

# Wellcome Trust Centre for Human Genetics – 2006 publications

## Genomics Core

### Head: Dr Ioannis Ragoussis

Caluseriu O., Mirza G., Ragoussis J., Chow E. W., MacCrimmon D., and Bassett A. S. (2006). Schizophrenia in an adult with 6p25 deletion syndrome. *Am J Med Genet A* 140: 1208-13.

Elvidge G. P., Glenny L., Appelhoff R. J., Ratcliffe P. J., Ragoussis J.\*, and Gleadle J. M. (2006). Concordant regulation of gene expression by hypoxia and 2-oxoglutarate-dependent dioxygenase inhibition: the role of HIF-1alpha, HIF-2alpha, and other pathways. *J Biol Chem* 281: 15215-26.  
\*co-corresponding author

Lai M. I., Jiang J., Silver N., Best S., Menzel S., Mijovic A., Colella S., Ragoussis J., Garner C., Weiss M. J., and Thein S. L. (2006). alpha-Haemoglobin stabilising protein is a quantitative trait gene that modifies the phenotype of beta-thalassaemia. *Br J Haematol* 133: 675-82.

Laun K., Coggill P., Palmer S., Sims S., Ning Z., Ragoussis J., Volpi E., Wilson N., Beck S., Ziegler A., and Volz A. (2006). The Leukocyte Receptor Complex in Chicken Is Characterized by Massive Expansion and Diversification of Immunoglobulin-Like Loci. *PLoS Genet* 2: e73.

Lonie L., Porter D. E., Fraser M., Cole T., Wise C., Yates L., Wakeling E., Blair E., Morava E., Monaco A. P., and Ragoussis J. (2006). 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* 27: 1160.

McGovern D. P., Butler H., Ahmad T., Paolucci M., van Heel D. A., Negro K., Hysi P., Ragoussis J., Travis S. P., Cardon L. R., and Jewell D. P. (2006). TUCAN (CARD8) Genetic Variants and Inflammatory Bowel Disease. *Gastroenterology* 131: 1190-6.

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Ragoussis J., and Elvidge G. (2006). Affymetrix GeneChip system: moving from research to the clinic. *Expert Rev Mol Diagn* 6: 145-52.

Ragoussis J. (2006) Genotyping systems for all. *Drug Discovery Today: Technologies*: in press

### Books/Book chapters/Book Reviews/Editorials

Markie, D. and Ragoussis J. Mitotic recombination of yeast artificial chromosomes (2<sup>nd</sup> edition). *Methods in Mol. Biol.* 2005 (in press)

Ragoussis J., Field S and Udalova I. Quantitative profiling of protein-DNA binding on microarrays. *Methods in Mol. Biol.* 2005 (in press)

Ragoussis J., Field S., and Udalova I. A. (2006b). Quantitative profiling of protein-DNA binding on microarrays. *Methods Mol Biol* 338: 261-80. Previously in press

Field S., Udalova I., and Ragoussis J. (2006). Accuracy and reproducibility of protein-DNA microarray technology. *Adv Biochem Eng Biotechnol* in press.

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### Dr G Elvidge

Elvidge G. (2006). Microarray expression technology: from start to finish. *Pharmacogenomics* 7: 123-34. –previously in press

Elvidge GP, Price TS, Glenny L and Ragoussis J. Development and evaluation of real competitive PCR for high-throughput quantitative applications. *Anal Biochem* 339: 231-41, 2005.

#### Reviews

Ragoussis J, Elvidge G. P., Kaur K., and Colella S. (2006). Matrix-assisted laser desorption/ionisation, time-of-flight mass spectrometry in genomics research. *PLoS Genet* 2: e100.

Ragoussis J, and Elvidge G. (2006). Affymetrix GeneChip system: moving from research to the clinic. *Expert Rev Mol Diagn* 6: 145-52.

