Italian scholarships, Fondazione Armenise-Harvard – 2016-2018
- Member of the New York Academy of Sciences – 2014-
- Member of the Scientific Advisory Board, Institute of Genetics and Molecular and Cellular Biology (IGMCB), Strasbourg, FR – 2020-present
- Member of the Advisory Board of Next Generation Diagnostics, Pozzuoli, Naples, Italy. 2021-present
- Member of the Advisory Board of Avilar Therapeutics, Waltham, MA, USA. 2021-present
- Member of the Advisory Board of Coave Therapeutics, Paris, France. 2022-
- Member of the Scientific Advisory Board of Human Technopole – 2022- present

SCIENTIFIC AWARDS

1987 FISME (Italian Federation for the Study of Inherited Diseases) Award given by the ARIN (Italian Society for Research of Neurologic Interest).

1988 AIRH (Italian Society for Research, Prevention and Cure of Handicaps) Award.
1989 FISME (Italian Federation for the Study of Inherited Diseases) Award given by Socrea-Sirtori for the best communication in Molecular Cytogenetics.
1991 Basil O'Connor Starter Scholar Research Award, March of Dimes: "Mapping and cloning of disease genes from the distal short arm of the human X chromosome".
1996 Guglia di Napoli Award.
1996 Rotary Club Napoli Award.
2000 Golden Medal award by the Journal "Le Scienze".
2000 Guido Scocozza Solidarity National Award
2001 "Arycanda per la Cultura" National Award
2002 The International Association of Lions Clubs Award
2002 International Award "Sebetia-Ter" for Biomedical Sciences
2003 Vomero Award
2003 "Città di Salerno" Award for Scientific Research
2004 "Stella for Communication and Research" award by the journal "Mondosalute"
2005 Palizzi for Science Award
2005 Visionary Award on behalf of The Vision of Children Foundation for the outstanding commitment and dedication to Vision Research
2007 "L'Altra Italia - Vite da Premio" Award
2007 European Society of Human Genetics International Award
2007 Megaris Award
2007 "Masaniello" Award
2007 Giuseppe Moscati Award
2007 Marcello Torre National Award for Civic Engagement
2009 Giovan Battista Basile Special Award
2009 Napoletani Eccellenti nel Mondo Award
2009 Stella di Tabor Award for Science, Research and Scientific Communication
2010 Capo D'Orlando Award
2010 PLEI Prestigio Professionale Award
2014 Alumni IPE (Institute for research and educational activities) Award
2014 Captains of the Year Award - Captains of Research
2014 Civitas Award
2016 Louis-Jeantet Prize for Medicine
2017 Lions International-Award for Excellence 2016-2017
2018 "Giusti nel mondo" Award
2019 Tommaso e Laura Leonetti Award
2021 Antonio Feltrinelli 2021 Prize for Biological Sciences and Applications, awarded by the Accademia dei Lincei

SCIENTIFIC SOCIETIES

- Academy of Medicine of Turin, Italy
- American Society of Human Genetics
- Biochemical Society
- European Molecular Biology Organization (EMBO)
- European Society of Human Genetics (ESHG)
- Human Genome Organization (HUGO)
- Italian Society of Medical Genetics

- Italian Society of Molecular Medicine
- Italian Society for the Study of Inborn Errors of Metabolism
- "L'Altra Napoli" Society

