

Curriculum Vitae

Name: Anthony P. Monaco

Position President, Tufts University

Visiting Professor of Genetics of Speech and Language Disorders, Wellcome Trust Centre for Human Genetics
University of Oxford

Address: Office of the President
Ballou Hall
Tufts University
Medford, MA 02155
USA
telephone: 1 617 627 3300
telefax: 1 617 627 3555
email: anthony.monaco@tufts.edu
<http://president.tufts.edu>

Wellcome Trust Centre for Human Genetics
University of Oxford
Roosevelt Drive
Headington, Oxford OX3 7BN
United Kingdom
telephone 44 1865 287502/503
telefax 44 1865 287650
e-mail anthony.monaco@well.ox.ac.uk
url: <http://www.well.ox.ac.uk/anthony-monaco-2>
<http://www.researcherid.com/rid/A-4495-2010>
http://www.researchgate.net/profile/Anthony_Monaco/
<http://scholar.google.co.uk/citations?user=rsUZ2eIAAAA&hl=en>

Home Address: Gifford House
161 Packard Avenue
Medford, MA 02155
USA

Date of Birth: October 10, 1959

Place of Birth: Wilmington, Delaware, USA

Citizenship: United States of America

Marital Status: Married, with three sons

Education:
1981 A.B. Princeton University, Princeton NJ USA

1987	Ph.D.	Independent Concentration in Neuroscience Neurobiology, The Program in Neuroscience Harvard University, Cambridge, MA, USA
1988	M.D.	Harvard Medical School, Boston, MA USA Medical Scientist Training Program

Postdoctoral Training:

1988-89	Harvard Medical School Moseley Travelling Fellow at Imperial Cancer Research Fund, London, UK
1989-90	Muscular Dystrophy Association Neuromuscular Disease Research Fellowship at ICRF, London, UK

Awards and Honors:

1985	American Society of Human Genetics Predoctoral Basic Science Award
1986	American Society of Human Genetics Travel Award for International Congress of Human Genetics, Berlin
1993	Gaetano Conte Prize for Basic Research on Muscular Diseases
1999	Fellow, Academy of Medical Sciences, UK
2002	Jacob's Ladder Lectureship and Award, The Canadian Foundation for the Control of Neurodegenerative Disorders
2003	Professorial Fellow, Merton College, Oxford
2003	Salesianum School Hall of Fame, Science
2006	Elected member, European Molecular Biology Organization (EMBO)
2006	Fellow of the Royal Society of Medicine, UK
2008	Selected as Highly Cited Researcher in Highly Cited.com
2010	Outstanding Physician Scientist Award in Neurological Research, European Genomics & Neurodegenerative Diseases
2013	Elected member, Association of American Physicians

Teaching Experience:

1982	Instructor in Neurobiology for the Harvard Health Professions Program Summer School
1988-1992, 1995	Instructor in European School of Medical Genetics, Sestri Levante, Italy
1988-1998	Instructor in Wellcome Trust Summer School "DNA Related Methods in Human Genetics"
1993	Instructor in Human Genetics (two week course) for Honours Biochemistry Students, The National University of Singapore
1995	Lecturer, First Advanced Course on Gene Therapy, Milan, Italy

1995	Lecturer, ISREC postgraduate student course, Geneva
1997	Lecturer to postgraduate students, Tunis, Tunisia
1997	Lecturer, course on Molecular Neurogenetics, Porto, Portugal
1997	Lecturer, Second Advanced Course on Gene Therapy, Venice, Italy
1998-2001	Lecturer for "Genetics and disease option", Department of Biochemistry, University of Oxford
1998-2007	Tutor for Stanford in Oxford programme, eight tutorials per student with written essays
1999-2000	Lecturer for "Methods and Techniques Course for DPhil Students", Nuffield Department of Medicine
2002-2007	Lecturer for MSc course in Neuroscience, University of Oxford
2003-2011	Lecturer in Human Genetics for the FHS Human Sciences undergraduates, University of Oxford
2012, 2013	Lecturer in Experimental College, Tufts University "A Taste of Tufts: A Sampling of Faculty Research."

Membership of professional and learned societies:

1981-	Sigma Xi Research Society, Phi Beta Kappa Society
1984-	Member, American Society of Human Genetics
1991-	Member, European Society of Human Genetics
1992-	Member, The Human Genome Organization
1995-2001	Foundation Member, World Muscle Society
1995-	Member, German Society of Neurogenetics
1995-	Charter Fellow, Molecular Medicine Society
1996-	Member, Autism Society of America
1996-	Member, The National Autistic Society (UK)
1999-	Fellow, Academy of Medical Sciences (UK)
2006-	Member, European Molecular Biology Organization (EMBO)
2006-	Fellow, Royal Society of Medicine
2013-	Member, Association of American Physicians

Other professional activities:

Editorial Boards

1989-2005	Editorial Board, Acta Myologica
1992-2011	Associate Editor, European Journal of Human Genetics
1992-2001	Editorial Board, Human Molecular Genetics
1994-2001	Editorial Board, Neuromuscular Disorders
1996-present	Editorial Board, Neurogenetics
1997-2013	Associate Editor, Genomics
1998-2004	Editorial Board, Brain
1998-2000	Advisory Board, Journal of Neural Transmission

2002-2005	Editorial Board, NeuroMolecular Medicine
2004-2010	Editorial Committee, Annual Review of Genomics and Human Genetics
2008-2010	Editorial Board, The American Journal of Human Genetics
2008-2012	Editorial Board, Journal of Neurodevelopmental Disorders
2008-present	Advisory Board, EMBO Molecular Medicine
2008-present	Editorial Board, European Psychiatric Review
2008-present	Associate Editor, Journal of Molecular Medicine
2009-present	Editorial Board, Molecular Autism

Scientific and Professional Committees

1989-1997	Editor, Human X Chromosome Committee Human Gene Mapping
1991-1993	External Advisory Committee, Baylor College of Medicine, Human Genome Center
1992	NIH Genome Center Review Committee for Human Chromosome 13, Columbia University
1993-1999	Scientific Committee, Telethon Foundation Italy
1994-1997	External Advisory Committee, NIH Program Project "Identification and Transgenic Studies of ALS Genes"
1994	Medical Research Council, HGMP Research Development Group
1994-1997	Medical Research Council, Cell and Molecular Board Grants Committee B
1995-1998	Genome Data Base, Quarterly Review Committee
1995-1998	Medical Research Council, Links with Industry Panel
1995-1998	External Advisory Committee, NIH Program Project "Neurogenetics"
1997-2010	Scientific Advisory Group, Autism Research Centre, University of Cambridge
1997-2001	HUGO Human Genome Mapping Committee
1998	International Evaluation Committee, Max-Delbrück Center for Molecular Medicine, Berlin
1998-1999	Member, Infrastructure Panel, The Wellcome Trust
1999-2001	Member, Biological Sciences Panel, Research and Assessment Exercise 2001
2001-2004	Member, Neurosciences Panel, The Wellcome Trust
2002, 2004-06, 2008	Member and Chair, Multidisciplinary International Peer Review Panel, Genome Canada
2003	Member, External Scientific Evaluation Committee, Molecular Cardiovascular Medicine Group, University of Leuven, Belgium
2003	Member, Review Panel, Genome España, Madrid
2004	Member, Site Visit Group, Nancy Lurie Marks Family Foundation, Boston, MA, USA
2004-2011	Member, External Advisory Committee, NIDA training grant, Departments of Psychiatry and Genetics,

	Virginia Commonwealth University VIPBG, Richmond, VA, USA
2005	Chair, Evaluation Committee, Interuniversity Attraction Poles, Belgian Science Policy Office
2005-2009	College of Experts, Medical Research Council, UK
2006	Expert Committee, Canada Foundation for Innovation, Toronto, Canada
2006-2007	Member, Science Funding Committee, Cancer Research UK
2006-2007	Member, Scientific Content and Advisory Panel for Generation Genome, a Wellcome Trust funded traveling museum exhibition celebrating the human genome and genetics.
2006-2007	Member, Advisory Panel for 'Choose your Character', an interactive traveling genetics exhibition
2007 & 2011	Review Panel and Site Visit, Genome Technology Branch, National Human Genome Research Institute, National Institutes of Health, USA
2007-present	Member, External Advisory Board, Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy
2008	Chairman, Quinquennial Review of the CRUK groups at the Strangeways Research Laboratories, Cambridge
2008-2011	Governor Member's Council, Oxfordshire & Buckinghamshire Mental Health Partnership NHS Foundation Trust
2008	Member, Review Panel, Genome España, Madrid
2008-2011	Member, TRAC Development Group, HEFCE
2010-2011	President, Advisory Scientific Committee, Centre for Biomedical Network Research in the Area of Rare Diseases, Valencia, Spain
2011-present	Trustee, Tufts University, Medford MA
2011-present	Director, Tufts Medical Center, Boston, MA
2011-2015	Member, NIH National Advisory Council for Human Genome Research
2011-present	Trustee, MacJannet Foundation, Wellesley Hills MA
2011-present	Trustee, Cummings Foundation, Woburn MA
2013-present	Elected member, Association of American Physicians
2013-present	Trustee, Salesianum School Board, Wilmington DE
2013-2014	Member, Steering Committee, Talloires Network
2015-present	Chair, Steering Committee, Talloires Network
2014-2017	Member, Executive Committee, NESAC
2015-2016	Chair, Executive Committee, NESAC
2015-present	Member, Business-Higher Education Forum
2016-2017	Chair, Board of Association of Independent Colleges and Universities in Massachusetts (AICUM), Inc.
2014-present	IMAGINE Scientific Advisory Board, Paris, France
2016-present	Member, WGBH Board of Trustees

Scientific Advisory Boards/Consultancies:

1993-1997	Scientific Advisory Board, Sequana Therapeutics Inc., La Jolla, CA, USA
-----------	---

1996-1999	Scientific Advisory Board, Hexagen Limited, Cambridge, UK, now Incyte Genomics Inc.
1997-2002	Science and Technology Advisory Board, Oxagen Limited, Abingdon UK
2003-present	Scientific Consultant, Oxagen Limited, Abingdon UK
1998-2003	Scientific Advisory Board, DNA Sciences Inc., Mountain View, CA, USA
2000-2002	Advisory Panel for Molecular Biology, Oxford University Press, Oxford, UK
2001-2006	Consultant, Triaj Inc., Brecksville, Ohio, USA
2005-present	Member, Gerson Lehrman Group Healthcare Council Scholars and Educators Program
2007-2008	Consultant, GlaxoSmithKline
2009-2013	Consultant, Wiley Rein LLP, Washington DC, USA
2009-2013	Scientific Advisory Board, Bio.logis, Frankfurt, Germany

University Committees:

1998-2007	Director of Wellcome Trust Centre for Human Genetics Chair, Management Committee, Group Heads Meeting, Information Technology Committee, Functional Genetics Facility Users Group, Health and Safety Committee
1998- 2007	Member, Division of Medical Sciences Board, Research Strategy and Implementation Group, University of Oxford
2005-2007	Member, RAE Preparation Group, Division of Medical Sciences, University of Oxford
2005-2011	Member, Merton College Oxford Governing Body and College Stipends Committee
2007-2011	Pro-Vice-Chancellor, Planning and Resources, University of Oxford Chair, Planning and Resource Allocation Committee Chair, Strategic Plan Review and Implementation Group Chair, Capital Steering Group Chair, Student Numbers Planning Sub-Committee Chair, Budget Sub-Committee Chair, FEC Senior Strategy Group Chair, Radcliffe Observatory Quarter Project Board Chair, Science Area Masterplan Project Board Chair, Sustainability Steering Committee Chair, Risk Management Steering Committee Chair, University Trusts Management Board Chair, Joint Student Teaching and Funding Support Group Chair, Fees Policy Group Chair, Somerville Project Sponsor Group

Chair, ROQ Infrastructure Project Sponsor Group
 Chair, Joint Panel on College Associations
 Chair, Services' Funding Working Group: Museums and University Collections
 Member, University Council
 Member, Energy Purchasing Task Group
 Member, Development Committee
 Member, John Fell OUP Research Fund Committee
 Member, Joint Resource Allocation Advisory Board
 Member, Buildings and Estates Sub-Committee
 Member, Science Area Infrastructure Project Sponsor Group
 Member, Health and Safety Management Committee
 Member, Research Staff Working Group
 Member, Humanities Division Review Panel
 Member, Social Sciences Division Review Panel
 Member, Strategic Plan Steering Group
 Member, Financial Processes Working Group of Council
 In attendance, General Purposes Committee
 In attendance, Finance Committee
 In attendance, Estates Bursars' Committee

2011-

President, Tufts University
 Member, Board of Trustees
 Member, Executive Committee of the Board of Trustees
 Member, standing committees of the Board of Trustees (Academic Affairs, Administration and Finance, Compensation Honorary Degree, Trusteeship, and University Advancement)
 President of the Faculties
 Chair, Academic Council
 Chair, President's Council on Diversity
 Chair, President's Council on Campus Sustainability
 Member, T10 Strategic Plan Steering and Executive Committees
 Member, TEAM Executive Committee
 Member, Executive Capital Committee
 Member, Executive Budget Committee
 Chair, President's Task Force on Sexual Misconduct Prevention
 Chair, Diversity and Inclusion Leadership Council
 Co-chair, Mental Health Task Force

Research Training and Appointments:

1978-1981

Summer and academic year research assistant
 Supervisor: Bartley G. Hoebel, PhD
 Psychology Department, Princeton University Research Areas:
 Neuropharmacology and physiology of feeding behavior;
 animal models of psychoactive drug self-administration

1981-1983

Summer and academic year medical school research.

	Supervisor: J. Allen Hobson, MD, Neurophysiology Laboratory, Department of Psychiatry, Harvard Medical School, Boston Research Areas: Neuropharmacology, physiology and anatomy of brainstem-visual cortex interactions during REM sleep
1984-1988	Doctoral research, Supervisor: Louis M. Kunkel, PhD, Division of Genetics, Department of Pediatrics, Children's Hospital and Harvard Medical School Research Areas: Molecular genetics of Duchenne and Becker muscular dystrophy
1988-1990	Postdoctoral research, Supervisor: Hans Lehrach, PhD, Genome Analysis Laboratory, Imperial Cancer Research Fund, London, UK Research Areas: Molecular genetics of the human X chromosome. Yeast artificial chromosome and cosmid libraries, physical mapping
1990-1995	Senior Scientist, Head of Human Genetics Laboratory, Imperial Cancer Research Fund, Institute of Molecular Medicine, John Radcliffe Hospital, Oxford
1994-1997	University Research Lecturer, Faculty of Clinical Medicine, University of Oxford Research areas: 199
1995-2007	Wellcome Principal Research Fellow, Wellcome Trust Centre for Human Genetics, University of Oxford Research Areas: Genetics of neurodevelopmental disorders including autism, specific language impairment and dyslexia; Positional cloning and functional analysis of monogenic and polygenic disease genes
1997-2011	Professor of Human Genetics, Faculty of Clinical Medicine, University of Oxford
1998-2007	Director, Wellcome Trust Centre for Human Genetics, University of Oxford
2007-2011	Pro-Vice-Chancellor (Planning and Resources) University of Oxford
2011-	President, Tufts University, Medford, MA, USA and Professor of Biology and Neuroscience, Tufts University
2011-2017	Visiting Professor in the Genetics of Speech and Language Disorders, University of Oxford

Current Grant Support:

Grant Type: Wellcome Trust Programme Grant

Grant Holder: AP Monaco, Z Molnar and A Velayos-Baeza

Grant Number: 092071

Title: The role of the KIAA0319 protein in neurodevelopment

Amount Awarded: £1,393,036

Dates: 1/10/10 - 31/12/16

Recently Completed Grant Support:

Grant Type: Medical Research Council Strategic Award
Grant Holders: AP Monaco and A Bailey and Autism Genome Project Consortium
Grant number: G060130 ID;79040
Title: Autism Genome Project
Amount awarded: £764,860 Dates: 23/04/07 – 30/09/10

Awarding Body: NLM Family Foundation and Simon's Foundation
Grant Holders: AP Monaco and A Bailey
Title: Identifying and understanding the actions of autism susceptibility genes
Amount awarded: £1,291,807 Dates: 1/10/06 – 30/09/10

Awarding Body: Medical Research Council
Grant Holders: S Paracchini, DF Newbury, AP Monaco, CD Steer, P Bolton, J Golding
Grant Number: G0800523
Title: Gene x gene and gene x environment interactions underlying speech, language and reading development
Amount awarded: £169,775 Dates: 1/10/08 – 30/09/10

Grant Type: Wellcome Trust Programme grant
Grant Holder: AP Monaco
Grant Number: 076566
Title: Genetics of specific reading and language disorders
Amount Awarded: £3,367,319 Dates: 1/10/05 – 30/09/10

Grant Type: Wellcome Trust Core Grant
Grant Holder: A P Monaco (no longer grant holder since May 2007)
Grant Number: ME030603 and 075491
Title: Mapping and characterization of susceptibility genes in multifactorial diseases
Amount Awarded: £10,016,270 Dates: 1/4/05 – 31/03/10

Grant Type: Wellcome Trust Equipment Grant
Grant Holders: R Mott, AP Monaco, DI Stuart, MI McCarthy, H Watkins, AVS Hill, D Kwiatkowski, J Flint
Grant Number: 079981/Z/06/Z
Title: Large scale data storage and backup strategy for the Wellcome Trust Centre for Human Genetics for the period 2006-2010
Amount Awarded: £195,129 Dates: 01/07/06 – 30/06/10

Awarding Body: FP6 EU dyslexia grant NEURODYS
Grant Holders: AP Monaco and L Cardon
Title: Dyslexia genes and neurobiological pathways
Amount awarded: £286,548 Dates: 1/07/06 – 31/03/10

Awarding Body: FP6 EU autism grant MOLGEN
Grant Holders: AP Monaco and A Bailey

Title: Using European and International populations to identify autism susceptibility loci

Amount awarded: £146,674

Dates: 1/12/05 – 30/04/09

Patents:

US Patent 5,239,060: Muscular dystrophy protein, dystrophin; Issued 24 August 1993

US Patent 5,541,074: Duchenne Muscular Dystrophy (MD Probes); Probes for and Methods of Diagnosis for MD (CIP); Issued 30 July 1996

US Patent 5,621,091: Probes for and nucleic acids encoding muscular dystrophy protein, dystrophin; Issued 15 April 1997

Australia Patent 633,249: Duchenne Muscular Dystrophy (MD Probes); Probes for and Methods of Diagnosis for MD (CIP); Issued 21 May 1993

Publications:

* indicates important contribution to field

ResearchID.com: A-4495-2010

<http://www.researcherid.com/rid/A-4495-2010>

Citations Total (Web of Science January 2017) 29,025

h-index 81

Average citation per article: 93.93

Reported as Highly Cited Researcher in ISI HighlyCited.Com

Publications as an undergraduate and medical student:

1. Hoebel BG, Hernandez L, Monaco AP, Miller WC. Amphetamine-induced overeating and overweight in rats. **Life Sci.** 1981 Jan 5;28(1):77-82. PMID: 6938757

2*. Hoebel BG, Monaco AP, Hernandez L, Aulisi EF, Stanley BG, Lenard L. Self-injection of amphetamine directly into the brain. **Psychopharmacology (Berl).** 1983;81(2):158-63. PMID: 6415748

3. Monaco AP, Baghdoyan HA, Nelson JP, Hobson JA. Cortical wave amplitude and eye movement direction are correlated in REM sleep but not in waking. **Arch Ital Biol.** 1984 Sep;122(3):213-23. PMID: 6517651

4. Baghdoyan HA, Monaco AP, Rodrigo-Angulo ML, Assens F, McCarley RW, Hobson JA. Microinjection of neostigmine into the pontine reticular formation of cats enhances desynchronized sleep signs. **J Pharmacol Exp Ther.** 1984 Oct;231(1):173-80. PMID: 6491973

Publications as a PhD student:

5. Kunkel LM, Lalande M, Monaco AP, Flint A, Middlesworth W, Latt SA. Construction of a human X-chromosome-enriched phage library which facilitates analysis of specific loci. **Gene.** 1985;33(3):251-8. PMID: 2989089