### Dr S Colella

Simeone A. M., Colella S., Krahe R., Johnson M. M., Mora E., and Tari A. M. (2006). N-(4-Hydroxyphenyl)retinamide and nitric oxide pro-drugs exhibit apoptotic and anti-invasive effects against bone metastatic breast cancer cells. *Carcinogenesis* 27: 568-77.

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Ragoussis J, Elvidge G. P., Kaur K., and Colella S. (2006). Matrix-assisted laser desorption/ionisation, time-of-flight mass spectrometry in genomics research. *PLoS Genet* 2: e100.

## Molecular Cytogenetics & Microscopy Core

### Head: Dr Emanuela Volpi

Laun K, Coggill P, Palmer S, Sims S, Ning Z, Ragoussis J, Volpi E, Wilson N, Beck S, Ziegler A, Volz A. The leukocyte receptor complex in chicken is characterized by massive expansion and diversification of immunoglobulin-like loci. *PLoS Genetics*, 2: e73, 2006.

Wilson ND, Ross LNJ, Crow TJ, Volpi EV. PCDH11 is X/Y homologous in Homo sapiens but not in Gorilla gorilla and Pan troglodytes. *Cytogenet Genome Res*, 114: 137-139, 2006.

Wilson ND, Ross LNJ, Mott R, Crow TJ, Volpi EV. Replication profile of PCDH11X and PCDH11Y, a gene pair located in the non-pseudoautosomal homologous region Xq21.3/Yp11.2. Submitted, 2006.

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Moralli D, Jefferson A, Simpson K, Volpi E, Larin Monaco Z. The centromere organisation and chromatin assembly on gene expressing HACs. International Cytogenetics and Genome Society Meeting 2006.

Suliman S, Wilson, Wass J, Volpi E, Karpe F, Gloyn AL. A novel autosomal dominant form of insulin resistance (IR) and polycystic ovarian syndrome (PCOS) resulting from a balanced translocation. Genomics of Hyperglycaemia Meeting 2006.

### Dr A Jefferson – group member

#### Books/Book chapters/Book Reviews/Editorials

Moralli D, Jefferson A, Simpson K, Volpi E, Larin Monaco Z. The centromere organisation and chromatin assembly on gene expressing HACs. International Cytogenetics and Genome Society Meeting 2006.

## Bioinformatics and Statistical Genetics Core

### Head: Professor Richard Mott

Mott R. (2006) Finding the molecular basis of complex genetic variation in humans and mice. *Philos Trans R Soc Lond B Biol Sci*. 361(1467):393-401. REVIEW

Fiddy S, Cattermole D, Xie D, Duan XY, Mott R. (2006) An integrated system for genetic analysis. *BMC Bioinformatics*. 2006 7:210.

Valdar W, Solberg LC, Gaugier D, Cookson WO, Rawlins JNP, Mott R, Flint J (2006) Genetic and environmental effects on complex traits in mice. *Genetics* (in press)

Valdar W, Solberg LC, Gaugier D, Burnett S, Klenerman P, Cookson WO, Taylor M, Rawlins JNP, Mott R, Flint J (2006) Genome-wide genetic association of complex traits in outbred mice. *Nature Genetics* 38(8):879-87

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Valdar W, Flint J, Mott R (2006) Simulating the collaborative cross: power of QTL detection and mapping resolution in large sets of recombinant inbred strains of mice. *Genetics* 172(3):1783-97.

Hanchard NA, Rockett KA, Spencer C, Coop G, Pinder M, Jallow M, Kimber M, McVean G, Mott R, Kwiatkowski DP. (2006) Screening for recently selected alleles by analysis of human haplotype similarity. *Am J Hum Genet*. Jan;78(1):153-9.