EDITORIAL BOARDS

Past

- American Journal of Medical Genetics: *Neuropsychiatric Genetics (Field Editor), Wiley-Liss, New York, N.Y.*
- Applied Bioinformatics, *Open Mind Journals, New Zealand*
- BMC Medical Genetics, *BioMed Central Ltd, London, UK*
- Clinical Genetics, *Munksgaard International,Copenhagen, Denmark. 2000-2005*
- Current Opinion in Genetics & Development, *Elsevier Science, London, U.K*
- EMBO Journal - European Molecular Biology Organization, *Embo Press, Germany*
- EMBO Reports, European Molecular Biology Organization, *Embo Press, Germany*
- Encyclopedia of the Human Genome, *Macmillan Reference Ltd., London, U.K.*
- European Journal of Human Genetics, *S. Karger, Basel, Switzerland*
- Genome Research, *Cold Spring Harbor Laboratory Press, New York, N.Y.*
- Human Molecular Genetics, *Oxford Academic, Oxford U.K. 1992-2001*
- Human Genetics, *Springer-Verlag International, Heidelberg, Germany*
- Molecular Genetics and Metabolism, *Academic Press, San Diego, CA until 2002*
- Mutation Research Genomics, *Elsevier Science, Amsterdam, The Netherlands*
- PathoGenetics (Chief Editor), *BioMed Central*
- Annual Review of Genomics and Human Genetics – *Annual Reviews, Palo Alto, CA*

Current

- Cell Stress, Shared Science Publishers OG, Graz, Austria
- EMBO Molecular Medicine, *Embo Press, Germany*
- The Online Metabolic and Molecular Bases of Inherited Disease – OMMBID, *The McGraw-Hill Companies, NY, USA from 2008 – present*
- eLife, *Castle Park, Cambridge, UK, 2012-present*

REVIEWER

Serves as a reviewer for: Science, Nature, Cell, American Journal of Human Genetics, American Journal of Medical Genetics, Clinical Genetics, European Journal of Human Genetics, Genetics, Genome Research, Genomics, Human Genetics, Human Molecular Genetics, Human Mutation, Mammalian Genome, Molecular Medicine Today, Nature Genetics, New England Journal of Medicine, Nucleic Acids Research, PCR Methods and Applications.

PATENTS

U.S. Patent Application No.10/775,678 and PCT Application PCT/US04/03632
Entitled: "Diagnosis and Treatment of Multiple Sulfatase Deficiency and Other Sulfatase Deficiency"
Filing date: February 10, 2004

Inventors: von Figura K, Schmidt B, Dierks T, Heartlein MW, ***Ballabio A***, Cosma MP

U.S. Patent Application No. 11/416453.
Entitled: "Methods and compositions for recovering or improving visual function".
Filing date: March 5, 2006
Inventors: Auricchio A, Surace EM, ***Ballabio A***.

European Patent Application No. 09152778
Entitled: "Molecules able to modulate the expression of at least a gene involved in degradative pathways and use thereof"
Filing date: February 13, 2009
Inventors: ***Ballabio A,*** Sardiello M

International Patent Application No. PCT/IB2010/056024 - WO2012/085622
Entitled: "Therapeutic strategies to treat CNS pathology in Mucopolysaccharidoses"
Filing date: 22 December, 2010
Inventors: Fraldi A, ***Ballabio A***

US Provisional Application N. 61/449,751 - WO2012120044
Entitled: "TFEB variants and uses thereof"
Filing date: 07 March 2011
Inventors: ***Ballabio A***, Settembre C, Medina Sanabria DL

US Provisioanl Application N. 61/579,793 23 - WO2012120048
Entitled: "TFEB phosphorylation inhibitors and uses thereof"
Filing date: December 2011
Inventors: ***Ballabio A***, Settembre C, Medina Sanabria DL

US Provisional Application N. 61/660,194 - WO2013186398
Entitled: "Use of TFEB for treating obesity"
Filing date: 15 June 2012
Inventors: ***Ballabio A***, Settembre C

European Patent Application N. 12164785.3 - WO2013156325
Entitled: "TFEB gene therapy of alpha-1-antitrypsin deficiency"
Filing date: 19 April 2012
Inventors: ***Ballabio A***, Brunetti-Pierri N, Pastore N

FUNDINGS

On-going

Funding Agency: Department of Defense U.S. Army Medical Research and Development Command
P.I. Ballabio A
Title: Targeting TFEB and TFE3 in Renal Tumorigenesis
Funding period: 07/2022-06/2025 Amount: 412.500$