- *6. Kunkel LM, Monaco AP, Middlesworth W, Ochs HD, Latt SA. Specific cloning of DNA fragments absent from the DNA of a male patient with an X chromosome deletion. **Proc Natl Acad Sci U S A**. 1985 Jul;82(14):4778-82. PMID: 2991893
- *7. Monaco AP, Bertelson CJ, Middlesworth W, Colletti CA, Aldridge J, Fischbeck KH, Bartlett R, Pericak-Vance MA, Roses AD, Kunkel LM. Detection of deletions spanning the Duchenne muscular dystrophy locus using a tightly linked DNA segment. **Nature**. 1985 Aug 29-Sep 4;316(6031):842-5. PMID: 2993910
- *8. Kunkel LM, Hejtmancik JF, Caskey CT, Speer A, Monaco AP, Middlesworth W, Colletti CA, Bertelson C, Müller U, Bresnan M, Shapiro F, Tantravahi U, Speer J, Latt SA, Bartlett R, Pericak-Vance MA, Roses AD, Thompson MW, Ray PN, Worton RG, Fischbeck KH, Gallano P, Coulon M, Duros C, Boue J, Junien C, Chelly J, Hamard G, Jeanpierre M, Lambert M, Kaplan JC, Emery A, Dorkins H, McGlade S, Davies KE, Boehm C, Arveiler B, Lemaire C, Morgan GJ, Denton MJ, Amos J, Bobrow M, Benham F, Boswinkel E, Cole C, Dubowitz V, Hart K, Hodgson S, Johnson L, Walker A, Roncuzzi L, Ferlini A, Nobile C, Romeo G, Wilcox DE, Affara NA, Ferguson-Smith MA, Lindolf M, Kaariainen H, de la Chapelle A, Ionasescu V, Searby C, Ionasescu R, Bakker E, van Ommen GJ, Pearson PL, Greenberg CR, Hamerton JL, Wrogemann K, Doherty RA, Polakowska R, Hyser C, Quirk S, Thomas N, Harper JF, Darras BT, Francke U. Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. **Nature**. 1986 Jul 3-9;322(6074):73-7. PMID: 3014348
- *9. Monaco AP, Neve RL, Colletti-Feener C, Bertelson CJ, Kurnit DM, Kunkel LM. Isolation of candidate cDNAs for portions of the Duchenne muscular dystrophy gene. **Nature**. 1986 Oct 16-22;323(6089):646-50. PMID: 3773991
10. Baehner RL, Kunkel LM, Monaco AP, Haines JL, Conneally PM, Palmer C, Heerema N, Orkin SH. DNA linkage analysis of X chromosome-linked chronic granulomatous disease. **Proc Natl Acad Sci U S A**. 1986 May;83(10):3398-401. PMID: 3010296
- *11. Royer-Pokora B, Kunkel LM, Monaco AP, Goff SC, Newburger PE, Baehner RL, Cole FS, Curnutte JT, Orkin SH. Cloning the gene for an inherited human disorder--chronic granulomatous disease--on the basis of its chromosomal location. **Nature**. 1986 Jul 3-9;322(6074):32-8. PMID: 2425263
12. Müller U, Tantravahi U, Monaco A, Stroh H, Kunkel LM, Latt SA. Repeated DNA sequences in the distal long arm of the human X chromosome. **Hum Genet**. 1986 Sep;74(1):24-9. PMID: 3019869
13. Fischbeck KH, Ritter AW, Tirschwell DL, Kunkel LM, Bertelson CJ, Monaco AP, Hejtmancik JF, Boehm C, Ionasescu V, Ionasescu R, et al. Recombination with pERT87 (DXS164) in families with X-linked muscular dystrophy. **Lancet**. 1986 Jul 12;2(8498):104. PMID: 2873362
14. van Ommen GJ, Verkerk JM, Hofker MH, Monaco AP, Kunkel LM, Ray P, Worton R, Wieringa B, Bakker E, Pearson PL. A physical map of 4 million bp around the Duchenne

- muscular dystrophy gene on the human X-chromosome. **Cell**. 1986 Nov 21;47(4):499-504. PMID: 2877741
15. Bertelson CJ, Bartley JA, Monaco AP, Colletti-Feener C, Fischbeck K, Kunkel LM. Localisation of Xp21 meiotic exchange points in Duchenne muscular dystrophy families. **J Med Genet**. 1986 Dec;23(6):531-7. PMID: 2879924
16. Kunkel LM, Monaco AP, Bertelson CJ, Colletti CA. Molecular genetics of Duchenne muscular dystrophy. **Cold Spring Harb Symp Quant Biol**. 1986;51 Pt 1:349-51. PMID: 3472731
17. Royer-Pokora B, Kunkel LM, Monaco AP, Goff SC, Newburger PE, Baehner RL, Cole FS, Curnutte JT, Orkin SH. Cloning the gene for the inherited disorder chronic granulomatous disease on the basis of its chromosomal location. **Cold Spring Harb Symp Quant Biol**. 1986;51 Pt 1:177-83. PMID: 3472714
18. Monaco AP, Bertelson CJ, Colletti-Feener C, Kunkel LM. Localization and cloning of Xp21 deletion breakpoints involved in muscular dystrophy. **Hum Genet**. 1987 Mar;75(3):221-7. PMID: 2881877
19. Boyd Y, Munro E, Ray P, Worton R, Monaco T, Kunkel L, Craig I. Molecular heterogeneity of translocations associated with muscular dystrophy. **Clin Genet**. 1987 Apr;31(4):265-72. PMID: 3594934
20. Monaco AP, Kunkel LM. A giant locus for the Duchenne and Becker muscular dystrophy gene. **Trends Genet** 1987; 3:33-37.
- *21. Koenig M, Hoffman EP, Bertelson CJ, Monaco AP, Feener C, Kunkel LM. Complete cloning of the Duchenne muscular dystrophy (DMD) cDNA and preliminary genomic organization of the DMD gene in normal and affected individuals. **Cell**. 1987 Jul 31;50(3):509-17. PMID: 3607877
22. Hart KA, Monaco AP, Kunkel LM, Bobrow M. A small deletion in the Duchenne/Becker muscular dystrophy locus--a functionally important region? **Hum Genet**. 1987 Sep;77(1):88-91. PMID: 3040577
- *23. Hoffman EP, Monaco AP, Feener CC, Kunkel LM. Conservation of the Duchenne muscular dystrophy gene in mice and humans. **Science**. 1987 Oct 16;238(4825):347-50. PMID: 3659917
24. Chamberlain JS, Grant SG, Reeves AA, Mullins LJ, Stephenson DA, Hoffman EP, Monaco AP, Kunkel LM, Caskey CT, Chapman VM. Regional localization of the murine Duchenne muscular dystrophy gene on the mouse X chromosome. **Somat Cell Mol Genet**. 1987 Nov;13(6):671-8. PMID: 2890215
25. Kunkel LM, Monaco AP, Hoffman E, Koenig M, Feener C, Bertelson C. Molecular studies of progressive muscular dystrophy (Duchenne). **Enzyme**. 1987;38(1-4):72-5. PMID: 3440453

*26. Koenig M, Monaco AP, Kunkel LM. The complete sequence of dystrophin predicts a rod-shaped cytoskeletal protein. **Cell**. 1988 Apr 22;53(2):219-28. PMID: 3282674

27. Burmeister M, Monaco AP, Gillard EF, van Ommen GJ, Affara NA, Ferguson-Smith MA, Kunkel LM, Lehrach H. A 10-megabase physical map of human Xp21, including the Duchenne muscular dystrophy gene. **Genomics**. 1988 Apr;2(3):189-202. PMID: 3397058

*28. Monaco AP, Bertelson CJ, Liechti-Gallati S, Moser H, Kunkel LM. An explanation for the phenotypic differences between patients bearing partial deletions of the DMD locus. **Genomics**. 1988 Jan;2(1):90-5. PMID: 3384440

29. Monaco AP, Kunkel LM. Cloning of the Duchenne/Becker muscular dystrophy locus. **Adv Hum Genet**. 1988;17:61-98. PMID: 3055851

Publications as a postdoctoral fellow:

30. Monaco AP. Dystrophin, the protein product of the Duchenne/Becker muscular dystrophy gene. **Trends Biochem Sci**. 1989 Oct;14(10):412-5. PMID: 2683261

31. Monaco AP. The Duchenne and Becker muscular dystrophy gene and protein product dystrophin. **Current Opinion in Pediatrics** 1989; 1:406-412

32. Carter ND, Morgan JE, Monaco AP, Schwartz MS, Jeffery S. Dystrophin expression and genotypic analysis of two cases of benign X linked myopathy (McLeod's syndrome). **J Med Genet**. 1990 Jun;27(6):345-7. PMID: 2193159

33. Davies KE, Mandel JL, Monaco AP, Nussbaum RL, Willard HF. Report of the committee on the genetic constitution of the X chromosome. **Cytogenet Cell Genet**. 1990;55(1-4):254-313. PMID: 2073838

34. Powell JF, Fodor FH, Cockburn DJ, Monaco AP, Craig IW. A dinucleotide repeat polymorphism at the DMD locus. **Nucleic Acids Res**. 1991 Mar 11;19(5):1159. PMID: 2020555

*35. Nizetić D, Zehetner G, Monaco AP, Gellen L, Young BD, Lehrach H. Construction, arraying, and high-density screening of large insert libraries of human chromosomes X and 21: their potential use as reference libraries. **Proc Natl Acad Sci U S A**. 1991 Apr 15;88(8):3233-7. PMID: 2014245

*36. Larin Z, Monaco AP, Lehrach H. Yeast artificial chromosome libraries containing large inserts from mouse and human DNA. **Proc Natl Acad Sci U S A**. 1991 May 15;88(10):4123-7. PMID: 2034658

37. Monaco AP, Müller U, Larin Z, Meier-Ewert S, Lehrach H. Isolation of the human sex determining region from a Y-enriched yeast artificial chromosome library. **Genomics**. 1991 Dec;11(4):1049-53. PMID: 1783376

38. Monaco AP, Lam VM, Zehetner G, Lennon GG, Douglas C, Nizetic D, Goodfellow PN, Lehrach H. Mapping irradiation hybrids to cosmid and yeast artificial chromosome libraries by direct hybridization of Alu-PCR products. **Nucleic Acids Res.** 1991 Jun 25;19(12):3315-8. PMID: 2062647
39. Ragoussis J, Monaco A, Mockridge I, Kendall E, Campbell RD, Trowsdale J. Cloning of the HLA class II region in yeast artificial chromosomes. **Proc Natl Acad Sci U S A.** 1991 May 1;88(9):3753-7. PMID: 1673791
40. Dietrich A, Kioschis P, Monaco AP, Gross B, Korn B, Williams SV, Sheer D, Heitz D, Oberle I, Toniolo D, et al. Molecular cloning and analysis of the fragile X region in man. **Nucleic Acids Res.** 1991 May 25;19(10):2567-72. PMID: 2041732
41. Hoheisel JD, Drmanac R, Larin Z, Lennon G, Monaco AP, Nizetic D, Ross M, Zehetner G, and Lehrach H. Use of high coverage libraries for an integrated analysis of genomic DNA. **Advances in Mol Gen** 1991; 4:125-132.
42. Grootsholten PM, Den Dunnen JT, Monaco AP, Anand R, and Van Ommen GJB. YAC mapping strategies applied to the DMD-gene. **Technique** 1991; 3:41-50.
43. Davies KE, Mandel JL, Monaco AP, Nussbaum RL, Willard HF. Report of the committee on the genetic constitution of the X chromosome. **Cytogenet Cell Genet.** 1990;55(1-4):254-313. PMID: 2073838
44. Monaco AP. Workshop report: Enabling technologies. **Cytogenet Cell Genet** 1991; 58:1845-1846.

Publications as Head of ICRF Human Genetics Laboratory:

- *45. Ho MF, Monaco AP, Blondin LA, van Ommen GJ, Affara NA, Ferguson-Smith MA, Lehrach H. Fine mapping of the McLeod locus (XK) to a 150-380-kb region in Xp21. **Am J Hum Genet.** 1992 Feb;50(2):317-30. PMID: 1734714
- *46. Monaco AP, Walker AP, Millwood I, Larin Z, Lehrach H. A yeast artificial chromosome contig containing the complete Duchenne muscular dystrophy gene. **Genomics.** 1992 Mar;12(3):465-73. PMID: 1559698
47. Den Dunnen JT, Grootsholten PM, Dauwerse JG, Walker AP, Monaco AP, Butler R, Anand R, Coffey AJ, Bentley DR, Steensma HY, Van Ommen GJB. Reconstruction of the 2.4 Mb human DMD-gene by homologous YAC recombination. **Hum Mol Genet.** 1992 Apr;1(1):19-28. PMID: 1301131
48. Hugnot JP, Gilgenkrantz H, Vincent N, Chafey P, Morris GE, Monaco AP, Berwald-Netter Y, Koulakoff A, Kaplan JC, Kahn A, et al. Distal transcript of the dystrophin gene initiated from an alternative first exon and encoding a 75-kDa protein widely distributed in nonmuscle tissues. **Proc Natl Acad Sci U S A.** 1992 Aug 15;89(16):7506-10. PMID: 1380160

49. Berger W, Meindl A, van de Pol TJ, Cremers FP, Ropers HH, Döerner C, Monaco A, Bergen AA, Lebo R, Warburg M, et al. Isolation of a candidate gene for Norrie disease by positional cloning. **Nat Genet.** 1992 Jun;1(3):199-203. Erratum in: *Nat Genet.* 1992 Sep;2(1):84. PMID: 1303235
50. Chen ZY, Sims KB, Coleman M, Donnai D, Monaco A, Breakefield XO, Davies KE, Craig IW. Characterization of a YAC containing part or all of the Norrie disease locus. **Hum Mol Genet.** 1992 Jun;1(3):161-4. PMID: 1303171
51. Bates GP, Valdes J, Hummerich H, Baxendale S, Le Paslier DL, Monaco AP, Tagle D, MacDonald ME, Altherr M, Ross M, et al. Characterization of a yeast artificial chromosome contig spanning the Huntington's disease gene candidate region. **Nat Genet.** 1992 Jun;1(3):180-7. PMID: 1303232
52. Maier E, Hoheisel JD, McCarthy L, Mott R, Grigoriev AV, Monaco AP, Larin Z, Lehrach H. Complete coverage of the *Schizosaccharomyces pombe* genome in yeast artificial chromosomes. **Nat Genet.** 1992 Jul;1(4):273-7. PMID: 1302023, Citations 85.
53. Brzustowicz LM, Kleyn PW, Boyce FM, Lien LL, Monaco AP, Penchaszadeh GK, Das K, Wang CH, Munsat TL, Ott J, et al. Fine-mapping of the spinal muscular atrophy locus to a region flanked by MAP1B and D5S6. **Genomics.** 1992 Aug;13(4):991-8. PMID: 1505990
54. Graeber MB, Monaco AP, Chelly J, Müller U. Isolation of DNTR polymorphisms from yeast artificial chromosomes encompassing X chromosomal loci PGK1 and DXS56. **Hum Genet.** 1992 Nov;90(3):270-4. PMID: 1339399
- *55. Tümer Z, Chelly J, Tommerup N, Ishikawa-Brush Y, Tønnesen T, Monaco AP, Horn N. Characterization of a 1.0 Mb YAC contig spanning two chromosome breakpoints related to Menkes disease. **Hum Mol Genet.** 1992 Oct;1(7):483-9. PMID: 1307248
56. Górecki DC, Monaco AP, Derry JM, Walker AP, Barnard EA, Barnard PJ. Expression of four alternative dystrophin transcripts in brain regions regulated by different promoters. **Hum Mol Genet.** 1992 Oct;1(7):505-10. PMID: 1307251
- *57. Walker AP, Chelly J, Love DR, Ishikawa-Brush Y, Récan D, Chaussain J-L, Oley CA, Conner JM, Yates J, Price DA, Super M, Bottani A, Steinmann B, Kaplan J-C, Davies KE, Monaco AP. A YAC contig in Xp21 containing the adrenal hypoplasia and glycerol kinase deficiency genes. **Hum Mol Genet** 1992; 1:579-585.
58. Mandel JL, Monaco AP, Nelson DL, Schlessinger D, Willard HF, Chipperfield M, Pearson P, Gilna P, Cinkosky M. Genome maps III. 1992. Wall Chart. **Science.** 1992 Oct 2;258(5079):87-102. PMID: 1439771
- *59. Mandel JL, Monaco AP, Nelson DL, Schlessinger D, Willard H. Genome analysis and the human X chromosome. **Science.** 1992 Oct 2;258(5079):103-9. PMID: 1439756
60. Knight JC, Reeves BR, Kearney L, Monaco AP, Lehrach H, Cooper CS. Localization of the synovial sarcoma $t(X;18)(p11.2;q11.2)$ breakpoint by

fluorescence in situ hybridization. **Hum Mol Genet.** 1992 Nov;1(8):633-7. PMID: 1338692

61. Muscatelli F, Monaco AP, Goodfellow PN, Hors-Cayla MC, Lehrach H, Fontes M. Isolation of new probes from Xq12-->q13: an example of the screening of reference libraries with Alu-PCR products from radiation hybrids. **Cytogenet Cell Genet.** 1992;61(2):109-13. PMID: 1395715

62. Baldrich K, Baldrich M, Monaco AP, Müller CR. Replication errors may contribute to the generation of large deletions and duplications in the dystrophin gene. **Hum Mutat.** 1992;1(4):280-7. PMID: 1301934

63. de Leeuw B, Berger W, Sinke RJ, Suijkerbuijk RF, Gilgenkrantz S, Geraghty MT, Valle D, Monaco AP, Lehrach H, Ropers HH, et al. Identification of a yeast artificial chromosome (YAC) spanning the synovial sarcoma-specific t(X;18)(p11.2;q11.2) breakpoint. **Genes Chromosomes Cancer.** 1993 Mar;6(3):182-9. PMID: 7682104

64. Bergen AA, Wapenaar MC, Schuurman EJ, Diergaarde PJ, Lerach H, Monaco AP, Bakker E, Bleeker-Wagemakers EM, van Ommen GJ. Detection of a new submicroscopic Norrie disease deletion interval with a novel DNA probe isolated by differential Alu PCR fingerprint cloning. **Cytogenet Cell Genet.** 1993;62(4):231-5. PMID: 8440142

*65. Chelly J, Tümer Z, Tønnesen T, Petterson A, Ishikawa-Brush Y, Tommerup N, Horn N, Monaco AP. Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. **Nat Genet.** 1993 Jan;3(1):14-9. PMID: 8490646

66. Wang Q, Ishikawa-Brush Y, Monaco AP, Nelson DL, Caskey CT, Pauly SP, Lenoir GM, Sylla BS. Physical mapping of Xq24-25 around loci closely linked to the X-linked lymphoproliferative syndrome locus: an overlapping YAC map and linkage between DXS12, DXS42, and DXS37. **Eur J Hum Genet.** 1993;1(1):64-71. PMID: 8069652

*67. Walker AP, Muscatelli F, Monaco AP. Isolation of the human Xp21 glycerol kinase gene by positional cloning. **Hum Mol Genet.** 1993 Feb;2(2):107-14. PMID: 8499898

68. Fairweather N, Chelly J, Monaco AP. Dinucleotide repeat polymorphisms from DXS106 and DXS227 YACs using a two stage approach. **Hum Mol Genet.** 1993 May;2(5):607-8. PMID: 8518803

69. Geraghty MT, Brody LC, Martin LS, Marble M, Kearns W, Pearson P, Monaco AP, Lehrach H, Valle D. The isolation of cDNAs from OATL1 at Xp 11.2 using a 480-kb YAC. **Genomics.** 1993 May;16(2):440-6. PMID: 8314581

70. Markiewicz S, DiSanto JP, Chelly J, Fairweather N, Le Marec B, Griscelli C, Graeber MB, Müller U, Fischer A, Monaco AP, et al. de Saint Basile G. Fine mapping of the human SCIDX1 locus at Xq12-13.1. **Hum Mol Genet.** 1993 Jun;2(6):651-4. PMID: 8353486

71. Lafrenière RG, Brown CJ, Rider S, Chelly J, Taillon-Miller P, Chinault AC, Monaco AP, Willard HF. 2.6 Mb YAC contig of the human X inactivation center region in Xq13: physical linkage of the RPS4X, PHKA1, XIST and DXS128E genes. **Hum Mol**

Genet. 1993 Aug;2(8):1105-15. PMID: 8401491

72. Francis MJ, Morrison KE, Campbell L, Grewal PK, Christodoulou Z, Daniels RJ, Monaco AP, Frischauf AM, McPherson J, Wasmuth J, et al. A contig of non-chimaeric YACs containing the spinal muscular atrophy gene in 5q13. **Hum Mol Genet.** 1993 Aug;2(8):1161-7. PMID: 8401497