### Dr Richard Copley

Bouurlat SJ, Juliusdottir T, Lowe C, Gerhart J, Freeman R, Aronowicz J, Kirschner M, Lander ES, Thorndyke M, Nakano H, Moroz L, Heyland A, Copley RR, Telford MJ. Phylogeny of all major deuterostome groups reveals the new phylum Xenoturbellida and monophyletic chordates

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Webster BL, Copley RR\*, Jenner RA, Mackenzie-Dodds JA, Bourlat SJ, Littlewood DTJ, Telford MJ. Mitogenomics and phylogenomics reveal priapulid worms as extant models of the ancestral ecdysozoan *Evol Dev*, 8:502-510 [\* joint first author]

Matus MQ, Copley RR, Dunn CW, Hejnal A, Eccleston H, Halanych KM, Martindale, MQ, Telford MJ. (2006) *Curr Biol* 16:R575-6 Broad taxon and gene sampling indicate that Chaetognaths are protostomes

Letunic I, Copley RR, Pils B, Pinkert S, Schultz J, Bork P (2006) *Nucleic Acids Res.* 34(Database issue):D257-60 SMART 5: domains in the context of genomes and networks.

Willis-Owen SA, Shifman S, Copley RR, Flint J (2006) *Mol Psychiatry* 11:121-122 DCNP1: a novel candidate gene for major depression

### Dr Andrew Morris

Morris AP (2006). A flexible Bayesian framework for modeling haplotype association with disease allowing for dominance effects of the underlying causative variants. *American Journal of Human Genetics* 79: 679-694.

Evans D, Morris AP, Cardon LR, Sham PC (2006). A note on the power to detect transmission distortion in parent-child trios via the transmission disequilibrium test. *Behaviour Genetics* (in press).

### Dr Jordana Tzenova-Bell

Shifman S, Tzenova-Bell J, Copley RR, Taylor MS, Williams RW, Mott R, Flint J (2006) A high resolution single nucleotide polymorphism genetic map of the mouse genome *PLoS Biol*, in press

Tosh K, Ravikumar M, Tzenova-Bell J, Meisner S, Hill AVS, and Pitchappan R. 2006. Variation in MICA and MICB genes and enhanced susceptibility to leprosy in South India. *Human Molecular Genetics*, in press.

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### Dr Denise Brocklebank

Andresen JM, Gayan J, Cherny SS, Brocklebank D, Alkorta-Aranburu G, Addis EA, Cardon LR, Housman DE, Wexler NS. (2006) Replication of twelve association studies for Huntington's disease residual age of onset in large Venezuelan kindreds. *J Med Genet*. 2006 Oct 3 [Epub ahead of print];

### Dr Martin Taylor

Taylor MS, Kai C, Kawai J, Carninci P, Hayashizaki Y, Semple CAM. Heterotachy in mammalian promoter evolution. *PLoS Genetics*. 2006 Apr 28;2(4):e30.

Smyth IM, Wilming L, Lee AW, Taylor MS, Gautier P, Barlow K, Wallis J, Martin S, Glithero R, Phillimore B, Pelan S, Andrew R, Holt K, Taylor R, McLaren S, Burton J, Bailey J, Sims S, Squares J, Plumb B, Joy A, Gibson R, Gilbert J, Hart E, Laird G, Loveland J, Mudge J, Steward C, Swarbreck D, Harrow J, North P, Leaves N, Greystrom J, Coppola M, Manjunath S, Campbell M, Smith M, Strachan G, Tofts C, Boal E, Cobley V, Hunter G, Kimberley C, Thomas D, Cave-Berry L, Weston P, Botcherby MR, White S, Edgar R, Cross SH, Irvani M, Hummerich H, Simpson EH, Johnson D, Hunsicker PR, Little PF, Hubbard T, Campbell RD, Rogers J, Jackson IJ. Genomic Anatomy of the TYRP1 (brown) Deletion Complex. *PNAS*. 103(10):3704-9

Taylor MS, Copley RR. Comparative genomics. In 'Bioinformatics for Geneticists (2nd edition)' (edited by Michael R. Barnes and Ian C. Gray. Wiley. In press. REVIEW

### Dr Jen Taylor

Chittenden, TW, Sherman JA, Xong F, Hall AE, Lanahan A, Taylor JM, Duan H, Pearlman J, Moore JH, Schwartz S, Simons M (2006) Transcription profiling in coronary artery disease: indications for novel markers of coronary collateralization. *Circulation* 114(17):1811-20

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. (2006) The chromosome 6p22 haplotype associated with dyslexia reduces the expression of KIAA0319, a novel gene involved in neuronal migration. *Hum Mol Genet*. 15(10):1659-66.