Funding Agency: Associazione Italiana per la Ricerca sul Cancro (AIRC) (Italian Association for Cancer Research)
P.I. Ballabio A

Title: Metastatic Cancer of Unknown Primary (CUP): a biological enigma and an unmet medical need
Funding period: 07/2018-01/2025 Amount: 2.770.000€

Funding Agency: Associazione Italiana per la Ricerca sul Cancro (AIRC) (Italian Association for Cancer Research) -
P.I. Ballabio A
Title: Targeting RagD to inhibit mTORC1 signaling in cancer
Funding period: 02/2019-01/2024 Amount: 936.000€

Completed (last 10 years)

Funding Agency: European Research Council
P.I. Ballabio A
Title: Lysosomics - Functional genomics of the lysosome
Funding period: 10/2016-09/2021 Amount: 2.362.562 EURO

Funding Agency: The Huffington Foundation
P.I. Ballabio A
Title: NRI Parkinson's disease project
Funding period: 01/2017-12/2021 Amount: $482,288 USD

Funding Agency: Regione Campania
P.I. Ballabio A
Title: From genomics to therapy of rare tumors
Funding period: 01/2018-12/2021 Amount: 11.800.000€

Funding Agency: National Institutes of Health (NIH)
P.I. Ballabio A
Title: Modulation of cellular clearance to treat human disease
Funding period: 09/2012-06/2021 Amount: $1,958,907 USD

Funding Agency: Beyond Batten Disease Foundation
P.I. Ballabio A
Title: New therapeutic strategies for Batten disease
Funding period: 08/2014-12/2017 Amount: 875,000USD

Funding Agency: National Institute of Health R01 NS078072-01A1
P.I. Ballabio A
Title: Modulation of cellular clearance to treat human disease
Funding period: 10/2011-11/2016 Amount: 1,250,000 USD

Funding Agency: Huffington Foundation
P.I. Ballabio A
Title: NRI Parkinson disease project
Funding Period: 01/01/2012-12/31/2016. Amount: $500,000 USD

Funding Agency: Center for Orphan Disease Research and Therapies
P.I. Ballabio A
Title: Modulation of cellular clearance to treat mucopolysaccharidoses type 1

Funding period: 01/2014-12/2016 Amount: 300,000USD

Funding Agency: Fondazione Telethon
P.I. Ballabio A
Title: Modulation of cellular clearance in lysosomal storage disease
Funding period: 07/2011-06/2015 Amount: 500.000,00€

Funding Agency: European Research Council
P.I. Ballabio A
Title: Modulating cellular clearance to cure human disease
Funding period: 03/2010-02/2015 Amount: 2,300.000,00€

Funding Agency: Team Sanfilippo Foundation
P.I. Ballabio A
Title: Modulation of cellular clearance to treat mucopolysaccharidosis
Funding period: 01/2012-12/2014 Amount: 75.000,00USD

Funding Agency: San Filippo Children's Research Foundation
P.I. Ballabio A
Title: Modulation of cellular clearance to clear mucopolysaccharidosis
Funding period: 01/2012-12/2014 Amount: 160.000,00CAD

PUBLICATIONS

Manuscripts
(375 publications in peer-reviewed journals)

1) ***Ballabio A***, Pallini R, Di Natale P.
Mucopolysaccharidosis III B: hybridization studies on fibroblasts from a mild case and fibroblasts from severe patients.
Clin Genet, 25:191-195. 1984.
2) Andria G, ***Ballabio A***, Parenti G, Di Maio S, Piccirillo A.
Steroid sulphatase deficiency is present in patients with the syndrome 'Ichthyosis and male hypogonadism' and with 'Rud syndrome'.
J Inher Metab Dis, 7:Suppl. 2, 159-160. 1984.
3) Andria G, ***Ballabio A***, Parenti G, Di Maio S, Piccirillo A.
Steroid sulphatase deficiency and hypogonadism.
Eur J Pediatr, 142:304-305. 1984.
4) ***Ballabio A***, Parenti G, Napolitano E, Di Natale P, Andria G.
Genetic complementation of steroid sulphatase after somatic cell hybridization of X linked ichthyosis and multiple sulphatase deficiency.
Hum Genet, 70:315-317. 1985.
5) Parenti G, ***Ballabio A***, Napolitano E, Di Natale P, Andria G.
Hybridization studies of steroid sulphatase on fibroblasts from patients affected by X linked ichthyosis with and without hypogonadism.
Persp Inher Metab Dis, 6:125-130. 1985.
6) Strisciuglio P, ***Ballabio A***, Parenti G.
Microtia with mental atresia and conductive deafness: mild and severe manifestations within the same sibship.
J Med Genet, 23:459-460. 1986.
7) ***Ballabio A***, Parenti G, Tippett P, Mondello C, Di Maio S, Tenore A, Andria G.