73. Rider SH, Monaco AP. Primers for the dinucleotide repeat at the DXS453 locus also recognizes the DXS983 locus. **Hum Mol Genet.** 1993 Sep;2(9):1510. PMID: 8242089

74. Thomas NS, Chelly J, Zonana J, Davies KJ, Morgan S, Gault J, Rack KA, Buckle VJ, Brockdorff N, Clarke A, Monaco AP. Characterisation of molecular DNA rearrangements within the Xq12-q13.1 region, in three patients with X-linked hypohidrotic ectodermal dysplasia (EDA). **Hum Mol Genet.** 1993 Oct;2(10):1679-85. PMID: 8268921

75. Graeber MB, Müller U, Monaco AP, Weber JL. Four dinucleotide repeat polymorphisms at the D7S804 locus. **Hum Mol Genet.** 1993 Dec;2(12):2195. PMID: 8111392

76. Suijkerbuijk RF, Meloni AM, Sinke RJ, de Leeuw B, Wilbrink M, Janssen HA, Geraghty MT, Monaco AP, Sandberg AA, Geurts van Kessel A. Identification of a yeast artificial chromosome that spans the human papillary renal cell carcinoma-associated t(X;1) breakpoint in Xp11.2. **Cancer Genet Cytogenet.** 1993 Dec;71(2):164-9. PMID: 8281521

77. Schlessinger D, Mandel JL, Monaco AP, Nelson DL, Willard HF. Report and abstracts of the Fourth International Workshop on Human X Chromosome Mapping 1993. St. Louis, Missouri, May 9-12, 1993. **Cytogenet Cell Genet.** 1993;64(3-4):147-94. PMID: 8404034

78. Cox RD, Meier-Ewert S, Ross M, Larin Z, Monaco AP, Lehrach H. Genome mapping and cloning of mutations using yeast artificial chromosomes. **Methods Enzymol.** 1993;225:637-53. PMID: 8231876

79. Larin Z, Monaco AP, Meier-Ewert S, Lehrach H. Construction and characterization of yeast artificial chromosome libraries from the mouse genome. **Methods Enzymol.** 1993;225:623-37. PMID: 8231875

80. Monaco AP. Molecular human genetics and the Duchenne/Becker muscular dystrophy gene. **Mol Cell Biol Hum Dis Ser.** 1993;3:1-11. PMID: 8111535

81. Pearce M, Blake DJ, Tinsley JM, Byth BC, Campbell L, Monaco AP, Davies KE. The utrophin and dystrophin genes share similarities in genomic structure. **Hum Mol Genet.** 1993 Nov;2(11):1765-72. PMID: 8281135

82. Chelly J, Monaco AP. Cloning the Wilson disease gene. **Nat Genet.** 1993 Dec;5(4):317-8. PMID: 8298634

83. Reed V, Rider S, Maslen GL, Hatchwell E, Blair HJ, Uwechue IC, Craig IW, Laval SH, Monaco AP, Boyd Y. A 2-Mb YAC contig encompassing three loci (DXF34, DXS14, and DXS390) that lie between Xp11.2 translocation breakpoints associated with

- incontinentia pigmenti type 1. **Genomics**. 1994 Apr;20(3):341-6. PMID: 8034305
84. Francis F, Benham F, See CG, Fox M, Ishikawa-Brush Y, Monaco AP, Weiss B, Rappold G, Hamvas RM, Lehrach H. Identification of YAC and cosmid clones encompassing the ZFX-POLA region using irradiation hybrid cell lines. **Genomics**. 1994 Mar 1;20(1):75-83. PMID: 8020959
85. Cochrane S, Bergoffen J, Fairweather ND, Müller E, Mostacciuolo ML, Monaco AP, Fischbeck KH, Haites NE. X linked Charcot-Marie-Tooth disease (CMTX1): a study of 15 families with 12 highly informative polymorphisms. **J Med Genet**. 1994 Mar;31(3):193-6. PMID: 7912286
86. Rack KA, Chelly J, Gibbons RJ, Rider S, Benjamin D, Lafreniere RG, Oscier D, Hendricks RW, Craig IW, Willard HF, Monaco AP, Buckle VJ. Absence of the XIST gene from late-replicating isodicentric X chromosomes in leukaemia. **Hum Mol Genet**. 1994 Jul;3(7):1053-9. PMID: 7981672
87. Müller U, Haberhausen G, Wagner T, Fairweather ND, Chelly J, Monaco AP. DXS106 and DXS559 flank the X-linked dystonia-parkinsonism syndrome locus (DYT3). **Genomics**. 1994 Sep 1;23(1):114-7. PMID: 7829058
88. George AM, Reed V, Glenister P, Chelly J, Tümer Z, Horn N, Monaco AP, Boyd Y. Analysis of Mnk, the murine homologue of the locus for Menkes disease, in normal and mottled (Mo) mice. **Genomics**. 1994 Jul 1;22(1):27-35. PMID: 7959788
89. Shipley JM, Clark J, Crew AJ, Birdsall S, Rocques PJ, Gill S, Chelly J, Monaco AP, Abe S, Gusterson BA, et al. The t(X;18)(p11.2;q11.2) translocation found in human synovial sarcomas involves two distinct loci on the X chromosome. **Oncogene**. 1994 May;9(5):1447-53. PMID: 8152806
90. Matfin G, Sheaves R, Muscatelli F, Walker A, Monaco A, Grant D, Nwose O, Wass JA. Gene deletion causing adrenal hypoplasia congenita and hypogonadotropic hypogonadism. **Clin Endocrinol (Oxf)**. 1994 Jun;40(6):807-8. PMID: 8033374
- *91. Fairweather N, Bell C, Cochrane S, Chelly J, Wang S, Mostacciuolo ML, Monaco AP, Haites NE. Mutations in the connexin 32 gene in X-linked dominant Charcot-Marie-Tooth disease (CMTX1) **Hum Mol Genet**. 1994 Jan;3(1):29-34. Erratum in: *Hum Mol Genet* 1994 Jun;3(6):1034. PMID: 8162049
92. Gong W, Kioschis P, Rogner U, Monaco AP, Poustka A. Identification of region-specific cosmid clones by hybridization with Alu-LINE PCR products of Yeast Artificial Chromosome clones. **Methods Mol Cell Biol** 1994; 4:269-272.
93. Monaco AP, Larin Z. YACs, BACs, PACs and MACs: artificial chromosomes as research tools. **Trends Biotechnol**. 1994 Jul;12(7):280-6. Review. PMID: 7765076
- *94. Ho M, Chelly J, Carter N, Danek A, Crocker P, Monaco AP. Isolation of the gene for McLeod syndrome that encodes a novel membrane transport protein. **Cell**. 1994 Jun

17;77(6):869-80. PMID: 8004674

95. Monaco AP. Isolation of genes from cloned DNA. **Curr Opin Genet Dev.** 1994 Jun;4(3):360-5. PMID: 7919912

96. Monaco AP, Larin Z, Lehrach H. Construction of yeast artificial chromosome libraries by pulsed-field gel electrophoresis. **Mol Biotechnol.** 1994 Jun;1(3):241-9. PMID: 7859163.

97. Hagemann T, Surosky R, Monaco AP, Lehrach H, Rosen FS, Kwan SP. Physical mapping in a YAC contig of 11 markers on the human X chromosome in Xp11.23. **Genomics.** 1994 May 1;21(1):262-5. PMID: 8088799

98. Stafford AN, Rider SH, Hopkin JM, Cookson WO, Monaco AP. A 2.8 Mb YAC contig in 11q12-q13 localizes candidate genes for atopy: Fc epsilon RI beta and CD20. **Hum Mol Genet.** 1994 May;3(5):779-85. PMID: 7521709

99. Huber I, Bitner-Glindzicz M, de Kok YJ, van der Maarel SM, Ishikawa-Brush Y, Monaco AP, Robinson D, Malcolm S, Pembrey ME, Brunner HG, et al. X-linked mixed deafness (DFN3): cloning and characterization of the critical region allows the identification of novel microdeletions. **Hum Mol Genet.** 1994 Jul;3(7):1151-4. PMID: 7981685

*100. Zanaria E, Muscatelli F, Bardoni B, Strom TM, Guioli S, Guo W, Lalli E, Moser C, Walker AP, McCabe ER, et al. An unusual member of the nuclear hormone receptor superfamily responsible for X-linked adrenal hypoplasia congenita. **Nature.** 1994 Dec 15;372(6507):635-41. PMID: 7990953

*101. Muscatelli F, Strom TM, Walker AP, Zanaria E, Récan D, Meindl A, Bardoni B, Guioli S, Zehetner G, Rabl W, et al. Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. **Nature.** 1994 Dec 15;372(6507):672-6. PMID: 7990958

102. Stone C, Pointon JJ, Jazwinska EC, Halliday JW, Powell LW, Robson KJ, Monaco AP, Weatherall DJ. Isolation of CA dinucleotide repeats close to D6S105; linkage disequilibrium with haemochromatosis. **Hum Mol Genet.** 1994 Nov;3(11):2043-6. PMID: 7874124

103. Willard HF, Cremers F, Mandel JL, Monaco AP, Nelson DL, Schlessinger D. Report and abstracts of the Fifth International Workshop on Human X Chromosome Mapping 1994. Heidelberg, Germany, April 24-27, 1994. **Cytogenet Cell Genet.** 1994;67(4):295-358. PMID: 7924455

104. Pericak-Vance MA, Barker DF, Bergoffen J, Chance P, Cochrane S, Dahl N, Exler M-C, Fain PR, Fairweather ND, Fischbeck K, Gal A, Haites N, Ionasescu R, Ionasescu VV, Kennerson ML, Monaco AP, Mostacciuolo M, Nicholson GA, Sillén A, Haines JL. Consortium fine localization of X-linked Charcot-Marie-Tooth disease (CMTX1): additional support that connexin32 is the defect in CMTX1. **Hum Hered.** 1995 May-Jun;45(3):121-8. PMID: 7615296

105. Fletcher FA, Huebner K, Shaffer LG, Fairweather ND, Monaco AP, Müller U, Druck T, Simoneaux DK, Chelly J, Belmont JW, et al. Assignment of the gene (EPLG2) encoding a high-affinity binding protein for the receptor tyrosine kinase elk to a 200-kilobasepair region in human chromosome Xq12. **Genomics**. 1995 Jan 1;25(1):334-5. PMID: 7774950

106. Villard L, Gecz J, Colleaux L, Lossi AM, Chelly J, Ishikawa-Brush Y, Monaco AP, Fontes M. Construction of a YAC contig spanning the Xq13.3 subband. **Genomics**. 1995 Mar 1;26(1):115-22. PMID: 7782069

107. Monaco AP, Chelly J. Menkes and Wilson diseases. **Adv Genet**. 1995;33:233-53. PMID: 7484454

108. de Kok YJ, van der Maarel SM, Bitner-Glindzicz M, Huber I, Monaco AP, Malcolm S, Pembrey ME, Ropers HH, Cremers FP. Association between X-linked mixed deafness and mutations in the POU domain gene POU3F4. **Science**. 1995 Feb 3;267(5198):685-8. PMID: 7839145

109. Tümer Z, Vural B, Tønnesen T, Chelly J, Monaco AP, Horn N. Characterization of the exon structure of the Menkes disease gene using vectorette PCR. **Genomics**. 1995 Apr 10;26(3):437-42. PMID: 7607665

Publications as Wellcome Trust Principal Research Fellow:

110. Bolton P, Powell J, Rutter M, Buckle V, Yates JR, Ishikawa-Brush Y, Monaco AP. Autism, mental retardation, multiple exostoses and short stature in a female with 46,X,t(X;8)(p22.13;q22.1). **Psychiatr Genet**. 1995 Summer;5(2):51-5. PMID: 7551962

*111. Muscatelli F, Walker AP, De Plaen E, Stafford AN, Monaco AP. Isolation and characterization of a MAGE gene family in the Xp21.3 region. **Proc Natl Acad Sci U S A**. 1995 May 23;92(11):4987-91. PMID: 7761436

112. Miller AP, Gustashaw K, Wolff DJ, Rider SH, Monaco AP, Eble B, Schlessinger D, Gorski JL, van Ommen GJ, Weissenbach J, et al. Three genes that escape X chromosome inactivation are clustered within a 6 Mb YAC contig and STS map in Xp11.21-p11.22. **Hum Mol Genet**. 1995 Apr;4(4):731-9. PMID: 7633424

113. Philippe C, Arnould C, Sloan F, van Bokhoven H, van der Velde-Visser SD, Chery M, Ropers HH, Gilgenkrantz S, Monaco AP, Cremers FP. A high-resolution interval map of the q21 region of the human X chromosome. **Genomics**. 1995 Jun 10;27(3):539-43. PMID: 7558039

114. Haberhausen G, Schmitt I, Köhler A, Peters U, Rider S, Chelly J, Terwilliger JD, Monaco AP, Müller U. Assignment of the dystonia-parkinsonism syndrome locus, DYT3, to a small region within a 1.8-Mb YAC contig of Xq13.1. **Am J Hum Genet**. 1995 Sep;57(3):644-50. PMID: 7668293

115. van der Maarel SM, Scholten

IHJM, Maat-Kievit JA, Huber I, de Kok YJM,
20

- de Wijs I, van Bokhoven H, den Dunnen JT, van Ommen GJB, Philippe C, Monaco AP, Smeets HJM, Ropers H-H, Cremers FPM. Yeast artificial chromosome cloning of the Xq13.3-q21.31 region and fine mapping of a deletion associated with choroideremia and nonspecific mental retardation. **Eur J Hum Genet.** 1995;3(4):207-18. PMID: 8528669
116. Blair HJ, Ho M, Monaco AP, Fisher S, Craig IW, Boyd Y. High-resolution comparative mapping of the proximal region of the mouse X chromosome. **Genomics.** 1995 Jul 20;28(2):305-10. PMID: 8530041
117. Fisher SE, Hatchwell E, Chand A, Ockenden N, Monaco AP, Craig IW. Construction of two YAC contigs in human Xp11.23-p11.22, one encompassing the loci OATL1, GATA, TFE3, and SYP, the other linking DXS255 to DXS146. **Genomics.** 1995 Sep 20;29(2):496-502. PMID: 8666400
118. Millwood IY, Blake DJ, Gauguier D, Monaco AP. Two polymorphic dinucleotide repeats in the rat dystrophin gene, including the conserved 3' UTR repeat. **Mamm Genome.** 1995 Sep;6(9):668-9. PMID: 8535080
119. Dry KL, Aldred MA, Edgar AE, Brown J, Manson FDC, Ho MF, Prosser J, Hardwick LJ, Lennon AA, Thomson K, van Keuren M, Kurnit DM, Bird AC, Jay M, Monaco AP, Wright AF. Identification of a novel gene, ETX1 from Xp21.1, a candidate gene for X-linked retinitis pigmentosa (RP3). **Hum Mol Genet.** 1995 Dec;4(12):2347-53. PMID: 8634709
120. Nelson DL, Ballabio A, Cremers F, Monaco AP, Schlessinger D. Report of the 6th international workshop on X chromosome mapping 1995. **Cytogenet Cell Genet** 1995; 71:308-336
121. Ho MF, Chalmers RM, Davis MB, Harding AE, Monaco AP. A novel point mutation in the McLeod syndrome gene in neuroacanthocytosis. **Ann Neurol.** 1996 May;39(5):672-5. PMID: 8619554
122. Ragoussis J, Monaco AP. Covering YAC-cloned DNA with phages and cosmids. **Methods Mol Biol.** 1996;54:157-66. PMID: 8597788
123. Larin Z, Monaco AP, Lehrach H. Generation of large insert YAC libraries. **Methods Mol Biol.** 1996;54:1-11. PMID: 8597781
124. Walker AP, Muscatelli F, Stafford AN, Chelly J, Dahl N, Blomquist HK, Delanghe J, Willems PJ, Steinmann B, Monaco AP. Mutations and phenotype in isolated glycerol kinase deficiency. **Am J Hum Genet.** 1996 Jun;58(6):1205-11. PMID: 8651297
125. Brown J, Dry KL, Edgar AJ, Pryde FE, Hardwick LJ, Aldred MA, Lester DH, Boyle S, Kaplan J, Dufier JL, Ho MF, Monaco AM, Musarella MA, Wright AF. Analysis of three deletion breakpoints in Xp21.1 and the further localization of RP3. **Genomics.** 1996 Oct 15;37(2):200-10. PMID: 8921393
126. de Gouyon B, Chatterjee A, Monaco A, Quaderi N, Brown SD, Herman GE. Comparative mapping on the mouse X chromosome defines a myotubular

myopathy equivalent region. **Mamm Genome**. 1996 Aug;7(8):575-9. PMID: 8678976

*127. Roest Crolius H, Ross MT, Grigoriev A, Knights CJ, Holloway E, Misfud J, Li K, Playford M, Gregory SG, Humphray SJ, Coffey AJ, See CG, Marsh S, Vatcheva R, Kumlien J, Labella T, Lam V, Rak KH, Todd K, Mott R, Graeser D, Rappold G, Zehetner G, Poustka A, Bentley DR, Monaco AP, Lehrach H. An integrated YAC map of the human X chromosome. **Genome Res**. 1996 Oct;6(10):943-55. PMID: 8908513

128. Kostrzewa M, Köhler A, Eppelt K, Hellam L, Fairweather ND, Levy ER, Monaco AP, Müller U. Assignment of genes encoding GABAA receptor subunits alpha 1, alpha 6, beta 2, and gamma 2 to a YAC contig of 5q33. **Eur J Hum Genet**. 1996;4(4):199-204. PMID: 8875185

*129. Maestrini E, Monaco AP, McGrath JA, Ishida-Yamamoto A, Camisa C, Hovnanian A, Weeks DE, Lathrop M, Uitto J, Christiano AM. A molecular defect in loricrin, the major component of the cornified cell envelope, underlies Vohwinkel's syndrome. **Nat Genet**. 1996 May;13(1):70-7. PMID: 8673107

130. Daniels GL, Weinauer F, Stone C, Ho M, Green CA, Jahn-Jochem H, Offner R, Monaco AP. A combination of the effects of rare genotypes at the XK and KEL blood group loci results in absence of Kell system antigens from the red blood cells. **Blood**. 1996 Nov 15;88(10):4045-50. PMID: 8916972

131. Monaco AP. Human genetics: dissecting Williams syndrome. **Curr Biol**. 1996 Nov 1;6(11):1396-8. PMID: 8939595

*132. Millwood IY, Bihoreau MT, Gauguier D, Hyne G, Levy ER, Kreutz R, Lathrop GM, Monaco AP. A gene-based genetic linkage and comparative map of the rat X chromosome. **Genomics**. 1997 Mar 1;40(2):253-61. PMID: 9119392

133. Zajac V, Kirchhoff T, Levy ER, Horsley SW, Miller A, Steichen-Gersdorf E, Monaco AP. Characterisation of X;17(q12;p13) translocation breakpoints in a female patient with hypomelanosis of Ito and choroid plexus papilloma. **Eur J Hum Genet**. 1997 Mar-Apr;5(2):61-8. PMID: 9195154

*134. Mejía JE, Monaco AP. Retrofitting vectors for Escherichia coli-based artificial chromosomes (PACs and BACs) with markers for transfection studies. **Genome Res**. 1997 Feb;7(2):179-86. PMID: 9049635

135. Woon PY, Gauguier D, Dubay C, Lathrop GM, Monaco AP. Construction of a yeast artificial chromosome library from rat DNA. **Rat Genome** 1997; 3:12-14.