### Dr William Valdar

Orton S, Herrera BM, Yee IM, Valdar W, Sadovnick AD, Ebers GC (2006) Increasing sex ratio of multiple sclerosis in Canada. *Lancet Neurology* (in press).

## Research Groups – Neurogenetics & Psychiatric Genetics

### Professor George Ebers

Dyment DA, Yee IML, Ebers GC, Sadovnick AD for the Canadian Collaborative Study Group. Multiple sclerosis in stepsiblings: recurrence risk and ascertainment. *J Neurol Neurosurg Psychiatry* (2006) 77(2):258-259.

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Kremenchutzky M, Rice GPA, Baskerville J, Wingerchuk DM, Ebers GC. The natural history of multiple sclerosis: a geographically based study IX. Observations on the progressive phase of the disease. *Brain* (2006) 129(Pt 3): 584-594.

Saarela J, Kallio S, Chen D, Montpetit A, Jokiaho A, Eva Choi, Asselta R, Bronnikov D, Lincoln M, Sadovnick AD, Tienari P, Koivisto K, Palotie A, Ebers GC, Hudson TJ, Peltonen L. PRKCA shows association to multiple sclerosis in two populations. *PLoS-Genet* (2006) 2(3, e42):0364-0375.

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Ebers G. Disease evolution in multiple sclerosis. (*J Neurol*)

Filippini G and Ebers GC. Multiple sclerosis and optic neuritis, in “Evidence-based Neurology and Neurosurgery”. Ed. Candelise L, et al. (Blackwell Publishing Ltd, Oxford)

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### Dr Simon Fisher

Fisher SE, Marcus GF. The eloquent ape: genes, brains and the evolution of language. *Nature Rev Genet* 7: 9-20, 2006.

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Fisher SE. How can animal studies help to uncover the roles of genes implicated in human speech and language disorders? In: *Transgenic and Knockout Models of Neuropsychiatric Disorders* (eds. Fisch GS, Flint, J) 127-149 (Humana press, USA) 2006.

### Professor Jonathan Flint

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### Dr Sam Knight

Koolen DA, Vissers LELM, Pfundt R, de Leeuw N, Knight SJL, Regan R, Kooy F, Reyniers E, Romano C, Fichera M, Schinzel A, Baumer A, Anderlid BM, Schoumans J, Knoers NV, v. Kessel AG, Sistermans EA, Veltman JA, Brunner HG, de Vries BBA. A New Frequent Microdeletion Syndrome due to a 17q21.31 deletion encompassing the MAPT gene, associated with a common inversion polymorphism, *Nature Genetics*. 38:999-1001, 2006.

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Knight S.J.L. and Regan R. Idiopathic Learning Disability and Genome Imbalance. *Cytogenetic and Genome Research*, in press.

### Professor Anthony Monaco

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### Other publications

Lonie L, Porter DE, Fraser M, Cole T, Wise C, Yates L, Wakling 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; 27(11):1160.

### Dr Richard Wade-Martins

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## Research Groups – Immunity & Inflammation

### Professor Adrian Hill

Hill AV (2006). Pre-erythrocytic malaria vaccines: towards greater efficacy. *Nature Reviews. Immunology* 6:21-32.

Dunachie SJ, Walther M, Vuola JM, Webster DP, Keating SM, Berthoud T, Andrews L, Bejon P, Poulton I, Butcher G, Watkins K, Sinden RE, Leach A, Moris P, Tornieporth N, Schneider J, Dubovsky F, Tierney E, Williams J, Gray Heppner D Jr, Gilbert SC, Cohen J, Hill AV (2006). A clinical trial of prime-boost immunisation with the candidate malaria vaccines RTS,S/AS02A and MVA-CS. *Vaccine* 24:2850-9.

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