X-linked ichthyosis, due to steroid sulphatase deficiency, associated with Kallmann syndrome (hypogonadotropic hypogonadism and anosmia): linkage relationships with Xg and cloned DNA sequences from the distal short arm of the X chromosome.
Hum Genet, 72:237-240. 1986.

8) Chase DS, Morris AH, ***Ballabio A***, Pepper S, Giannelli F, Adinolfi M.
Genetics of Hunter syndrome: carrier detection, new mutations, segregation and linkage analysis.
Ann Hum Genet, 50:349-360. 1986.
9) Sebastio G, Hunziker W, ***Ballabio A***, Auricchio S, Semenza G.
On the primary site of control in the spontaneous development of small-intestinal sucrase-isomaltase after birth.
Febs Lett, 208:460-464. 1986.
10) Andria G, ***Ballabio A***, Parenti G.
X-linked ichthyosis due to steroid sulphatase deficiency, associated with hypogonadism and anosmia.
Ann Neurol, 22:98. 1987.
11) ***Ballabio A***, Carrozzo R, Persico MG, Andria G.
Studies on the molecular basis of steroid sulphatase deficiency in man.
Protides of the Biol Fluids, 35:41-44. 1987.
12) Parenti G, ***Ballabio A***, Hoogeveen AT, van der Loos CM, Jobsis AC, Andria G.
Studies on cross-reacting material to steroid sulphatase in fibroblasts from patients affected by different types of steroid sulphatase deficiency.
J Inher Metab Dis, 10:224-228. 1987.
13) ***Ballabio A***, Parenti G, Carrozzo R, Sebastio G, Andria G, Buckle V, Fraser N, Craig I, Rocchi M, Romeo G, Jobsis AC, Persico MG.
Isolation and characterization of a steroid sulfatase cDNA clone: genomic deletions in patients with X-chromosome-linked ichthyosis.
Proc Natl Acad Sci USA, 84:4519-4523. 1987.
14) ***Ballabio A***, Sebastio G, Carrozzo R, Parenti G, Piccirillo A, Persico MG, Andria G.
Deletions of the steroid sulphatase gene in "classical" X-linked ichthyosis and in X linked ichthyosis associated with Kallmann syndrome.
Hum Genet, 77:338-341. 1987.
15) Fraser N, ***Ballabio A***, Zollo M, Persico G, Craig I.
Identification of incomplete coding sequences for steroid sulphatase on the human Y chromosome: evidence for an ancestral pseudoautosomal gene?
Development, 101:Suppl., 127-132. 1987.
16) ***Ballabio A***, Parenti G, Carrozzo R, Coppa G, Felici L, Migliori V, Silengo M, Franceschini P, Andria G.
X/Y translocation in a family with X-linked ichthyosis, chondrodysplasia punctata, and mental retardation: DNA analysis reveals deletion of the steroid sulphatase gene and translocation of its Y pseudogene.
Clin Genet, 34:31-37. 1988.
17) Piccirillo A, Auricchio L, Fabbrocini G, Parenti G, ***Ballabio A***, Delfino M.
Ocular findings and skin histology in a group of patients with X-linked ichthyosis.
Br J Dermatol, 119:185-188. 1988.
18) ***Ballabio A***, Carrozzo R, Parenti G, Gil A, Zollo M, Persico MG, Gillard E, Affara N, Yates J, Ferguson-Smith MA, Frants RR, Eriksson AW, Andria G.
Molecular heterogeneity of steroid sulfatase deficiency: A multicenter study on 57 unrelated patients, at DNA and protein levels.
Genomics, 4:36-40. 1989.
19) ***Ballabio A***., Carrozzo R., Gil A., Gillard B., Affara N., Ferguson-Smith M.A., Fraser N., Craig I., Rocchi M., Romeo G., Andria G.
Molecular characterization of human X/Y translocations suggests their aetiology through aberrant exchange between homologous sequences on Xp and Yq.
Ann Hum Genet, 53:9-14. 1989.