*136. Philippe C, Porter DE, Emerton ME, Wells DE, Simpson AH, Monaco AP. Mutation screening of the EXT1 and EXT2 genes in patients with hereditary multiple exostoses. **Am J Hum Genet**. 1997 Sep;61(3):520-8. PMID: 9326317

137. Ishikawa-Brush Y, Powell JF, Bolton P, Miller AP, Francis F, Willard HF, Lehrach H, Monaco AP. Autism and multiple exostoses associated with an X;8 translocation

occurring within the GRPR gene and 3' to the SDC2 gene. **Hum Mol Genet.** 1997 Aug;6(8):1241-50. PMID: 9259269

138. Boultonwood J, Fidler C, Soularue P, Strickson AJ, Kostrzewa M, Jaju RJ, Cotter FE, Fairweather N, Monaco AP, Müller U, Lovett M, Jabs EW, Auffray C, Wainscoat JS. Novel genes mapping to the critical region of the 5q- syndrome. **Genomics.** 1997 Oct 1;45(1):88-96. PMID: 9339364

*139. Szepietowski P, Rochette J, Berquin P, Piussan C, Lathrop GM, Monaco AP. Familial infantile convulsions and paroxysmal choreoathetosis: a new neurological syndrome linked to the pericentromeric region of human chromosome 16. **Am J Hum Genet.** 1997 Oct;61(4):889-98. PMID: 9382100

*140. Rubio JP, Danek A, Stone C, Chalmers R, Wood N, Verellen C, Ferrer X, Malandrini A, Fabrizi GM, Manfredi M, Vance J, Pericak-Vance M, Brown R, Rudolf G, Picard F, Alonso E, Brin M, Németh AH, Farrall M, Monaco AP. Chorea-acanthocytosis: genetic linkage to chromosome 9q21. **Am J Hum Genet.** 1997 Oct;61(4):899-908. PMID: 9382101

141. Renault B, Hovnanian A, Bryce S, Chang JJ, Lau S, Sakuntabhai A, Monk S, Carter S, Ross CJ, Pang J, Twells R, Chamberlain S, Monaco AP, Strachan T, Kucherlapati R. A sequence-ready physical map of a region of 12q24.1. **Genomics.** 1997 Oct 15;45(2):271-8. PMID: 9344649

142. Millwood IY and Monaco AP. Report on the rat X chromosome. **Rat Genome** 1997; 3:179-190.

143. Larin Z, Monaco AP, Lehrach H. Generation of large insert yeast artificial chromosome libraries. **Mol Biotechnol.** 1997 Oct;8(2):147-53. PMID: 9406185

144. Sloan-Béna F, Philippe C, LeHeup B, Wuilque F, Levy ER, Chéry M, Jonveaux P, Monaco AP. Characterisation of an inverted X chromosome (p11.2q21.3) associated with mental retardation using FISH. **J Med Genet.** 1998 Feb;35(2):146-50. PMID: 9507395.

145. Lurquin C, De Smet C, Brasseur F, Muscatelli F, Martelange V, De Plaen E, Brasseur R, Monaco AP, Boon T. Two members of the human MAGEB gene family located in Xp21.3 are expressed in tumors of various histological origins. **Genomics.** 1997 Dec 15;46(3):397-408. PMID: 9441743

146. Szepietowski P, Monaco AP. Recent progress in the genetics of human epilepsies. **Neurogenetics.** 1998 Mar;1(3):153-63. PMID: 10737118

*147. Fisher SE, Vargha-Khadem F, Watkins KE, Monaco AP, Pembrey ME. Localisation of a gene implicated in a severe speech and language disorder. **Nat Genet.** 1998 Feb;18(2):168-70. Erratum in: *Nat Genet* 1998 Mar;18(3):298. PMID: 9462748

148. Kostrzewa M, Krings BW, Dixon MJ, Eppelt K, Köhler A, Grady DL, Steinberger D, Fairweather ND, Moyzis RK, Monaco AP, Müller U. Integrated physical and transcript map of 5q31.3-qter. **Eur J Hum Genet.** 1998 May-Jun;6(3):266-74. PMID: 9781031

149. Greenfield A, Carrel L, Pennisi D, Philippe C, Quaderi N, Siggers P, Steiner K, Tam PP, Monaco AP, Willard HF, Koopman P. The UTX gene escapes X inactivation in mice and humans. **Hum Mol Genet.** 1998 Apr;7(4):737-42. PMID: 9499428

*150. The International Molecular Genetic Study of Autism Consortium. A full genome screen for autism with evidence for linkage to a region on chromosome 7q. **Hum Mol Genet** 1998; 7:571-578

*151. Francis MJ, Jones EE, Levy ER, Ponnambalam S, Chelly J, Monaco AP. A Golgi localization signal identified in the Menkes recombinant protein. **Hum Mol Genet.** 1998 Aug;7(8):1245-52. PMID: 9668166

151. Monk S, Sakuntabhai A, Carter SA, Bryce SD, Cox R, Harrington L, Levy E, Ruiz-Perez VL, Katsantoni E, Kodvawala A, Munro CS, Burge S, Larrègue M, Nagy G, Rees JL, Lathrop M, Monaco AP, Strachan T, Hovnanian A. Refined genetic mapping of the darier locus to a <1-cM region of chromosome 12q24.1, and construction of a complete, high-resolution P1 artificial chromosome/bacterial artificial chromosome contig of the critical region. **Am J Hum Genet.** 1998 Apr;62(4):890-903. PMID: 9529352

*152. Woon PY, Osoegawa K, Kaisaki PJ, Zhao B, Catanese JJ, Gauguier D, Cox R, Levy ER, Lathrop GM, Monaco AP, de Jong PJ. Construction and characterization of a 10-fold genome equivalent rat P1-derived artificial chromosome library. **Genomics.** 1998 Jun 15;50(3):306-16. PMID: 9676425

153. Hovnanian A, Rebouillat D, Mattei MG, Levy ER, Marié I, Monaco AP, Hovanessian AG. The human 2',5'-oligoadenylate synthetase locus is composed of three distinct genes clustered on chromosome 12q24.2 encoding the 100-, 69-, and 40-kDa forms. **Genomics.** 1998 Sep 15;52(3):267-77. PMID: 9790745

154. Fisher SE, Stein JF, Monaco AP. A genome-wide search strategy for identifying quantitative trait loci involved in reading and spelling disability (developmental dyslexia). **Eur Child Adolesc Psychiatry.** 1999;8 Suppl 3:47-51. Review. PMID: 10638370

155. Lee WL, Tay A, Ong HT, Goh LM, Monaco AP, Szepetowski P. Association of infantile convulsions with paroxysmal dyskinesias (ICCA syndrome): confirmation of linkage to human chromosome 16p12-q12 in a Chinese family. **Hum Genet.** 1998 Nov;103(5):608-12. PMID: 9860304

156. Maestrini E, Marlow AJ, Weeks DE, Monaco AP. Molecular genetic investigations of autism. **J Autism Dev Disord.** 1998 Oct;28(5):427-37. Review. PMID: 9813778

157. Kaisaki PJ, Woon PY, Wallis RH, Monaco AP, Lathrop M, Gauguier D. Localization of tub and uncoupling proteins (Ucp) 2 and 3 to a region of rat chromosome 1 linked to glucose intolerance and adiposity in the Goto-Kakizaki (GK) type 2 diabetic rat. **Mamm Genome.** 1998 Nov;9(11):910-2. PMID: 9799845

*158. Fisher SE, Marlow AJ, Lamb J, Maestrini E, Williams DF, Richardson AJ,

Weeks DE, Stein JF, Monaco AP. A quantitative-trait locus on chromosome 6p influences different aspects of developmental dyslexia. **Am J Hum Genet.** 1999 Jan;64(1):146-56. PMID: 9915953

*159. Sakuntabhai A, Ruiz-Perez V, Carter S, Jacobsen N, Burge S, Monk S, Smith M, Munro CS, O'Donovan M, Craddock N, Kucherlapati R, Rees JL, Owen M, Lathrop GM, Monaco AP, Strachan T, Hovnanian A. Mutations in ATP2A2, encoding a Ca²⁺ pump, cause Darier disease. **Nat Genet.** 1999 Mar;21(3):271-7. PMID: 10080178

160. Rubio JP, Levy ER, Dobson-Stone C, Monaco AP. Genomic organization of the human galpha14 and Galphaq genes and mutation analysis in chorea-acanthocytosis (CHAC). **Genomics.** 1999 Apr 1;57(1):84-93. PMID: 10191087

*161. Francis MJ, Jones EE, Levy ER, Martin RL, Ponnambalam S, Monaco AP. Identification of a di-leucine motif within the C terminus domain of the Menkes disease protein that mediates endocytosis from the plasma membrane. **J Cell Sci.** 1999 Jun;112 (Pt 11):1721-32. PMID: 10318764

162. Szepetowski P, Monaco AP. Electronic identification and chromosomal assignment by radiation hybrid mapping of human expressed sequence tags corresponding to new potassium channel genes. **Neurogenetics.** 1999 Apr;2(2):115-20. PMID: 10369888

163. Maestrini E, Lai C, Marlow A, Matthews N, Wallace S, Bailey A, Cook EH, Weeks DE, Monaco AP. Serotonin transporter (5-HTT) and gamma-aminobutyric acid receptor subunit beta3 (GABRB3) gene polymorphisms are not associated with autism in the IMGSA families. The International Molecular Genetic Study of Autism Consortium. **Am J Med Genet.** 1999 Oct 15;88(5):492-6. PMID: 10490705

164. Goodyer ID, Jones EE, Monaco AP, Francis MJ. Characterization of the Menkes protein copper-binding domains and their role in copper-induced protein relocalization. **Hum Mol Genet.** 1999 Aug;8(8):1473-8. PMID: 10400994

*165. Maestrini E, Korge BP, Ocaña-Sierra J, Calzolari E, Cambiaghi S, Scudder PM, Hovnanian A, Monaco AP, Munro CS. A missense mutation in connexin26, D66H, causes mutilating keratoderma with sensorineural deafness (Vohwinkel's syndrome) in three unrelated families. **Hum Mol Genet.** 1999 Jul;8(7):1237-43. PMID: 10369869

166. Németh AH, Nolte D, Dunne E, Niemann S, Kostrzewa M, Peters U, Fraser E, Bochukova E, Butler R, Brown J, Cox RD, Levy ER, Ropers HH, Monaco AP, Müller U. Refined linkage disequilibrium and physical mapping of the gene locus for X-linked dystonia-parkinsonism (DYT3). **Genomics.** 1999 Sep 15;60(3):320-9. PMID: 10493831

167. Park KJ, Shin KH, Ku JL, Cho TJ, Lee SH, Choi IH, Phillippe C, Monaco AP, Porter DE, Park JG. Germline mutations in the EXT1 and EXT2 genes in Korean patients with hereditary multiple exostoses. **J Hum Genet.** 1999;44(4):230-4. PMID: 10429361

168. Dotti MT, Battisti C, Malandrini A, Federico A, Rubio JP, Ciriarello G, Monaco AP. McLeod syndrome and neuroacanthocytosis with a novel mutation in

the XK gene. **Mov Disord.** 2000 Nov;15(6):1282-4. PMID: 11104227

169. Dobson-Stone C, Cox RD, Lonie L, Southam L, Fraser M, Wise C, Bernier F, Hodgson S, Porter DE, Simpson AH, Monaco AP. Comparison of fluorescent single-strand conformation polymorphism analysis and denaturing high-performance liquid chromatography for detection of EXT1 and EXT2 mutations in hereditary multiple exostoses. **Eur J Hum Genet.** 2000 Jan;8(1):24-32. PMID: 10713884

170. Bolino A, Levy ER, Muglia M, Conforti FL, LeGuern E, Salih MA, Georgiou DM, Christodoulou RK, Hausmanowa-Petrusewicz I, Mandich P, Gambardella A, Quattrone A, Devoto M, Monaco AP. Genetic refinement and physical mapping of the CMT4B gene on chromosome 11q22. **Genomics.** 2000 Jan 15;63(2):271-8. PMID: 10673338

*171. Bolino A, Muglia M, Conforti FL, LeGuern E, Salih MA, Georgiou DM, Christodoulou K, Hausmanowa-Petrusewicz I, Mandich P, Schenone A, Gambardella A, Bono F, Quattrone A, Devoto M, Monaco AP. Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. **Nat Genet.** 2000 May;25(1):17-9. PMID: 10802647

172. Francks C, Fisher SE, Marlow AJ, Richardson AJ, Stein JF, Monaco AP. A sibling-pair based approach for mapping genetic loci that influence quantitative measures of reading disability. **Prostaglandins Leukot Essent Fatty Acids.** 2000 Jul-Aug;63(1-2):27-31. PMID: 10970709

*173. Sudbrak R, Brown J, Dobson-Stone C, Carter S, Ramser J, White J, Healy E, Dissanayake M, Larrègue M, Perrussel M, Lehrach H, Munro CS, Strachan T, Burge S, Hovnanian A, Monaco AP. Hailey-Hailey disease is caused by mutations in ATP2C1 encoding a novel Ca(2+) pump. **Hum Mol Genet.** 2000 Apr 12;9(7):1131-40. PMID: 10767338

174. Lamb JA, Moore J, Bailey A, Monaco AP. Autism: recent molecular genetic advances. **Hum Mol Genet.** 2000 Apr 12;9(6):861-8. Erratum in: Hum Mol Genet 2000 May 22;9(9):1461. PMID: 10767308

175. Maestrini E, Paul A, Monaco AP, Bailey A. Identifying autism susceptibility genes. **Neuron.** 2000 Oct;28(1):19-24. PMID: 11086979

176. Lai CS, Fisher SE, Hurst JA, Levy ER, Hodgson S, Fox M, Jeremiah S, Povey S, Jamison DC, Green ED, Vargha-Khadem F, Monaco AP. The SPCH1 region on human 7q31: genomic characterization of the critical interval and localization of translocations associated with speech and language disorder. **Am J Hum Genet.** 2000 Aug;67(2):357-68. Epub 2000 Jul 5. PMID: 10880297

178. McDonnell N, Ramser J, Francis F, Vinet MC, Rider S, Sudbrak R, Riesselman L, Yaspo ML, Reinhardt R, Monaco AP, Ross F, Kahn A, Kearney L, Buckle V, Chelly J. Characterization of a highly complex region in Xq13 and mapping of three isodicentric breakpoints associated with preleukemia. **Genomics.** 2000 Mar 15;64(3):221-9. PMID: 10756090

179. Danek A, Tison F, Rubio J, Oechsner M, Kalckreuth W, Monaco AP. The chorea of McLeod syndrome. **Mov Disord.** 2001 Sep;16(5):882-9. PMID: 11746618
180. Bolino A, Lonie LJ, Zimmer M, Boerkoel CF, Takashima H, Monaco AP, Lupski JR. Denaturing high-performance liquid chromatography of the myotubularin-related 2 gene (MTMR2) in unrelated patients with Charcot-Marie-Tooth disease suggests a low frequency of mutation in inherited neuropathy. **Neurogenetics.** 2001 Mar;3(2):107-9. PMID: 11354824
181. Marlow AJ, Fisher SE, Richardson AJ, Francks C, Talcott JB, Monaco AP, Stein JF, Cardon LR. Investigation of quantitative measures related to reading disability in a large sample of sib-pairs from the UK. **Behav Genet.** 2001 Mar;31(2):219-30. PMID: 11545538
182. Caraballo R, Pavek S, Lemainque A, Gastaldi M, Echenne B, Motte J, Genton P, Cersósimo R, Humbertclaude V, Fejerman N, Monaco AP, Lathrop MG, Rochette J, Szepetowski P. Linkage of benign familial infantile convulsions to chromosome 16p12-q12 suggests allelism to the infantile convulsions and choreoathetosis syndrome. **Am J Hum Genet.** 2001 Mar;68(3):788-94. Epub 2001 Feb 13. PMID: 11179027
- *183. Rampoldi L, Dobson-Stone C, Rubio JP, Danek A, Chalmers RM, Wood NW, Verellen C, Ferrer X, Malandrini A, Fabrizi GM, Brown R, Vance J, Pericak-Vance M, Rudolf G, Carrè S, Alonso E, Manfredi M, Németh AH, Monaco AP. A conserved sorting-associated protein is mutant in chorea-acanthocytosis. **Nat Genet.** 2001 Jun;28(2):119-20. PMID: 11381253
- *184. International Molecular Genetic Study of Autism Consortium (IMGSAC). Further characterization of the autism susceptibility locus AUTS1 on chromosome 7q. **Hum Mol Genet.** 2001 Apr 15;10(9):973-82. PMID: 11392322
- *185. Lai CS, Fisher SE, Hurst JA, Vargha-Khadem F, Monaco AP. A forkhead-domain gene is mutated in a severe speech and language disorder. **Nature.** 2001 Oct 4;413(6855):519-23. PMID: 11586359
- *186. International Molecular Genetic Study of Autism Consortium (IMGSAC). A genomewide screen for autism: strong evidence for linkage to chromosomes 2q, 7q, and 16p. **Am J Hum Genet.** 2001 Sep;69(3):570-81. Epub 2001 Jul 30. PMID: 11481586
187. Danek A, Rubio JP, Rampoldi L, Ho M, Dobson-Stone C, Tison F, Symmans WA, Oechsner M, Kalckreuth W, Watt JM, Corbett AJ, Hamdalla HH, Marshall AG, Sutton I, Dotti MT, Malandrini A, Walker RH, Daniels G, Monaco AP. McLeod neuroacanthocytosis: genotype and phenotype. **Ann Neurol.** 2001 Dec;50(6):755-64. PMID: 11761473
188. Monaco AP, Bailey AJ. Autism. The search for susceptibility genes. **Lancet.** 2001 Dec;358 Suppl:S3. PMID: 11784552
- *189. Fisher SE, Francks C, Marlow AJ, MacPhie IL, Newbury DF, Cardon LR,

Ishikawa-Brush Y, Richardson AJ, Talcott JB, Gayán J, Olson RK, Pennington BF, Smith SD, DeFries JC, Stein JF, Monaco AP. Independent genome-wide scans identify a chromosome 18 quantitative-trait locus influencing dyslexia. **Nat Genet.** 2002 Jan;30(1):86-91. Epub 2001 Dec 17. PMID: 11743577

190. Bonora E, Bacchelli E, Levy ER, Blasi F, Marlow A, Monaco AP, Maestrini E; International Molecular Genetic Study of Autism Consortium (IMGSAC). Mutation screening and imprinting analysis of four candidate genes for autism in the 7q32 region. **Mol Psychiatry.** 2002;7(3):289-301. PMID: 11920156

191. Dobson-Stone C, Fairclough R, Dunne E, Brown J, Dissanayake M, Munro CS, Strachan T, Burge S, Sudbrak R, Monaco AP, Hovnanian A. Hailey-Hailey disease: molecular and clinical characterization of novel mutations in the ATP2C1 gene. **J Invest Dermatol.** 2002 Feb;118(2):338-43. PMID: 11841554

192. Bolino A, Marigo V, Ferrera F, Loader J, Romio L, Leoni A, Di Duca M, Cinti R, Cecchi C, Feltri ML, Wrabetz L, Ravazzolo R, Monaco AP. Molecular characterization and expression analysis of Mtmr2, mouse homologue of MTMR2, the Myotubularin-related 2 gene, mutated in CMT4B. **Gene.** 2002 Jan 23;283(1-2):17-26. PMID: 11867209

*193. SLI Consortium. A genomewide scan identifies two novel loci involved in specific language impairment. **Am J Hum Genet.** 2002 Feb;70(2):384-98. Epub 2002 Jan 4. PMID: 11791209

194. Francks C, Fisher SE, Olson RK, Pennington BF, Smith SD, DeFries JC, Monaco AP. Fine mapping of the chromosome 2p12-16 dyslexia susceptibility locus: quantitative association analysis and positional candidate genes SEMA4F and OTX1. **Psychiatr Genet.** 2002 Mar;12(1):35-41. PMID: 11901358

*195. Francks C, Fisher SE, MacPhie IL, Richardson AJ, Marlow AJ, Stein JF, Monaco AP. A genomewide linkage screen for relative hand skill in sibling pairs. **Am J Hum Genet.** 2002 Mar;70(3):800-5. Epub 2002 Jan 3. Erratum in: *Am J Hum Genet* 2002 Apr;70(4):1075. PMID: 11774074

196. Fisher SE, Francks C, McCracken JT, McGough JJ, Marlow AJ, MacPhie IL, Newbury DF, Crawford LR, Palmer CG, Woodward JA, Del'Homme M, Cantwell DP, Nelson SF, Monaco AP, Smalley SL. A genomewide scan for loci involved in attention-deficit/hyperactivity disorder. **Am J Hum Genet.** 2002 May;70(5):1183-96. Epub 2002 Mar 28. PMID: 11923911

197. Newbury DF, Bonora E, Lamb JA, Fisher SE, Lai CS, Baird G, Jannoun L, Slonims V, Stott CM, Merricks MJ, Bolton PF, Bailey AJ, Monaco AP; International Molecular Genetic Study of Autism Consortium. FOXP2 is not a major susceptibility gene for autism or specific language impairment. **Am J Hum Genet.** 2002 May;70(5):1318-27. Epub 2002 Mar 13. PMID: 11894222

198. Rampoldi L, Danek A, Monaco AP. Clinical features and molecular bases of

neuroacanthocytosis. **J Mol Med.** 2002 Aug;80(8):475-91. Epub 2002 Jun 18. PMID: 12185448

199. Lamb JA, Parr JR, Bailey AJ, Monaco AP. Autism: in search of susceptibility genes. **Neuromolecular Med.** 2002;2(1):11-28. PMID: 12230302

200. Newbury DF, Monaco AP. Talking genes - the molecular basis of language impairment. **Biologist (London).** 2002 Dec;49(6):255-60. PMID: 12486301

*201. Enard W, Przeworski M, Fisher SE, Lai CS, Wiebe V, Kitano T, Monaco AP, Pääbo S. Molecular evolution of FOXP2, a gene involved in speech and language. **Nature.** 2002 Aug 22;418(6900):869-72. Epub 2002 Aug 14. PMID: 12192408

*202. Cobbold C, Ponnambalam S, Francis MJ, Monaco AP. Novel membrane traffic steps regulate the exocytosis of the Menkes disease ATPase. **Hum Mol Genet.** 2002 Nov 1;11(23):2855-66. PMID: 12393797

203. Beyer KS, Blasi F, Bacchelli E, Klauck SM, Maestrini E, Poustka A; International Molecular Genetic Study of Autism Consortium (IMGSAC). Mutation analysis of the coding sequence of the MECP2 gene in infantile autism. **Hum Genet.** 2002 Oct;111(4-5):305-9. Epub 2002 Aug 14. Erratum in: Hum Genet. 2003 Apr;112(4):436. PMID: 12384770

204. Dobson-Stone C, Danek A, Rampoldi L, Hardie RJ, Chalmers RM, Wood NW, Bohlega S, Dotti MT, Federico A, Shizuka M, Tanaka M, Watanabe M, Ikeda Y, Brin M, Goldfarb LG, Karp BI, Mohiddin S, Fananapazir L, Storch A, Fryer AE, Maddison P, Sibon I, Trevisol-Bittencourt PC, Singer C, Caballero IR, Aasly JO, Schmierer K, Dengler R, Hiersemenzel LP, Zeviani M, Meiner V, Lossos A, Johnson S, Mercado FC, Sorrentino G, Dupré N, Rouleau GA, Volkmann J, Arpa J, Lees A, Geraud G, Chouinard S, Németh A, Monaco AP. Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. **Eur J Hum Genet.** 2002 Nov;10(11):773-81. PMID: 12404112

205. Smalley SL, Kustanovich V, Minassian SL, Stone JL, Ogdie MN, McGough JJ, McCracken JT, MacPhie IL, Francks C, Fisher SE, Cantor RM, Monaco AP, Nelson SF. Genetic linkage of attention-deficit/hyperactivity disorder on chromosome 16p13, in a region implicated in autism. **Am J Hum Genet.** 2002 Oct;71(4):959-63. Epub 2002 Aug 14. PMID: 12187510

206. Dobson-Stone C, Rampoldi L, Bader B, Velayos Baeza A, Walker RH, Danek A, Monaco AP. Choreo-Acanthocytosis. In: Pagon RA, Bird TC, Dolan CR, Stephens K, editors. GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993.- 2002 Jun 14 [updated 2010 Jul 6]. PMID: 20301561

207. Newbury DF, Monaco AP. Molecular genetics of speech and language disorders. **Curr Opin Pediatr.** 2002 Dec;14(6):696-701. Review. PMID: 12436038

208. Francks C, MacPhie IL, Monaco AP. The genetic basis of dyslexia. **Lancet Neurol.** 2002 Dec;1(8):483-90. Review. PMID: 12849333

209. Bochukova EG, Jefferson A, Francis MJ, Monaco AP. Genomic studies of gene expression: regulation of the Wilson disease gene. **Genomics**. 2003 Jun;81(6):531-42. PMID: 12782122
210. Bohlega S, Al-Jishi A, Dobson-Stone C, Rampoldi L, Saha P, Murad H, Kareem A, Roberts G, Monaco AP. Chorea-acanthocytosis: clinical and genetic findings in three families from the Arabian peninsula. **Mov Disord**. 2003 Apr;18(4):403-7. PMID: 12671946
211. Francks C, DeLisi LE, Fisher SE, Laval SH, Rue JE, Stein JF, Monaco AP. Confirmatory evidence for linkage of relative hand skill to 2p12-q11. **Am J Hum Genet**. 2003 Feb;72(2):499-502. PMID: 12596796
212. Marlow AJ, Fisher SE, Francks C, MacPhie IL, Cherny SS, Richardson AJ, Talcott JB, Stein JF, Monaco AP, Cardon LR. Use of multivariate linkage analysis for dissection of a complex cognitive trait. **Am J Hum Genet**. 2003 Mar;72(3):561-70. Epub 2003 Feb 13. PMID: 12587094
213. Ogdie MN, Macphie IL, Minassian SL, Yang M, Fisher SE, Francks C, Cantor RM, McCracken JT, McGough JJ, Nelson SF, Monaco AP, Smalley SL. A genomewide scan for attention-deficit/hyperactivity disorder in an extended sample: suggestive linkage on 17p11. **Am J Hum Genet**. 2003 May;72(5):1268-79. Epub 2003 Apr 8. PMID: 12687500
- *214. Cobbold C, Coventry J, Ponnambalam S, Monaco AP. The Menkes disease ATPase (ATP7A) is internalized via a Rac1-regulated, clathrin- and caveolae-independent pathway. **Hum Mol Genet**. 2003 Jul 1;12(13):1523-33. PMID: 12812980
215. Previtali SC, Zerega B, Sherman DL, Brophy PJ, Dina G, King RH, Salih MM, Feltri L, Quattrini A, Ravazzolo R, Wrabetz L, Monaco AP, Bolino A. Myotubularin-related 2 protein phosphatase and neurofilament light chain protein, both mutated in CMT neuropathies, interact in peripheral nerve. **Hum Mol Genet**. 2003 Jul 15;12(14):1713-23. PMID: 12837694
216. Francks C, Fisher SE, Marlow AJ, MacPhie IL, Taylor KE, Richardson AJ, Stein JF, Monaco AP. Familial and genetic effects on motor coordination, laterality, and reading-related cognition. **Am J Psychiatry**. 2003 Nov;160(11):1970-7. Erratum in: *Am J Psychiatry*. 2004 Jan;161(1):185. PMID: 14594743
217. Bonora E, Beyer KS, Lamb JA, Parr JR, Klauck SM, Benner A, Paolucci M, Abbott A, Ragoussis I, Poustka A, Bailey AJ, Monaco AP; International Molecular Genetic Study of Autism (IMGSAC). Analysis of reelin as a candidate gene for autism. **Mol Psychiatry**. 2003 Oct;8(10):885-92. PMID: 14515139
218. Bacchelli E, Blasi F, Biondolillo M, Lamb JA, Bonora E, Barnby G, Parr J, Beyer KS, Klauck SM, Poustka A, Bailey AJ, Monaco AP, Maestrini E; International Molecular Genetic Study of Autism Consortium (IMGSAC). Screening of nine candidate genes for autism on chromosome 2q reveals rare nonsynonymous variants in the cAMP-GEFII gene. **Mol Psychiatry**. 2003 Nov;8(11):916-24. PMID: 14593429

219. Fisher SE, Lai CS, Monaco AP. Deciphering the genetic basis of speech and language disorders. **Annu Rev Neurosci.** 2003;26:57-80. Epub 2003 Jan 8. PMID: 12524432
- *220. Lai CS, Gerrelli D, Monaco AP, Fisher SE, Copp AJ. FOXP2 expression during brain development coincides with adult sites of pathology in a severe speech and language disorder. **Brain.** 2003 Nov;126(Pt 11):2455-62. Epub 2003 Jul 22. PMID: 12876151
221. Francks C, DeLisi LE, Shaw SH, Fisher SE, Richardson AJ, Stein JF, Monaco AP. Parent-of-origin effects on handedness and schizophrenia susceptibility on chromosome 2p12-q11. **Hum Mol Genet.** 2003 Dec 15;12(24):3225-30. Epub 2003 Oct 28. PMID: 14583442
222. Barnby G, Monaco AP. Strategies for autism candidate gene analysis. **Novartis Found Symp.** 2003;251:48-63; discussion 63-9, 109-11, 281-97. PMID: 14521187
223. Cobbold C, Monaco AP, Sivaprasadarao A, Ponnambalam S. Aberrant trafficking of transmembrane proteins in human disease. **Trends Cell Biol.** 2003 Dec;13(12):639-47. PMID: 14624842
224. Cobbold C, Coventry J, Ponnambalam S, Monaco AP. Actin and microtubule regulation of trans-Golgi network architecture, and copper-dependent protein transport to the cell surface. **Mol Membr Biol.** 2004 Jan-Feb;21(1):59-66. PMID: 14668139
225. Loo SK, Fisher SE, Francks C, Ogdie MN, MacPhie IL, Yang M, McCracken JT, McGough JJ, Nelson SF, Monaco AP, Smalley SL. Genome-wide scan of reading ability in affected sibling pairs with attention-deficit/hyperactivity disorder: unique and shared genetic effects. **Mol Psychiatry.** 2004 May;9(5):485-93. PMID: 14625563
226. Scerri TS, Fisher SE, Francks C, MacPhie IL, Paracchini S, Richardson AJ, Stein JF, Monaco AP. Putative functional alleles of DYX1C1 are not associated with dyslexia susceptibility in a large sample of sibling pairs from the UK. **J Med Genet.** 2004 Nov;41(11):853-7. PMID: 15520411
- *227. SLI Consortium (SLIC). Highly significant linkage to the SLI1 locus in an expanded sample of individuals affected by specific language impairment. **Am J Hum Genet.** 2004 Jun;74(6):1225-38. Epub 2004 May 3. PMID: 15133743
- *228. Francks C, Paracchini S, Smith SD, Richardson AJ, Scerri TS, Cardon LR, Marlow AJ, MacPhie IL, Walter J, Pennington BF, Fisher SE, Olson RK, DeFries JC, Stein JF, Monaco AP. A 77-kilobase region of chromosome 6p22.2 is associated with dyslexia in families from the United Kingdom and from the United States. **Am J Hum Genet.** 2004 Dec;75(6):1046-58. Epub 2004 Oct 22. PMID: 15514892
229. Velayos-Baeza A, Vettori A, Copley RR, Dobson-Stone C, Monaco AP. Analysis of the human VPS13 gene family. **Genomics.** 2004 Sep;84(3):536-49. PMID: 15498460
230. Dobson-Stone C, Velayos-Baeza A, Filippone LA, Westbury S, Storch A, Erdmann T, Wroe SJ, Leenders KL, Lang AE, Dotti MT, Federico A, Mohiddin SA, Fananapazir L, Daniels G, Danek A, Monaco AP. Chorein detection for the diagnosis of chorea-

acanthocytosis. **Ann Neurol.** 2004 Aug;56(2):299-302. PMID: 15293285

231. D'Adamo P, Bacchelli E, Blasi F, Lipp HP, Toniolo D, Maestrini E. and the International Molecular Genetic Study of Autism Consortium (IMGSAC). DNA variants in the human RAB3A gene are not associated with autism. **Genes Brain Behav.** 2004 Apr;3(2):123-4. PMID: 15005721.

232. Porter DE, Lonie L, Fraser M, Dobson-Stone C, Porter JR, Monaco AP, Simpson AH. Severity of disease and risk of malignant change in hereditary multiple exostoses. A genotype-phenotype study. **J Bone Joint Surg Br.** 2004 Sep;86(7):1041-6. PMID: 15446535

*233. Lamb JA, Barnby G, Bonora E, Sykes N, Bacchelli E, Blasi F, Maestrini E, Broxholme J, Tzenova J, Weeks D, Bailey AJ, Monaco AP; International Molecular Genetic Study of Autism Consortium (IMGSAC). Analysis of IMGSAC autism susceptibility loci: evidence for sex limited and parent of origin specific effects. **J Med Genet.** 2005 Feb;42(2):132-7. PMID: 15689451

234. Lossos A, Dobson-Stone C, Monaco AP, Soffer D, Rahamim E, Newman JP, Mohiddin S, Fananapazir L, Lerer I, Linetsky E, Reches A, Argov Z, Abramsky O, Gadoth N, Sadeh M, Gomori JM, Boher M, Meiner V. Early clinical heterogeneity in choreoacanthocytosis. **Arch Neurol.** 2005 Apr;62(4):611-4. PMID: 15824261

235. Gayán J, Willcutt EG, Fisher SE, Francks C, Cardon LR, Olson RK, Pennington BF, Smith SD, Monaco AP, DeFries JC. Bivariate linkage scan for reading disability and attention-deficit/hyperactivity disorder localizes pleiotropic loci. **J Child Psychol Psychiatry.** 2005 Oct;46(10):1045-56. PMID: 16178928

236. Bonora E, Lamb JA, Barnby G, Sykes N, Moberly T, Beyer KS, Klauck SM, Poustka F, Bacchelli E, Blasi F, Maestrini E, Battaglia A, Haracopos D, Pedersen L, Isager T, Eriksen G, Viskum B, Sorensen EU, Brondum-Nielsen K, Cotterill R, Engeland H, Jonge M, Kemner C, Steggehuis K, Scherpenisse M, Rutter M, Bolton PF, Parr JR, Poustka A, Bailey AJ, Monaco AP; International Molecular Genetic Study of Autism Consortium. Mutation screening and association analysis of six candidate genes for autism on chromosome 7q. **Eur J Hum Genet.** 2005 Feb;13(2):198-207. PMID: 15523497

237. Barnby G, Abbott A, Sykes N, Morris A, Weeks DE, Mott R, Lamb J, Bailey AJ, Monaco AP; International Molecular Genetics Study of Autism Consortium. Candidate-gene screening and association analysis at the autism-susceptibility locus on chromosome 16p: evidence of association at GRIN2A and ABAT. **Am J Hum Genet.** 2005 Jun;76(6):950-66. Epub 2005 Apr 13. PMID: 15830322

238. Dobson-Stone C, Velayos-Baeza A, Jansen A, Andermann F, Dubeau F, Robert F, Summers A, Lang AE, Chouinard S, Danek A, Andermann E, Monaco AP. Identification of a VPS13A founder mutation in French Canadian families with chorea-acanthocytosis. **Neurogenetics.** 2005 Sep;6(3):151-8. Epub 2005 Sep 28. PMID: 15918062

239. Al-Asmi A, Jansen AC, Badhwar A, Dubeau F, Tampieri D, Shustik C, Mercho S, Savard G, Dobson-Stone C, Monaco AP, Andermann F, Andermann E. Familial

- temporal lobe epilepsy as a presenting feature of choreoacanthocytosis. **Epilepsia**. 2005 Aug;46(8):1256-63. PMID: 16060937
240. MacDermot KD, Bonora E, Sykes N, Coupe AM, Lai CS, Vernes SC, Vargha-Khadem F, McKenzie F, Smith RL, Monaco AP, Fisher SE. Identification of FOXP2 truncation as a novel cause of developmental speech and language deficits. **Am J Hum Genet**. 2005 Jun;76(6):1074-80. Epub 2005 Apr 22. PMID: 15877281
241. Newbury DF, Bishop DV, Monaco AP. Genetic influences on language impairment and phonological short-term memory. **Trends Cogn Sci**. 2005 Nov;9(11):528-34. Epub 2005 Sep 26. PMID: 16188486
242. Blasi F, Bacchelli E, Carone S, Toma C, Monaco AP, Bailey AJ, Maestrini E; International Molecular Genetic Study of Autism Consortium (IMGSAC). SLC25A12 and CMYA3 gene variants are not associated with autism in the IMGSAC multiplex family sample. **Eur J Hum Genet**. 2006 Jan;14(1):123-6. PMID: 16205742
243. Ogdie MN, Bakker SC, Fisher SE, Francks C, Yang MH, Cantor RM, Loo SK, van der Meulen E, Pearson P, Buitelaar J, Monaco A, Nelson SF, Sinke RJ, Smalley SL. Pooled genome-wide linkage data on 424 ADHD ASPs suggests genetic heterogeneity and a common risk locus at 5p13. **Mol Psychiatry**. 2006 Jan;11(1):5-8. PMID: 16205734
244. Howell GJ, Holloway ZG, Cobbold C, Monaco AP, Ponnambalam S. Cell biology of membrane trafficking in human disease. **Int Rev Cytol**. 2006;252:1-69. PMID: 16984815
245. Paracchini S, Thomas A, Castro S, Lai C, Paramasivam M, Wang Y, Keating BJ, Taylor JM, Hacking DF, Scerri T, Francks C, Richardson AJ, Wade-Martins R, Stein JF, Knight JC, Copp AJ, Loturco J, Monaco AP. The chromosome 6p22 haplotype associated with dyslexia reduces the expression of KIAA0319, a novel gene involved in neuronal migration. **Hum Mol Genet**. 2006 May 15;15(10):1659-66. Epub 2006 Apr 6. PMID: 16600991
246. Blasi F, Bacchelli E, Pesaresi G, Carone S, Bailey AJ, Maestrini E; International Molecular Genetic Study of Autism Consortium (IMGSAC). Absence of coding mutations in the X-linked genes neuroligin 3 and neuroligin 4 in individuals with autism from the IMGSAC collection. **Am J Med Genet B Neuropsychiatr Genet**. 2006 Apr 5;141(3):220-1. PMID: 16508939
247. Harold D, Paracchini S, Scerri T, Dennis M, Cope N, Hill G, Moskvina V, Walter J, Richardson AJ, Owen MJ, Stein JF, Green ED, O'Donovan MC, Williams J, Monaco AP. Further evidence that the KIAA0319 gene confers susceptibility to developmental dyslexia. **Mol Psychiatry**. 2006 Dec;11(12):1085-91, 1061. Epub 2006 Oct 10. PMID: 17033633
248. Parr JR, Lamb JA, Bailey AJ, Monaco AP. Response to paper by Molloy et al.: linkage on 21q and 7q in autism subset with regression. **Mol Psychiatry**. 2006 Jul;11(7):617-9; author reply 619. PMID: 16801976

249. Flint JA, Monaco AP. Focus on behavioural genetics. **Eur J Hum Genet** 2006; 14(6):647-648.
250. Lonie L, Porter DE, Fraser M, Cole T, Wise C, Yates L, Wakeling E, Blair E, Morava E, Monaco AP, Ragoussis J. Determination of the mutation spectrum of the EXT1/EXT2 genes in British Caucasian patients with multiple osteochondromas, and exclusion of six candidate genes in EXT negative cases. **Hum Mutat.** 2006 Nov;27(11):1160. PMID: 17041877
251. Paracchini S, Scerri T, Monaco AP. The Genetic Lexicon of Dyslexia. **Annu Rev Genomics Hum Genet.** 2007; 8:57-79. PMID: 17444811
- 252*. Autism Genome Project Consortium; Szatmari P, Paterson AD, Zwaigenbaum L, Roberts W, Brian J, Liu XQ, Vincent JB, Skaug JL, Thompson AP, Senman L, Feuk L, Qian C, Bryson SE, Jones MB, Marshall CR, Scherer SW, Vieland VJ, Bartlett C, Mangin LV, Goedken R, Segre A, Pericak-Vance MA, Cuccaro ML, Gilbert JR, Wright HH, Abramson RK, Betancur C, Bourgeron T, Gillberg C, Leboyer M, Buxbaum JD, Davis KL, Hollander E, Silverman JM, Hallmayer J, Lotspeich L, Sutcliffe JS, Haines JL, Folstein SE, Piven J, Wassink TH, Sheffield V, Geschwind DH, Bucan M, Brown WT, Cantor RM, Constantino JN, Gilliam TC, Herbert M, Lajonchere C, Ledbetter DH, Lese-Martin C, Miller J, Nelson S, Samango-Sprouse CA, Spence S, State M, Tanzi RE, Coon H, Dawson G, Devlin B, Estes A, Flodman P, Klei L, McMahon WM, Minshew N, Munson J, Korvatska E, Rodier PM, Schellenberg GD, Smith M, Spence MA, Stodgell C, Tepper PG, Wijsman EM, Yu CE, Roge B, Mantoulan C, Wittemeyer K, Poustka A, Felder B, Klauck SM, Schuster C, Poustka F, Bolte S, Feineis-Matthews S, Herbrecht E, Schmotzer G, Tsiantis J, Papanikolaou K, Maestrini E, Bacchelli E, Blasi F, Carone S, Toma C, Van Engeland H, de Jonge M, Kemner C, Koop F, Langemeijer M, Hijimans C, Staal WG, Baird G, Bolton PF, Rutter ML, Weisblatt E, Green J, Aldred C, Wilkinson JA, Pickles A, Le Couteur A, Berney T, McConachie H, Bailey AJ, Francis K, Honeyman G, Hutchinson A, Parr JR, Wallace S, Monaco AP, Barnby G, Kobayashi K, Lamb JA, Sousa I, Sykes N, Cook EH, Guter SJ, Leventhal BL, Salt J, Lord C, Corsello C, Hus V, Weeks DE, Volkmar F, Tauber M, Fombonne E, Shih A. Mapping autism risk loci using genetic linkage and chromosomal rearrangements. **Nat Genet.** 2007 Mar;39(3):319-28. Epub 2007 Feb 18. PMID: 17322880.
253. Monaco AP. The SLI Consortium. Multivariate linkage analysis of specific language impairment (SLI). **Ann Hum Genet.** 2007 Sep;71(Pt 5):660-73. Epub 2007 Mar 27. PMID: 17388790
- 254*. Francks C, Maegawa S, Laurén J, Abrahams BS, Velayos-Baeza A, Medland SE, Colella S, Groszer M, McAuley EZ, Caffrey TM, Timmusk T, Pruunsild P, Koppel I, Lind PA, Matsumoto-Itaba N, Nicod J, Xiong L, Jooper R, Enard W, Krinsky B, Nanba E, Richardson AJ, Riley BP, Martin NG, Strittmatter SM, Möller HJ, Rujescu D, St Clair D, Muglia P, Roos JL, Fisher SE, Wade-Martins R, Rouleau GA, Stein JF, Karayiorgou M, Geschwind DH, Ragoussis J, Kendler KS, Airaksinen MS, Oshimura M, DeLisi LE, Monaco AP. LRR11 on chromosome 2p12 is a maternally suppressed gene that is associated paternally with handedness and schizophrenia. **Mol Psychiatry.** 2007 Dec;12(12):1129-39, 1057. Epub 2007 Jul 31. PMID: 17667961