20) ***Ballabio A***., Bardoni B., Carrozzo R., Andria G., Bick D., Campbell L., Hamel B., Ferguson-Smith M.A., Gimelli G., Fraccaro M., Maraschio P., Zuffardi O., Guioli S., Camerino G.
Contiguous gene syndromes due to deletions in the distal short arm of the human X chromosome.
Proc Natl Acad Sci USA, 86:10001-10005. 1989.
21) ***Ballabio A***, Gibbs RA, Caskey CT.
PCR test for cystic fibrosis deletion.
Nature, 343:220. 1990.
22) Ross MT, ***Ballabio A***, Craig IW.
Long-range physical mapping around the human steroid sulfatase locus.
Genomics, 6:528-539, 1990.
23) ***Ballabio A***, Ranier JE, Chamberlain JS, Zollo M, Caskey CT.
Screening for steroid sulfatase (STS) gene deletions by multiplex DNA amplification.
Hum Genet, 84:571-573. 1990.
24) Meitinger T, Heye B, Petit C, Levilliers J, Golla A, Moraine C, Dalla Piccola B, Sippell WG, Murken J, ***Ballabio A***.
Definitive localization of X-linked Kallmann syndrome (hypogonadotropic hypogonadism and anosmia) to Xp22.3: close linkage to the hypervariable repeat sequence CRI-S232.
Am J Hum Genet, 47:664-669. 1990.
25) ***Ballabio A***, Bardoni B, Guioli S, Basler E, Camerino G.
Two families of low-copy-number repeats are interspersed on Xp22.3: implications for the high frequency of deletions in this region.
Genomics, 8:263-270. 1990.
26) Bick D and ***Ballabio A***.
Chromosome abnormalities in and pathogenesis of Kallmann syndrome.
Am J Med Genet, 37:298. 1990.
27) ***Ballabio A***.
Contiguous deletion syndromes.
Curr Opin Genet Devel, 1:25-29. 1991.
28) Brown CJ, ***Ballabio A***, Rupert JL, Lafreniere RG, Grompe M, Tonlorenzi R, Willard HF.
A gene from the region of the human X inactivation centre is expressed exclusively from the inactive X chromosome.
Nature, 349:38-44. 1991.
29) Brown CJ, Lafreniere RG, Powers VE, Sebastio G, ***Ballabio A***, Pettigrew AL, Ledbetter DH, Levy E, Craig IW, Willard HF.
Localization of the X inactivation centre on the human X chromosome in Xq13.
Nature, 349:82-84. 1991.
30) Borsani G, Tonlorenzi R, Simmler MC, Dandolo L, Arnaud D, Capra V, Grompe M, Pizzuti A, Muzny D, Lawrence C, Willard HF, Avner P, ***Ballabio A***.
Characterization of a murine gene expressed from the inactive X chromosome.
Nature, 351:325-329. 1991
31) Pieretti M, Tonlorenzi R, ***Ballabio A***.
Rapid assembly of l phage contigs within YAC clones.
Nucl Acids Res, 19:2795-2796. 1991.
32) Nelson DL, ***Ballabio A***, Victoria MF, Pieretti M, Bies RD, Gibbs RA, Maley JA, Chinault AC, Webster TD, Caskey CT.
Alu-primed polymerase chain reaction for regional assignment of 110 yeast artificial chromosome clones from the human X chromosome: identification of clones associated with a disease locus.
Proc Natl Acad Sci USA, 88:6157-6161. 1991.
33) Bardoni B, Zuffardi O, Guioli S, ***Ballabio A***, Simi P, Cavalli P, Grimoldi MG, Fraccaro M, Camerino G.