Molecular Psychiatry cover image: Francks C, Maegawa S, Laurén J, Abrahams BS, Velayos-Baeza A, Medland SE, Colella S, Groszer M, McAuley EZ, Caffrey TM, Timmusk T, Pruunsild P, Koppel I, Lind PA, Matsumoto-Itaba N, Nicod J, Xiong L, Joobor R, Enard W, Krinsky B, Nanba E, Richardson AJ, Riley BP, Martin NG, Strittmatter SM, Möller HJ, Rujescu D, St Clair D, Muglia P, Roos JL, Fisher SE, Wade-Martins R, Rouleau GA, Stein JF, Karayiorgou M, Geschwind DH, Ragoussis J, Kendler KS, Airaksinen MS, Oshimura M, Delisi LE, Monaco AP. LRRTM1 protein is located in the endoplasmic reticulum (ER) in mammalian cells. **Mol Psychiatry**. 2007 Dec;12(12):1057. PMID: 18043708

255. Velayos-Baeza A, Toma C, da Roza S, Paracchini S, Monaco AP. Alternative splicing in the dyslexia-associated gene KIAA0319. **Mamm Genome**. 2007 Sep;18(9):627-634. PMID: 17846832

256. Toma C, Rossi M, Sousa I, Blasi F, Bacchelli E, Alen R, Vanhala R, Monaco AP, Järvelä I, Maestrini E; International Molecular Genetic Study of Autism Consortium. Is ASMT a susceptibility gene for autism spectrum disorders? A replication study in European populations. **Mol Psychiatry**. 2007 Nov;12(11):977-9. PMID: 17957233

257. Holloway ZG, Grabski R, Szul T, Styers ML, Coventry JA, Monaco AP, Sztul E. Activation of ADP-ribosylation factor regulates biogenesis of the ATP7A-containing trans-Golgi network compartment and its Cu-induced trafficking. **Am J Physiol Cell Physiol**. 2007 Dec;293(6):C1753-67. Epub 2007 Oct 3. PMID: 17913844

258. Ruiz-Sandoval JL, García-Navarro V, Chiquete E, Dobson-Stone C, Monaco AP, Alvarez-Palazuelos LE, Padilla-Martínez JJ, Barrera-Chairez E, Rodríguez-Figueroa EI, Pérez-García G. Choreoacanthocytosis in a Mexican family. **Arch Neurol**. 2007 Nov;64(11):1661-4. PMID: 17998451

Publications as Pro-Vice-Chancellor (Planning and Resources):

259. Falcaro M, Pickles A, Newbury DF, Addis L, Banfield E, Fisher SE, Monaco AP, Simkin Z, Conti-Ramsden G; The SLI Consortium. Genetic and phenotypic effects of phonological short-term memory and grammatical morphology in specific language impairment. **Genes Brain Behav**. 2008 Jun;7(4):393-402. Epub 2007 Nov 12. PMID 18005161

260. Velayos-Baeza A, Toma C, Paracchini S, Monaco AP. The dyslexia-associated gene KIAA0319 encodes highly N- and O-glycosylated plasma membrane and secreted isoforms. **Hum Mol Genet**. 2008 Mar 15;17(6):859-71. Epub 2007 Dec 6. PMID: 18063668

261. Murphy JE, Vohra RS, Dunn S, Holloway ZG, Monaco AP, Homer-Vanniasinkam S, Walker JH, Ponnambalam S. Oxidised LDL internalisation by the LOX-1 scavenger receptor is dependent on a novel cytoplasmic motif and is regulated by dynamin-2. **J Cell Sci**. 2008 Jul 1;121(Pt 13):2136-47. Epub 2008 Jun 10. PMID: 18544637

262. Liu XQ, Paterson AD, Szatmari P; Autism Genome Project Consortium. Genome-wide linkage analyses of quantitative and categorical autism subphenotypes. **Biol Psychiatry**. 2008 Oct 1;64(7):561-70. Epub 2008 Jul 16. PMID: 18632090

- 263*. Paracchini S, Steer CD, Buckingham LL, Morris AP, Ring S, Scerri T, Stein J, Pembrey ME, Ragoussis J, Golding J, Monaco AP. Association of the KIAA0319 Dyslexia Susceptibility Gene With Reading Skills in the General Population. **Am J Psychiatry**. 2008 Dec;165(12):1576-84. Epub 2008 Oct 1. PMID: 18829873
- 264*. Vernes SC, Newbury DF, Abrahams BS, Winchester L, Nicod J, Groszer M, Alarcón M, Oliver PL, Davies KE, Geschwind DH, Monaco AP, Fisher SE. A Functional Genetic Link between Distinct Developmental Language Disorders. **N Engl J Med**. 2008 Nov 27;359(22):2337-45. Epub 2008 Nov 5. PMID: 18987363
265. Sousa I, Clark TG, Toma C, Kobayashi K, Choma M, Holt R, Sykes NH, Lamb JA, Bailey AJ, Battaglia A, Maestrini E, Monaco AP. MET and autism susceptibility: family and case-control studies. **Eur J Hum Genet**. 2009 Jun;17(6):749-58. Epub 2008 Nov 12. PMID: 19002214
266. Pagnamenta AT, Wing K, Akha ES, Knight SJ, Bölte S, Schmötzer G, Duketis E, Poustka F, Klauck SM, Poustka A, Ragoussis J, Bailey AJ, Monaco AP. A 15q13.3 microdeletion segregating with autism. **Eur J Hum Genet**. 2009 May;17(5):687-92. Epub 2008 Dec 3. PMID: 19050728
267. Asher JE, Lamb JA, Brocklebank D, Cazier JB, Maestrini E, Addis L, Sen M, Baron-Cohen S, Monaco AP. A Whole-Genome Scan and Fine-Mapping Linkage Study of Auditory-Visual Synesthesia Reveals Evidence of Linkage to Chromosomes 2q24, 5q33, 6p12, and 12p12. **Am J Hum Genet**. 2009 Feb;84(2):279-85. Epub 2009 Feb 5. PMID: 19200526
268. Newbury DF, Warburton PC, Wilson N, Bacchelli E, Carone S, Lamb JA, Maestrini E, Volpi EV, Mohammed S, Baird G, Monaco AP; The International Molecular Genetic Study of Autism Consortium (IMGSAC). Mapping of partially overlapping de novo deletions across an autism susceptibility region (AUTS5) in two unrelated individuals affected by developmental delays with communication impairment. **Am J Med Genet A**. 2009 Feb 15;149A(4):588-597, PMID: 19267418
- 269*. Dennis MY, Paracchini S, Scerri TS, Prokunina-Olsson L, Knight JC, Wade-Martins R, Coggill P, Beck S, Green ED, Monaco AP. A Common Variant Associated with Dyslexia Reduces Expression of the KIAA0319 Gene. **PLoS Genet**. 2009 Mar;5(3):e1000436. Epub 2009 Mar 27. PMID: 19325871
270. Vernes SC, Macdermot KD, Monaco AP, Fisher SE. Assessing the impact of FOXP1 mutations on developmental verbal dyspraxia. **Eur J Hum Genet**. 2009 Oct;17(10):1354-8. Epub 2009 Apr 8. PMID: 19352412
271. Sykes NH, Toma C, Wilson N, Volpi EV, Sousa I, Pagnamenta AT, Tancredi R, Battaglia A, Maestrini E, Bailey AJ, Monaco AP. Copy number variation and association analysis of SHANK3 as a candidate gene for autism in the IMGSAC collection. **Eur J Hum Genet**. 2009 Oct;17(10):1347-53. Epub 2009 Apr 22. PMID: 19384346

- 272*. Maestrini E, Pagnamenta AT, Lamb JA, Bacchelli E, Sykes NH, Sousa I, Toma C, Barnby G, Butler H, Winchester L, Scerri TS, Minopoli F, Reichert J, Cai G, Buxbaum JD, Korvatska O, Schellenberg GD, Dawson G, Bildt AD, Minderaa RB, Mulder EJ, Morris AP, Bailey AJ, Monaco AP. High-density SNP association study and copy number variation analysis of the AUTS1 and AUTS5 loci implicate the IMMP2L-DOCK4 gene region in autism susceptibility. **Mol Psychiatry**. 2010 Sep;15(9):954-68. Epub 2009 Apr 28. PMID: 19401682
273. Levecque C, Velayos-Baeza A, Holloway ZG, Monaco AP. The dyslexia-associated protein KIAA0319 interacts with Adaptor Protein 2 and follows the classical clathrin-mediated endocytosis pathway. **Am J Physiol Cell Physiol**. 2009 Jul;297(1):C160-8. Epub 2009 May 6. PMID: 19419997
- 274*. Newbury DF, Winchester L, Addis L, Paracchini S, Buckingham LL, Clark A, Cohen W, Cowie H, Dworzynski K, Everitt A, Goodyer IM, Hennessy E, Kindley AD, Miller LL, Nasir J, O'Hare A, Shaw D, Simkin Z, Simonoff E, Slonims V, Watson J, Ragoussis J, Fisher SE, Seckl JR, Helms PJ, Bolton PF, Pickles A, Conti-Ramsden G, Baird G, Bishop DV, Monaco AP. CMIP and ATP2C2 Modulate Phonological Short-Term Memory in Language Impairment. **Am J Hum Genet**. 2009 Aug;85(2):264-72. Epub 2009 Jul 30 PMID: 19646677
275. Pagnamenta, A and Monaco, A, Chromosomal Copy Number Variation in Psychiatric Disorders, **European Psychiatric Review** 2009 2(1): 8-12
276. Weiss LA, Arking DE & The Gene Discovery Project of Johns Hopkins & the Autism Consortium A genome-wide linkage and association scan reveals novel loci for autism. **Nature** 2009 Oct 8;461(7265):802-8. PMID: 19812673
277. Newbury DF, Fisher SE, Monaco AP. Recent advances in the genetics of language impairment. **Genome Med**. 2010 Jan 26;2(1):6. PMID: 20193051.
278. Addis L, Friederici AD, Kotz SA, Sabisch B, Barry J, Richter N, Ludwig AA, Räsänen R, Albert FW, Pääbo S, Newbury DF, Monaco AP. A locus for an auditory processing deficit and language impairment in an extended pedigree maps to 12p13.31-q14.3. **Genes Brain Behav**. 2010 Aug;9(6):545-61. Epub Mar 25. PMID: 20345892.
279. Pagnamenta AT, Bacchelli E, de Jonge MV, Mirza G, Scerri TS, Minopoli F, Chiocchetti A, Ludwig KU, Hoffmann P, Paracchini S, Lowy E, Harold DH, Chapman JA, Klauck SM, Poustka F, Houben RH, Staal WG, Ophoff RA, O'Donovan MC, Williams J, Nöthen MM, Schulte-Körne G, Deloukas P, Ragoussis J, Bailey AJ, Maestrini E, Monaco AP; International Molecular Genetic Study Of Autism Consortium. Characterization of a Family with Rare Deletions in CNTNAP5 and DOCK4 Suggests Novel Risk Loci for Autism and Dyslexia. **Biol Psychiatry**. 2010 Aug 15;68(4):320-328. Epub 2010 Mar 26. PMID: 20346443
280. Holt R, Barnby G, Maestrini E, Bacchelli E, Brocklebank D, Sousa I, Mulder EJ, Kantojarvi K, Jarvela I, Klauck SM, Poustka F, Bailey AJ, Monaco AP. Linkage and candidate gene studies of autism spectrum disorders in European populations. **Eur J Hum Genet**. 2010 Sep;18(9):1013-9. Epub 2010 May 5. Erratum in: Eur J Hum Genet. 2010

Sep;18(9):1020. Parr, Jeremy [added]. PubMed PMID: 20442744.

281*. Pinto D, Pagnamenta AT, Klei L, Anney R, Merico D, Regan R, Conroy J, Magalhaes TR, Correia C, Abrahams BS, Almeida J, Bacchelli E, Bader GD, Bailey AJ, Baird G, Battaglia A, Berney T, Bolshakova N, Bölte S, Bolton PF, Bourgeron T, Brennan S, Brian J, Bryson SE, Carson AR, Casallo G, Casey J, Chung BH, Cochrane L, Corsello C, Crawford EL, Crossett A, Cytrynbaum C, Dawson G, de Jonge M, Delorme R, Drmic I, Duketis E, Duque F, Estes A, Farrar P, Fernandez BA, Folstein SE, Fombonne E, Freitag CM, Gilbert J, Gillberg C, Glessner JT, Goldberg J, Green A, Green J, Guter SJ, Hakonarson H, Heron EA, Hill M, Holt R, Howe JL, Hughes G, Hus V, Iglizzi R, Kim C, Klauck SM, Kolevzon A, Korvatska O, Kustanovich V, Lajonchere CM, Lamb JA, Laskawiec M, Leboyer M, Le Couteur A, Leventhal BL, Lionel AC, Liu XQ, Lord C, Lotspeich L, Lund SC, Maestrini E, Mahoney W, Mantoulan C, Marshall CR, McConachie H, McDougle CJ, McGrath J, McMahon WM, Merikangas A, Migita O, Minshew NJ, Mirza GK, Munson J, Nelson SF, Noakes C, Noor A, Nygren G, Oliveira G, Papanikolaou K, Parr JR, Parrini B, Paton T, Pickles A, Pilorge M, Piven J, Ponting CP, Posey DJ, Poustka A, Poustka F, Prasad A, Ragoussis J, Renshaw K, Rickaby J, Roberts W, Roeder K, Roge B, Rutter ML, Bierut LJ, Rice JP, Salt J, Sansom K, Sato D, Segurado R, Sequeira AF, Senman L, Shah N, Sheffield VC, Soorya L, Sousa I, Stein O, Sykes N, Stoppioni V, Strawbridge C, Tancredi R, Tansey K, Thiruvahindrapduram B, Thompson AP, Thomson S, Tryfon A, Tsiantis J, Van Engeland H, Vincent JB, Volkmar F, Wallace S, Wang K, Wang Z, Wassink TH, Webber C, Weksberg R, Wing K, Wittmeyer K, Wood S, Wu J, Yaspan BL, Zurawiecki D, Zwaigenbaum L, Buxbaum JD, Cantor RM, Cook EH, Coon H, Cuccaro ML, Devlin B, Ennis S, Gallagher L, Geschwind DH, Gill M, Haines JL, Hallmayer J, Miller J, Monaco AP, Nurnberger JI Jr, Paterson AD, Pericak-Vance MA, Schellenberg GD, Szatmari P, Vicente AM, Vieland VJ, Wijsman EM, Scherer SW, Sutcliffe JS, Betancur C. Functional impact of global rare copy number variation in autism spectrum disorders. **Nature**. 2010 Jul 15;466(7304):368-72. Epub 2010 Jun 9. PubMed PMID: 20531469.

282*. Anney R, Klei L, Pinto D, Regan R, Conroy J, Magalhaes TR, Correia C, Abrahams BS, Sykes N, Pagnamenta AT, Almeida J, Bacchelli E, Bailey AJ, Baird G, Battaglia A, Berney T, Bolshakova N, Bölte S, Bolton PF, Bourgeron T, Brennan S, Brian J, Carson AR, Casallo G, Casey J, Chu S, Cochrane L, Corsello C, Crawford EL, Crossett A, Dawson G, de Jonge M, Delorme R, Drmic I, Duketis E, Duque F, Estes A, Farrar P, Fernandez BA, Folstein SE, Fombonne E, Freitag CM, Gilbert J, Gillberg C, Glessner JT, Goldberg J, Green A, Green J, Guter SJ, Hakonarson H, Heron EA, Hill M, Holt R, Howe JL, Hughes G, Hus V, Iglizzi R, Kim C, Klauck SM, Kolevzon A, Korvatska O, Kustanovich V, Lajonchere CM, Lamb JA, Laskawiec M, Leboyer M, Le Couteur A, Leventhal BL, Lionel AC, Liu XQ, Lord C, Lotspeich L, Lund SC, Maestrini E, Mahoney W, Mantoulan C, Marshall CR, McConachie H, McDougle CJ, McGrath J, McMahon WM, Melhem NM, Merikangas A, Migita O, Minshew NJ, Mirza GK, Munson J, Nelson SF, Noakes C, Noor A, Nygren G, Oliveira G, Papanikolaou K, Parr JR, Parrini B, Paton T, Pickles A, Piven J, Posey DJ, Poustka A, Poustka F, Prasad A, Ragoussis J, Renshaw K, Rickaby J, Roberts W, Roeder K, Roge B, Rutter ML, Bierut LJ, Rice JP, Salt J, Sansom K, Sato D, Segurado R, Senman L, Shah N, Sheffield VC, Soorya L, Sousa I, Stoppioni V, Strawbridge C, Tancredi R, Tansey K, Thiruvahindrapduram B, Thompson AP, Thomson S, Tryfon A, Tsiantis J, Van Engeland H, Vincent JB, Volkmar F, Wallace S, Wang K, Wang Z, Wassink TH, Wing K, Wittmeyer K, Wood S, Yaspan BL, Zurawiecki D, Zwaigenbaum L, Betancur C,

Buxbaum JD, Cantor RM, Cook EH, Coon H, Cuccaro ML, Gallagher L, Geschwind DH, Gill M, Haines JL, Miller J, Monaco AP, Nurnberger JI Jr, Paterson AD, Pericak-Vance MA, Schellenberg GD, Scherer SW, Sutcliffe JS, Szatmari P, Vicente AM, Vieland VJ, Wijsman EM, Devlin B, Ennis S, Hallmayer J. A genomewide scan for common alleles affecting risk for autism. **Hum Mol Genet.** 2010 Oct 15;19(20):4072-82. Epub 2010 Jul 27. PMID: 20663923

283. Sousa I, Clark TG, Holt R, Pagnamenta AT, Mulder EJ, Minderaa RB, Bailey AJ, Battaglia A, Klauck SM, Poustka F, Monaco AP; International Molecular Genetic Study of Autism Consortium (IMGSAC). Polymorphisms in leucine-rich repeat genes are associated with autism spectrum disorder susceptibility in populations of European ancestry. **Mol Autism.** 2010 Mar 25;1(1):7. PMID: 20678249, Central PMCID: PMC2913944.