A deletion map of the human Yq11 region: implications for the evolution of the Y chromosome and tentative mapping of a locus involved in spermatogenesis.
Genomics, 11:443-451. 1991.

34) Franco B, Guioli S, Pragliola A, Incerti B, Bardoni B, Tonlorenzi R, Carrozzo R, Maestrini E, Pieretti M, Taillon-Miller P, Brown CJ, Willard HF, Lawrence C, Persico MG, Camerino G, ***Ballabio A***.
A gene deleted in Kallmann's syndrome shares homology with neural cell adhesion and axonal path-finding molecules.
Nature, 353:529-536. 1991.

35) ***Ballabio A***, Zollo, M, Carrozzo R, Caiulo A, Zuffardi O, Cascioli CF, Viggiano D, Strisciuglio P.
Deletion of the distal short arm of the X chromosome (Xp) in a patient with short stature, chondrodysplasia punctata and X-linked ichthyosis due to steroid sulfatase deficiency.
Am J Med Genet, 41:184-187. 1991.

36) ***Ballabio A,*** Camerino G.
A gene for X-linked Kallmann syndrome: a human neuronal migration defect.
Curr Opin Genet Devel, 2:417-421. 1992.

37) ***Ballabio A,*** Willard HF.
Mammalian X-chromosome inactivation and the XIST gene.
Curr Opin Genet Devel, 2:439-447. 1992.

38) Carrozzo R, Ellison J, Yen P, Taillon-Miller P, Brownstein BH, Persico G, ***Ballabio A***, Shapiro L.
Isolation and characterization of a yeast artificial chromosome (YAC) contig around the human steroid sulfatase gene.
Genomics, 12:7-12. 1992.

39) Palmieri G, Capra V, Romano G, D'Urso M, Johnson S, Schlessinger D, Morris P, Hopwood J, Di Natale P, Gatti R, ***Ballabio A***.
The iduronate sulfatase gene: isolation of a 1.2-Mb YAC contig spanning the entire gene and identification of heterogeneous deletions in patients with Hunter syndrome.
Genomics, 12:52-57. 1992.

40) Grompe M, Pieretti M, Caskey CT, ***Ballabio A***.
The sulfatase gene family: cross-species PCR cloning using the MOPAC technique.
Genomics, 12:755-760. 1992.

41) Basler E, Grompe M, Parenti G, Yates, J, ***Ballabio A***.
Identification of point mutations in the steroid sulfatase gene of three patients with X linked ichthyosis.
Am J Hum Genet, 50:483-491. 1992.

42) Wapenaar MC, Petit C, Basler E, ***Ballabio A***, Henke A, Rappold GA, van Paassen HMB, Blonden LAJ, van Ommen GJB.
Physical mapping of 14 new DNA markers isolated from the human distal Xp region.
Genomics, 13:167-175. 1992.

43) Bick D, Franco B, Sherins RJ, Heye B, Pike L, Crawford J, Maddalena A, Incerti B, Pragliola A, Meitinger T, ***Ballabio A***.
Brief report: Intragenic deletion of the *Kalig-1* gene in Kallmann's syndrome.
New Engl J Med, 326:1752-1755. 1992.

44) Bernatowicz LF, Li X-M, Carrozzo R, ***Ballabio A***, Mohandas T, Yen PH, Shapiro L.
Sequence analysis of a partial deletion of the human steroid sulfatase gene reveals 3 bp of homology at deletion breakpoints.
Genomics, 13: 892-893, 1992.

45) ***Ballabio A*** and Andria G.

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