284. Horn D, Kapeller J, Rivera-Brugués N, Moog U, Lorenz-Depiereux B, Eck S, Hempel M, Wagenstaller J, Gawthrop A, Monaco AP, Bonin M, Riess O, Wohlleber E, Illig T, Bezzina CR, Franke A, Spranger S, Villavicencio-Lorini P, Seifert W, Rosenfeld J, Klopocki E, Rappold GA, Strom TM. Identification of FOXP1 deletions in three unrelated patients with mental retardation and significant speech and language deficits. **Hum Mutat.** 2010 Nov;31(11):E1851-60. PMID: 20848658

285. Paracchini S, Ang QW, Stanley FJ, Monaco AP, Pennell CE, Whitehouse AJ. Analysis of dyslexia candidate genes in the Raine cohort representing the general Australian population. **Genes Brain Behav.** 2010 Mar;10(2):158-65. doi: 10.1111/j.1601-183X.2010.00651.x. Epub 2010 Oct 19. PMID: 20846247

286. Noor A, Whibley A, Marshall CR, Gianakopoulos PJ, Piton A, Carson AR, Orlic-Milacic M, Lionel AC, Sato D, Pinto D, Drmic I, Noakes C, Senman L, Zhang X, Mo R, Gauthier J, Crosbie J, Pagnamenta AT, Munson J, Estes AM, Fiebig A, Franke A, Schreiber S, Stewart AF, Roberts R, McPherson R, Guter SJ, Cook EH Jr, Dawson G, Schellenberg GD, Battaglia A, Maestrini E; Autism Genome Project Consortium, Jeng L, Hutchison T, Rajcan-Separovic E, Chudley AE, Lewis SM, Liu X, Holden JJ, Fernandez B, Zwaigenbaum L, Bryson SE, Roberts W, Szatmari P, Gallagher L, Stratton MR, Gecz J, Brady AF, Schwartz CE, Schachar RJ, Monaco AP, Rouleau GA, Hui CC, Lucy Raymond F, Scherer SW, Vincent JB. Disruption at the PTCHD1 Locus on Xp22.11 in Autism Spectrum Disorder and Intellectual Disability. **Sci Transl Med.** 2010 Sep 15;2(49):49ra68. PMID: 20844286

287. Newbury DF, Monaco AP. Genetic advances in the study of speech and language disorders. **Neuron.** 2010 Oct 21;68(2):309-20. PMID: 20955937

288*. Velayos-Baeza A, Levecque C, Kobayashi K, Holloway ZG, Monaco AP. The dyslexia-associated KIAA0319 protein undergoes proteolytic processing with {gamma}-secretase-independent intramembrane cleavage. **J Biol Chem.** 2010 Dec 17;285(51):40148-62. Epub 2010 Oct 13. PMID: 20943657

289. Pagnamenta AT, Khan H, Walker S, Gerrelli D, Wing K, Bonaglia MC, Giorda R, Berney T, Mani E, Molteni M, Pinto D, Le Couteur A, Hallmayer J, Sutcliffe JS, Szatmari P, Paterson AD, Scherer SW, Vieland VJ, Monaco AP. Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning

- disability. **J Med Genet.** 2011 Jan;48(1):48-54. Epub 2010 Oct 23. PMID: 20972252
- 290*. Scerri TS, Brandler WM, Paracchini S, Morris AP, Ring SM, Richardson AJ, Talcott JB, Stein J, Monaco AP. PCSK6 is associated with handedness in individuals with dyslexia. **Hum Mol Genet.** 2011 Feb 1;20(3):608-14. Epub 2010 Nov 4. PMID: 21051773
291. Scerri TS, Paracchini S, Morris A, Macphie IL, Richardson AJ, Talcott J, Stein J, Smith SD, Pennington BF, Olson RK, Defries JC, Monaco AP. Identification of candidate genes for dyslexia susceptibility on chromosome 18. **PLoS One.** 2010 Oct 28;5(10):e13712. Erratum in: PLoS One. 2010;5(12). doi: 10.1371/annotation/2294a38b-878d-42f0-9faf-0822db4a0248. Richardson, Alex J [added]. PMID: 21060895
292. Paracchini S, Monaco AP, Knight JC. An Allele-specific Gene Expression Assay to Test the Functional Basis of Genetic Associations. **J Vis Exp.** 2010 Nov 3;(45). pii: 2279. doi: 10.3791/2279. PMID: 21085102
293. Newbury DF, Paracchini S, Scerri TS, Winchester L, Addis L, Richardson AJ, Walter J, Stein JF, Talcott JB, Monaco AP. Investigation of Dyslexia and SLI Risk Variants in Reading- and Language-Impaired Subjects. **Behav Genet.** 2011 Jan;41(1):90-104. Epub 2010 Dec 17. PMID: 21165691
294. Villanueva P, Newbury DF, Jara L, De Barbieri Z, Mirza G, Palomino HM, Fernández MA, Cazier JB, Monaco AP, Palomino H. Genome-wide analysis of genetic susceptibility to language impairment in an isolated Chilean population. **Eur J Hum Genet.** 2011 Jun;19(6):687-95. Epub 2011 Jan 19. PMID: 21248734
295. Scerri TS, Morris AP, Buckingham LL, Newbury DF, Miller LL, Monaco AP, Bishop DV, Paracchini S. DCDC2, KIAA0319 and CMIP Are Associated with Reading-Related Traits. **Biol Psychiatry.** 2011 Aug 1;70(3):237-45. Epub 2011 Mar 31. PMID: 21457949
296. Vieland VJ, Hallmayer J, Huang Y, Pagnamenta AT, Pinto D, Khan H, Monaco AP, Paterson AD, Scherer SW, Sutcliffe JS, Szatmari P; The Autism Genome Project (AGP). Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. **J Neurodev Disord.** 2011 Jun;3(2):113-123. Epub 2011 Jan 19. PMID:21484201
297. Pagnamenta AT, Holt R, Yusuf M, Pinto D, Wing K, Betancur C, Scherer SW, Volpi EV, Monaco AP. A family with autism and rare copy number variants disrupting the Duchenne/Becker muscular dystrophy gene DMD and TRPM3. **J Neurodev Disord.** 2011 Jun;3(2):124-31. Epub 2011 Feb 12. PMID:21484199
298. Anney RJ, Kenny EM, O'Dushlaine C, Yaspan BL, Parkhomenka E; The Autism Genome Project, Buxbaum JD, Sutcliffe J, Gill M, Gallagher L; The AGP Members, Bailey AJ, Fernandez BA, Szatmari P, Scherer SW, Patterson A, Marshall CR, Pinto D, Vincent JB, Fombonne E, Betancur C, Delorme R, Leboyer M, Bourgeron T, Mantoulan C, Roge B, Tauber M, Freitag CM, Poustka F, Duketis E, Klauck SM, Poustka A, Papanikolaou K, Tsiantis J, Gallagher L, Gill M, Anney R, Bolshakova N, Brennan S, Hughes G, McGrath J, Merikangas A, Ennis S, Green A, Casey JP, Conroy JM, Regan R, Shah N, Maestrini E, Bacchelli E, Minopoli F, Stoppioni V, Battaglia A, Iglizzo R, Parrini B, Tancredi R, Oliveira G, Almeida J, Duque F, Vicente A, Correia C, Magalhaes TR, Gillberg C, Nygren G, Jonge MD, Van Engeland H, Vorstman JA, Wittmeyer K, Baird G, Bolton PF, Rutter ML,

Green J, Lamb JA, Pickles A, Parr JR, Couteur AL, Berney T, McConachie H, Wallace S, Coutanche M, Foley S, White K, Monaco AP, Holt R, Farrar P, Pagnamenta AT, Mirza GK, Ragoussis J, Sousa I, Sykes N, Wing K, Hallmayer J, Cantor RM, Nelson SF, Geschwind DH, Abrahams BS, Volkmar F, Pericak-Vance MA, Cuccaro ML, Gilbert J, Cook EH, Guter SJ, Jacob S, Nurnberger Jr JI, McDougle CJ, Posey DJ, Lord C, Corsello C, Hus V, Buxbaum JD, Kolevzon A, Soorya L, Parkhomenko E, Leventhal BL, Dawson G, Vieland VJ, Hakonarson H, Glessner JT, Kim C, Wang K, Schellenberg GD, Devlin B, Klei L, Minshew N, Sutcliffe JS, Haines JL, Lund SC, Thomson S, Yaspan BL, Coon H, Miller J, McMahon WM, Munson J, Estes A, Wijsman EM. Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. **Eur J Hum Genet.** 2011 Oct;19(10):1082-9. doi: 10.1038/ejhg.2011.75. Epub 2011 Apr 27. PMID:21522181

299. Holt R, Monaco AP. Links between genetics and pathophysiology in the autism spectrum disorders. **EMBO Mol Med.** 2011 Aug;3(8):438-50. doi: 10.1002/emmm.201100157. PMID:21805639

300. Casey JP, Magalhaes T, Conroy JM, Regan R, Shah N, Anney R, Shields DC, Abrahams BS, Almeida J, Bacchelli E, Bailey AJ, Baird G, Battaglia A, Berney T, Bolshakova N, Bolton PF, Bourgeron T, Brennan S, Cali P, Correia C, Corsello C, Coutanche M, Dawson G, de Jonge M, Delorme R, Duketis E, Duque F, Estes A, Farrar P, Fernandez BA, Folstein SE, Foley S, Fombonne E, Freitag CM, Gilbert J, Gillberg C, Glessner JT, Green J, Guter SJ, Hakonarson H, Holt R, Hughes G, Hus V, Iglizzi R, Kim C, Klauck SM, Kolevzon A, Lamb JA, Leboyer M, Le Couteur A, Leventhal BL, Lord C, Lund SC, Maestrini E, Mantoulan C, Marshall CR, McConachie H, McDougle CJ, McGrath J, McMahon WM, Merikangas A, Miller J, Minopoli F, Mirza GK, Munson J, Nelson SF, Nygren G, Oliveira G, Pagnamenta AT, Papanikolaou K, Parr JR, Parrini B, Pickles A, Pinto D, Piven J, Posey DJ, Poustka A, Poustka F, Ragoussis J, Roge B, Rutter ML, Sequeira AF, Soorya L, Sousa I, Sykes N, Stoppioni V, Tancredi R, Tauber M, Thompson AP, Thomson S, Tsiantis J, Van Engeland H, Vincent JB, Volkmar F, Vorstman JA, Wallace S, Wang K, Wassink TH, White K, Wing K, Wittemeyer K, Yaspan BL, Zwaigenbaum L, Betancur C, Buxbaum JD, Cantor RM, Cook EH, Coon H, Cuccaro ML, Geschwind DH, Haines JL, Hallmayer J, Monaco AP, Nurnberger JI Jr, Pericak-Vance MA, Schellenberg GD, Scherer SW, Sutcliffe JS, Szatmari P, Vieland VJ, Wijsman EM, Green A, Gill M, Gallagher L, Vicente A, Ennis S. A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. **Hum Genet.** 2012 Apr;131(4):565-79. doi: 10.1007/s00439-011-1094-6. Epub 2011 Oct 14. PMID: 21996756.

301. Velayos-Baeza A, Holinski-Feder E, Neitzel B, Bader B, Critchley EM, Monaco AP, Danek A, Walker RH. Chorea-acanthocytosis genotype in the original critchley kentucky neuroacanthocytosis kindred. **Arch Neurol.** 2011 Oct;68(10):1330-3. PMID:21987550.

302. Leblond CS, Heinrich J, Delorme R, Proepper C, Betancur C, Huguet G, Konyukh M, Chaste P, Ey E, Rastam M, Anckarsäter H, Nygren G, Gillberg IC, Melke J, Toro R, Regnault B, Fauchereau F, Mercati O, Lemièrre N, Skuse D, Poot M, Holt R, Monaco AP, Järvelä I, Kantojärvi K, Vanhala R, Curran S, Collier DA, Bolton P, Chicchetti A, Klauck SM, Poustka F, Freitag CM, Waltes R, Kopp M, Duketis E, Bacchelli E, Minopoli F, Ruta L, Battaglia A, Mazzone L, Maestrini E, Sequeira AF, Oliveira B, Vicente A, Oliveira G, Pinto D, Scherer SW, Zelenika D, Delepine M, Lathrop M, Bonneau D, Guinchat V,

Devillard F, Assouline B, Mouren MC, Leboyer M, Gillberg C, Boeckers TM, Bourgeron T. Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. **PLoS Genet.** 2012 Feb;8(2):e1002521. doi: 10.1371/journal.pgen.1002521. Epub 2012 Feb 9. PMID:22346768

303. Holt R, Sykes NH, Conceição IC, Cazier JB, Anney RJ, Oliveira G, Gallagher L, Vicente A, Monaco AP, Pagnamenta AT. CNVs leading to fusion transcripts in individuals with autism spectrum disorder. **Eur J Hum Genet.** 2012 Nov;20(11):1141-7. doi: 10.1038/ejhg.2012.73. Epub 2012 May 2. PMID:22549408

304*. Anney R, Klei L, Pinto D, Almeida J, Bacchelli E, Baird G, Bolshakova N, Bölte S, Bolton PF, Bourgeron T, Brennan S, Brian J, Casey J, Conroy J, Correia C, Corsello C, Crawford EL, de Jonge M, Delorme R, Duketis E, Duque F, Estes A, Farrar P, Fernandez BA, Folstein SE, Fombonne E, Gilbert J, Gillberg C, Glessner JT, Green A, Green J, Guter SJ, Heron EA, Holt R, Howe JL, Hughes G, Hus V, Iglizoi R, Jacob S, Kenny GP, Kim C, Klevzon A, Kustanovich V, Lajonchere CM, Lamb JA, Law-Smith M, Leboyer M, Le Couteur A, Leventhal BL, Liu XQ, Lombard F, Lord C, Lotspeich L, Lund SC, Magalhaes TR, Mantoulan C, McDougle CJ, Melhem NM, Merikangas A, Minshew NJ, Mirza GK, Munson J, Noakes C, Papanikolaou K, Pagnamenta AT, Parrini B, Paton T, Pickles A, Posey DJ, Poustka F, Ragoussis J, Regan R, Renshaw K, Roberts W, Roeder K, Roge B, Rutter ML, Schlitt S, Shah N, Sheffield VC, Soorya L, Sousa I, Stoppioni V, Sykes N, Tancredi R, Thompson AP, Thomson S, Tryfon A, Tsiantis J, Van Engeland H, Vincent JB, Volkmar F, Vorstman J, Wallace S, Wing K, Wittemeyer K, Wood S, Zurawiecki D, Zwaigenbaum L, Bailey AJ, Battaglia A, Cantor RM, Coon H, Cuccaro ML, Dawson G, Ennis S, Freitag CM, Geschwind DH, Haines JL, Klauck SM, McMahon WM, Maestrini E, Miller J, Monaco AP, Nelson SF, Nurnberger JI Jr, Oliveira G, Parr JR, Pericak-Vance MA, Piven J, Schellenberg GD, Scherer SW, Vicente AM, Wassink TH, Wijsman EM, Betancur C, Buxbaum JD, Cook EH, Gallagher L, Gill M, Hallmayer J, Paterson AD, Sutcliffe JS, Szatmari P, Vieland VJ, Hakonarson H, Devlin B. Individual common variants exert weak effects on risk for Autism Spectrum Disorders. **Hum Mol Genet.** 2012 Nov 1;21(21):4781-92. doi: 10.1093/hmg/dd3301. Epub 2012 Jul 26. PMID:22843504

305. Newbury DF, Mari F, Akha ES, Macdermot KD, Canitano R, Monaco AP, Taylor JC, Renieri A, Fisher SE, Knight SJ. Dual copy number variants involving 16p11 and 6q22 in a case of childhood apraxia of speech and pervasive developmental disorder. **Eur J Hum Genet.** 2013 Apr;21(4):361-5. doi: 10.1038/ejhg.2012.166. Epub 2012 Aug 22. PMID:22909776

306. Scerri TS, Darki F, Newbury DF, Whitehouse AJ, Peyrard-Janvid M, Matsson H, Ang QW, Pennell CE, Ring S, Stein J, Morris AP, Monaco AP, Kere J, Talcott JB, Klingberg T, Paracchini S. The dyslexia candidate locus on 2p12 is associated with general cognitive ability and white matter structure. **PLoS One.** 2012;7(11):e50321. doi: 10.1371/journal.pone.0050321. Epub 2012 Nov 28. PMID:23209710

307. Holloway ZG, Baeza AV, Howell GJ, Levecque C, Ponnambalam S, Sztul E, Monaco AP. Trafficking of the Menkes copper transporter ATP7A is regulated by clathrin, AP-2,

AP-1 and Rab22-dependent steps. **Mol Biol Cell.** 2013 Jun;24(11):1735-48. doi: 10.1091/mbc.E12-08-0625. Epub 2013 Apr 17. PMID:23596324

308*. Cross-Disorder Group of the Psychiatric Genomics Consortium, Lee SH, Ripke S, Neale BM, Faraone SV, Purcell SM, Perlis RH, Mowry BJ, Thapar A, Goddard ME, Witte JS, Absher D, Agartz I, Akil H, Amin F, Andreassen OA, Anjorin A, Anney R, Anttila V, Arking DE, Asherson P, Azevedo MH, Backlund L, Badner JA, Bailey AJ, Banaschewski T, Barchas JD, Barnes MR, Barrett TB, Bass N, Battaglia A, Bauer M, Bayés M, Bellivier F, Bergen SE, Berrettini W, Betancur C, Bettecken T, Biederman J, Binder EB, Black DW, Blackwood DH, Bloss CS, Boehnke M, Boomsma DI, Breen G, Breuer R, Bruggeman R, Cormican P, Buccola NG, Buitelaar JK, Bunney WE, Buxbaum JD, Byerley WF, Byrne EM, Caesar S, Cahn W, Cantor RM, Casas M, Chakravarti A, Chambert K, Choudhury K, Cichon S, Cloninger CR, Collier DA, Cook EH, Coon H, Cormand B, Corvin A, Coryell WH, Craig DW, Craig IW, Crosbie J, Cuccaro ML, Curtis D, Czamara D, Datta S, Dawson G, Day R, De Geus EJ, Degenhardt F, Djurovic S, Donohoe GJ, Doyle AE, Duan J, Dudbridge F, Duketis E, Ebstein RP, Edenberg HJ, Elia J, Ennis S, Etain B, Fanous A, Farmer AE, Ferrier IN, Flickinger M, Fombonne E, Foroud T, Frank J, Franke B, Fraser C, Freedman R, Freimer NB, Freitag CM, Friedl M, Frisén L, Gallagher L, Gejman PV, Georgieva L, Gershon ES, Geschwind DH, Giegling I, Gill M, Gordon SD, Gordon-Smith K, Green EK, Greenwood TA, Grice DE, Gross M, Grozeva D, Guan W, Gurling H, De Haan L, Haines JL, Hakonarson H, Hallmayer J, Hamilton SP, Hamshere ML, Hansen TF, Hartmann AM, Hautzinger M, Heath AC, Henders AK, Herms S, Hickie IB, Hipolito M, Hoefels S, Holmans PA, Holsboer F, Hoogendijk WJ, Hottenga JJ, Hultman CM, Hus V, Ingason A, Ising M, Jamain S, Jones EG, Jones I, Jones L, Tzeng JY, Kähler AK, Kahn RS, Kandaswamy R, Keller MC, Kennedy JL, Kenny E, Kent L, Kim Y, Kirov GK, Klauck SM, Klei L, Knowles JA, Kohli MA, Koller DL, Konte B, Korszun A, Krabbendam L, Krasucki R, Kuntsi J, Kwan P, Landén M, Långström N, Lathrop M, Lawrence J, Lawson WB, Leboyer M, Ledbetter DH, Lee PH, Lencz T, Lesch KP, Levinson DF, Lewis CM, Li J, Lichtenstein P, Lieberman JA, Lin DY, Linszen DH, Liu C, Lohoff FW, Loo SK, Lord C, Lowe JK, Lucae S, Macintyre DJ, Madden PA, Maestrini E, Magnusson PK, Mahon PB, Maier W, Malhotra AK, Mane SM, Martin CL, Martin NG, Mattheisen M, Matthews K, Mattingsdal M, McCarroll SA, McGhee KA, McGough JJ, McGrath PJ, McGuffin P, McInnis MG, McIntosh A, McKinney R, McLean AW, McMahan FJ, McMahan WM, McQuillin A, Medeiros H, Medland SE, Meier S, Melle I, Meng F, Meyer J, Middeldorp CM, Middleton L, Milanova V, Miranda A, Monaco AP, Montgomery GW, Moran JL, Moreno-De-Luca D, Morken G, Morris DW, Morrow EM, Moskvina V, Muglia P, Mühleisen TW, Muir WJ, Müller-Myhsok B, Murtha M, Myers RM, Myin-Germeys I, Neale MC, Nelson SF, Nievergelt CM, Nikolov I, Nimgaonkar V, Nolen WA, Nöthen MM, Nurnberger JI, Nwulia EA, Nyholt DR, O'Dushlaine C, Oades RD, Olincy A, Oliveira G, Olsen L, Ophoff RA, Osby U, Owen MJ, Palotie A, Parr JR, Paterson AD, Pato CN, Pato MT, Penninx BW, Pergadia ML, Pericak-Vance MA, Pickard BS, Pimm J, Piven J, Posthuma D, Potash JB, Poustka F, Propping P, Puri V, Quedstedt DJ, Quinn EM, Ramos-Quiroga JA, Rasmussen HB, Raychaudhuri S, Rehnström K, Reif A, Ribasés M, Rice JP, Rietschel M, Roeder K, Roeyers H, Rossin L, Rothenberger A, Rouleau G, Ruderfer D, Rujescu D, Sanders AR, Sanders SJ, Santangelo SL, Sergeant JA, Schachar R, Schalling M, Schatzberg AF, Scheftner WA, Schellenberg GD, Scherer SW, Schork NJ, Schulze TG, Schumacher J, Schwarz M, Scolnick E, Scott LJ, Shi J, Shilling PD, Shyn SI, Silverman JM, Slager SL, Smalley SL, Smit JH, Smith EN, Sonuga-Barke EJ, St Clair D, State M,

Steffens M, Steinhausen HC, Strauss JS, Strohmaier J, Stroup TS, Sutcliffe JS, Szatmari P, Szelinger S, Thirumalai S, Thompson RC, Todorov AA, Tozzi F, Treutlein J, Uhr M, van den Oord EJ, Van Grootheest G, Van Os J, Vicente AM, Vieland VJ, Vincent JB, Visscher PM, Walsh CA, Wassink TH, Watson SJ, Weissman MM, Werge T, Wienker TF, Wijsman EM, Willemsen G, Williams N, Willsey AJ, Witt SH, Xu W, Young AH, Yu TW, Zammit S, Zandi PP, Zhang P, Zitman FG, Zöllner S; International Inflammatory Bowel Disease Genetics Consortium (IIBDGC), Devlin B, Kelsoe JR, Sklar P, Daly MJ, O'Donovan MC, Craddock N, Sullivan PF, Smoller JW, Kendler KS, Wray NR. Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. **Nat Genet.** 2013 Sep;45(9):984-94. doi: 10.1038/ng.2711. Epub 2013 Aug 11. PMID:23933821

309. Becker J, Czamara D, Scerri TS, Ramus F, Csépe V, Talcott JB, Stein J, Morris A, Ludwig KU, Hoffmann P, Honbolygó F, Tóth D, Fauchereau F, Bogliotti C, Iannuzzi S, Chaix Y, Valdois S, Billard C, George F, Soares-Boucaud I, Gérard CL, van der Mark S, Schulz E, Vaessen A, Maurer U, Lohvansuu K, Lyytinen H, Zucchelli M, Brandeis D, Blomert L, Leppänen PH, Bruder J, Monaco AP, Müller-Myhsok B, Kere J, Landerl K, Nöthen MM, Schulte-Körne G, Paracchini S, Peyrard-Janvid M, Schumacher J. Genetic analysis of dyslexia candidate genes in the European cross-linguistic NeuroDys cohort. **Eur J Hum Genet.** 2014 May;22(5):675-80. doi: 10.1038/ejhg.2013.199. Epub 2013 Sep 11. PMID:24022301

310*. Brandler WM, Morris AP, Evans DM, Scerri TS, Kemp JP, Timpson NJ, St Pourcain B, Smith GD, Ring SM, Stein J, Monaco AP, Talcott JB, Fisher SE, Webber C, Paracchini S. Common variants in left/right asymmetry genes and pathways are associated with relative hand skill. **PLoS Genet.** 2013 Sep;9(9):e1003751. doi:10.1371/journal.pgen.1003751. Epub 2013 Sep 12. PMID:24068947

311. Nudel R, Simpson NH, Baird G, O Hare A, Conti-Ramsden G, Bolton PF, Hennessy ER, Monaco AP, Knight JC, Winney B, Fisher SE, Newbury DF. Associations of HLA alleles with specific language impairment. **J Neurodev Disord.** 2014 Jan 17;6(1):1. doi: 10.1186/1866-1955-6-1. PMID:24433325

312. Nudel R, Simpson NH, Baird G, O'Hare A, Conti-Ramsden G, Bolton PF, Hennessy ER; the SLI Consortium, Ring SM, Smith GD, Francks C, Paracchini S, Monaco AP, Fisher SE, Newbury DF. Genome-wide association analyses of child genotype effects and parent-of-origin effects in specific language impairment. **Genes Brain Behav.** 2014 Apr;13(4):418-29. doi: 10.1111/gbb.12127. Epub 2014 Mar 24. PMID:24571439

313. Ceroni F, Sagar A, Simpson NH, Gawthrop AJ, Newbury DF, Pinto D, Francis SM, Tessman DC, Cook EH, Monaco AP, Maestrini E, Pagnamenta AT, Jacob S. A Deletion Involving CD38 and BST1 Results in a Fusion Transcript in a Patient With Autism and Asthma. **Autism Res.** 2014 Apr;7(2):254-63. doi: 10.1002/aur.1365. Epub 2014 Mar 13. PMID:24634087

314. Newbury DF, Monaco AP, Paracchini S. Reading and language disorders: the

importance of both quantity and quality. **Genes** (Basel). 2014 Apr 4;5(2):285-309. doi: 10.3390/genes5020285. PMID:24705331

315*. Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, Klei L, Thiruvahindrapuram B, Xu X, Ziman R, Wang Z, Vorstman JA, Thompson A, Regan R, Pilorge M, Pellecchia G, Pagnamenta AT, Oliveira B, Marshall CR, Magalhaes TR, Lowe JK, Howe JL, Griswold AJ, Gilbert J, Duketis E, Dombroski BA, De Jonge MV, Cuccaro M, Crawford EL, Correia CT, Conroy J, Conceição IC, Chiocchetti AG, Casey JP, Cai G, Cabrol C, Bolshakova N, Bacchelli E, Anney R, Gallinger S, Cotterchio M, Casey G, Zwaigenbaum L, Wittemeyer K, Wing K, Wallace S, van Engeland H, Tryfon A, Thomson S, Soorya L, Rogé B, Roberts W, Poustka F, Mouga S, Minshew N, McInnes LA, McGrew SG, Lord C, Leboyer M, Le Couteur AS, Kolevzon A, Jiménez González P, Jacob S, Holt R, Guter S, Green J, Green A, Gillberg C, Fernandez BA, Duque F, Delorme R, Dawson G, Chaste P, Café C, Brennan S, Bourgeron T, Bolton PF, Bölte S, Bernier R, Baird G, Bailey AJ, Anagnostou E, Almeida J, Wijsman EM, Vieland VJ, Vicente AM, Schellenberg GD, Pericak-Vance M, Paterson AD, Parr JR, Oliveira G, Nurnberger JI, Monaco AP, Maestrini E, Klauck SM, Hakonarson H, Haines JL, Geschwind DH, Freitag CM, Folstein SE, Ennis S, Coon H, Battaglia A, Szatmari P, Sutcliffe JS, Hallmayer J, Gill M, Cook EH, Buxbaum JD, Devlin B, Gallagher L, Betancur C, Scherer SW. Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. **Am J Hum Genet.** 2014 May 1;94(5):677-94. doi: 10.1016/j.ajhg.2014.03.018. Epub 2014 Apr 24. PMID:24768552

316. Simpson NH, Addis L, Brandler WM, Slonims V, Clark A, Watson J, Scerri TS, Hennessy ER, Bolton PF, Conti-Ramsden G, Fairfax BP, Knight JC, Stein J, Talcott JB, O'Hare A, Baird G, Paracchini S, Fisher SE, Newbury DF; SLI Consortium. Increased prevalence of sex chromosome aneuploidies in specific language impairment and dyslexia. **Dev Med Child Neurol.** 2014 Apr;56(4):346-53. doi: 10.1111/dmcn.12294. Epub 2013 Oct 9. PMID:24117048

317. Hadley D, Wu ZL, Kao C, Kini A, Mohamed-Hadley A, Thomas K, Vazquez L, Qiu H, Mentch F, Pellegrino R, Kim C, Connolly J; AGP Consortium, Glessner J, Hakonarson H. The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. **Nat Commun.** 2014 Jun 13;5:4074. doi: 10.1038/ncomms5074. PMID:24927284

318. Gialluisi A, Newbury DF, Wilcutt EG, Olson RK, DeFries JC, Brandler WM, Pennington BF, Smith SD, Scerri TS, Simpson NH; The SLI Consortium, Luciano M, Evans DM, Bates TC, Stein JF, Talcott JB, Monaco AP, Paracchini S, Francks C, Fisher SE. Genome-wide screening for DNA variants associated with reading and language traits. **Genes Brain Behav.** 2014 Sep;13(7):686-701. doi: 10.1111/gbb.12158. Epub 2014 Aug 29. PMID:25065397

319. Leblond CS, Nava C, Polge A, Gauthier J, Huguet G, Lumbroso S, Giuliano F, Stordeur C, Depienne C, Mouzat K, Pinto D, Howe J, Lemièrre N, Durand CM, Guibert J, Ey E, Toro R, Peyre H, Mathieu A, Amsellem F, Rastam M, Gillberg IC, Rappold GA, Holt R, Monaco AP, Maestrini E, Galan P, Heron D, Jacquette A, Afenjar A, Rastetter A, Brice

- A, Devillard F, Assouline B, Laffargue F, Lespinasse J, Chiesa J, Rivier F, Bonneau D, Regnault B, Zelenika D, Delepine M, Lathrop M, Sanlaville D, Schluth-Bolard C, Edery P, Perrin L, Tabet AC, Schmeisser MJ, Boeckers TM, Coleman M, Sato D, Szatmari P, Scherer SW, Rouleau GA, Betancur C, Leboyer M, Gillberg C, Delorme R, Bourgeron T. Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. **PLoS Genet.** 2014 Sep 4;10(9):e1004580. doi: 10.1371/journal.pgen.1004580. eCollection 2014 Sep. PMID:25188300
320. Simpson NH, Ceroni F, Reader RH, Covill LE, Knight JC; the SLI Consortium, Hennessy ER, Bolton PF, Conti-Ramsden G, O'Hare A, Baird G, Fisher SE, Newbury DF; the SLI Consortium. Genome-wide analysis identifies a role for common copy number variants in specific language impairment. **Eur J Hum Genet.** 2015 Jan 14. doi: 10.1038/ejhg.2014.296. [Epub ahead of print] PMID:25585696
321. Maier R, Moser G, Chen GB, Ripke S; Cross-Disorder Working Group of the Psychiatric Genomics Consortium, Coryell W, Potash JB, Scheftner WA, Shi J, Weissman MM, Hultman CM, Landén M, Levinson DF, Kendler KS, Smoller JW, Wray NR, Lee SH; Cross-Disorder Working Group of the Psychiatric Genomics Consortium. Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. **Am J Hum Genet.** 2015 Feb 5;96(2):283-94. doi: 10.1016/j.ajhg.2014.12.006. Epub 2015 Jan 29. PMID:25640677
322. Pettigrew KA, Fajutrao Valles SF, Moll K, Northstone K, Ring S, Pennell C, Wang C, Leavett R, Hayiou-Thomas ME, Thompson P, Simpson NH, Fisher SE; SLI Consortium, Whitehouse AJ, Snowling MJ, Newbury DF, Paracchini S. Lack of replication for the myosin-18B association with mathematical ability in independent cohorts. **Genes Brain Behav.** 2015 Apr;14(4):369-76. doi: 10.1111/gbb.12213. Epub 2015 Apr 1. PMID:25778778
323. Shore R, Covill L, Pettigrew KA, Brandler WM, Diaz R, Xu Y, Tello JA, Talcott JB, Newbury DF, Stein J, Monaco AP, Paracchini S. The handedness-associated PCSK6 locus spans an intronic promoter regulating novel transcripts. **Hum Mol Genet.** 2016 Feb 21. pii: ddw047. [Epub ahead of print] PMID:26908617
324. Yang RY, Xue H, Yu L, Velayos-Baeza A, Monaco AP, Liu FT. Identification of VPS13C as a Galectin-12-Binding Protein That Regulates Galectin-12 Protein Stability and Adipogenesis. **PLoS One.** 2016 Apr 13;11(4):e0153534. doi: 10.1371/journal.pone.0153534. eCollection 2016. PMID:27073999
325. Pettigrew KA, Frinton E, Nudel R, Chan MT, Thompson P, Hayiou-Thomas ME, Talcott JB, Stein J, Monaco AP, Hulme C, Snowling MJ, Newbury DF, Paracchini S. Further evidence for a parent-of-origin effect at the NOP9 locus on language-related phenotypes. **J Neurodev Disord.** 2016 Jun 14;8:24. doi: 10.1186/s11689-016-9157-6. eCollection 2016. PMID:27307794
326. Mehta ZB, Fine N, Pullen TJ, Cane MC, Hu M, Chabosseau P, Meur G, Velayos-Baeza A, Monaco AP, Marselli L, Marchetti P, Rutter GA. Changes in the expression of the type 2 diabetes-associated gene VPS13C in the β cell are associated with glucose intolerance in

humans and mice. **Am J Physiol Endocrinol Metab.** 2016 Aug 1;311(2):E488-507. doi: 10.1152/ajpendo.00074.2016. Epub 2016 Jun 21. PMID:27329800

327. Martinez-Garay I, Guidi LG, Holloway ZG, Bailey MA, Lyngholm D, Schneider T, Donnison T, Butt SJ, Monaco AP, Molnár Z, Velayos-Baeza A. Normal radial migration and lamination are maintained in dyslexia-susceptibility candidate gene homolog Kiaa0319 knockout mice. **Brain Struct Funct.** 2016 Aug 10. [Epub ahead of print] PMID:27510895

Book Chapters and Book Reviews:

1. Monaco AP, Hernandez L, Hoebel BG. Nucleus accumbens: site of amphetamine self-injection; comparison with the lateral ventricle. In The neurobiology of the nucleus accumbens. RB Chronister and JF DeFrance (eds). Brunswick, Maine: Haer Institute, 1980.

2. Kunkel LM, Monaco AP, Middlesworth W, Bertelson C, Fischbeck K. Molecular analysis of the DNA surrounding the Duchenne muscular dystrophy locus. ICN-UCLA symp. Mol. Cell Biol. VIII, In Molecular Biology of Muscle Development. Emerson C, Fischman D, Nadal-Ginard B, Siddiqui MAQ (eds), Academic Press, 1986; pp.865-874.

3. Kunkel LM, Bertelson CJ, Monaco AP, Colletti CA. Molecular and genetic analysis of the region surrounding Duchenne muscular dystrophy. In Current Communications in Molecular Biology: DNA probes: Applications in Genetic and Infectious Disease and Cancer. Lerman LS (ed). Cold Spring Harbor, NY, 1987; pp.37-42.

4. Kunkel LM, Monaco AP, Colletti-Feener C, Bertelson C. Approaching the gene responsible for Duchenne muscular dystrophy. In Expression of Neural Genes. Alan R. Liss, NY; 1987; pp.125-131.

5. Monaco AP, Kunkel LM. Reverse genetics and the Duchenne muscular dystrophy locus. In Evolutionary Mechanisms in Sex Determination. S Wachtel (ed.) CRC Press Inc., NY; 1988; pp.233-239.

6. Monaco AP. The tools of molecular genetics: DNA probes, library screening, DNA sequencing and expression vectors. In Molecular Genetics in Diseases of Brain, Nerve and Muscle. LP Rowland, DS Wood, EA Schon and S DiMauro (eds.), Oxford University Press, NY and Oxford; 1989; pp.145-155.

7. Lehrach H, Drmanac R, Hoheisel J, Larin Z, Lennon G, Nizetic D, Monaco AP, Zehetner G and Poutska A. Hybridization fingerprinting in genome mapping and sequencing. In Genome Analysis Volume 1: Genetic and Physical Mapping. KE Davies and S.M.Tilghman (eds.), Cold Spring Harbor Laboratory Press, Cold Spring Harbor, NY; 1990; pp.39-81.

*8. Monaco AP, Larin Z and Lehrach H. Construction of yeast artificial chromosome libraries by pulsed field gel electrophoresis. In Methods in Molecular Biology: Vol 12: Pulsed-Field Gel Electrophoresis. M Burmeister and L Ulanovsky (eds.), The Humana Press, Totowa, NJ; 1992; pp.225-234.

9. Larin Z, Monaco AP and Lehrach H. Large insert yeast artificial chromosome libraries. In YAC Libraries: A User's Guide. D. Nelson and B. Brownstein (eds), W.H. Freeman and Co., New York, NY, 1993; pp.43-56.
- *10. Monaco AP, Walker AP, Ho MF, Chelly J, Clarke E, Ishikawa-Brush Y, Muscatelli F. Molecular genetics and physical mapping in human Xp21. In Cold Spring Harbor Genome Analysis Volume 5: Regional Physical Mapping. KE Davies and SM Tilghman (eds), Cold Spring Harbor Press, Cold Spring Harbor, NY, 1993; pp.35-61.
11. Monaco AP., Book Review of "Molecular Basis of Inherited Disease (2nd edn) (Kay E. Davies and Andrew P. Read)" **Trends Genet** 1993; 9:94.
12. Monaco AP, Larin Z. Generating subclones from large insert genomic libraries. In Current Protocols in Human Genetics. NC Dracopoli, JL Haines, BR Korf, DT Moir, CC Morton, CE Seidman, JG Seidman, DR Smith (eds), Current Protocols, Brooklyn, NY; 1994 Supplement 3; Unit 5.11.
13. Monaco AP., Book Review of "Duchenne Muscular Dystrophy (2nd edn.) by Alan E. H. Emery, **Am J Hum Genet** 1994; 54:212-213.
- *14. Ho MF and Monaco AP. PFGE in physical mapping. In Pulsed Field Gel Electrophoresis: A Practical Approach. AP Monaco (ed), Oxford University Press, Oxford 1995; pp.21-43.
15. Nelson DL, Ballabio A, Cremers F, Monaco AP, Schlessinger D, Willard HF. Report of the committee on the genetic constitution of the X chromosome. In Human Gene Mapping 1994, a Compendium. AJ Cuticchia (ed). The Johns Hopkins University Press, Baltimore, 1995; pp.787-891.
16. Monaco AP, Human genetic methods. Book Review of Human Molecular Genetics: Methods in Molecular Genetics (vol 8) edited by Kenneth W Adolph. **Trends Genet** (1996); 12:488.
17. Bailey A and Monaco AP. Revealing the roots of autism. **MRC News** Autumn/Winter 1996; 72:32-35.
18. Francis MJ and Monaco AP. The identification of motifs involved in the intracellular trafficking of the Menkes disease protein. In Handbook of Copper Toxicology and Pharmacology; EJ Massaro (ed). Humana Press, Totowa, NJ. 2002.
19. Dobson-Stone C, Rampoldi L, Monaco AP. The spectrum of mutations and possible function of the CHAC gene. In Neuroacanthocytosis Syndromes A Danek (ed), Springer 2004, pp. 169-175.
20. Monaco AP. Recipe for the Mind. Book Review of The Birth Of The Mind by Gary Marcus PhD. **Nature** 2004 ; 427:681.

21. Newbury DF, Monaco AP. (2008) The Application of Molecular Genetics to the Study of Language Impairments. In Understanding Developmental Language Disorders: From Theory to Practice CF Norbury, JB Tomblin & DVM Bishop (eds), Psychology Press. July 2008, pp. 79-92.

22. Velayos-Baeza A, Levecque C, Dobson-Stone C, Monaco AP. The function of chorein. In Neuroacanthocytosis Syndromes II. R.H. Walker (ed), Springer-Verlag, Berlin Heidelberg, 2008, pp. 87-105.

23. Sousa I, Holt R, Pagnamenta A, Monaco M. Unravelling the Genetics of Autism Spectrum Disorders. In: Researching the Autism Spectrum. Roth I, Rezaie P, (eds) Cambridge: Cambridge University Press, 2011, pp. 53-111.