

Gail Norbury's publications and presentations

Sagoo GS, **Norbury G**, Mohammed S, Kroese M. Whole-exome sequencing in clinical genetics. A health economic analysis. May 2017 (in press)

Shaw AC, Izatt L, Kulkarni A, Ruddy D, Sodha N, Tripathi V, Green M, **Norbury G**. Data from 1100 families used to model investigation strategies in hereditary bowel cancer. European Hereditary Tumour Group Meeting 2016. Mallorca.

Sagoo GS, Mohammed S, Barton G, **Norbury G**, Ahn JW, Ogilvie CM, Kroese M. 2015. Cost Effectiveness of Using Array-CGH for Diagnosing Learning Disability. Appl Health Econ Health Policy. 2015 Aug;13(4):421-32

Watts S, **G. Norbury**, L. Izatt, C. Jacobs, A. Kulkarni, D. Ruddy, A. Shaw. Challenging the clinical effectiveness of offering mismatch repair testing to patients with ovarian cancer. Cancer Genetics Group Meeting and International Meeting on the Psychosocial Aspects of Hereditary Cancer 2015 Manchester

Brittain H, Ridout C, Campbell J, Jacobs C, **Norbury G**, Ruddy D, Izatt L, Shaw A, Kulkarni A. Outcomes from BRCA1 and BRCA2 testing in an epithelial ovarian cancer (EOC) cohort. BSGM 2014.

Thomas E, Y Patel, S Lillis, M Irving, M Holder-Espinasse, C Deshpande, D Josifova, L Robert, L Izatt, F Flinter, A Saxena, C Ogilvie, **G Norbury**, J Ahn, M Yau, S Mohammed, M Simpson. Whole exome sequencing with virtual panel analysis for molecular diagnosis in the genetics clinic. BSGM 2014.

Levene S, **G Norbury**, G Say, M Jackson, C Patch Experience of offering self-funded carrier screening for an expanded panel of recessive conditions to Ashkenazi Jewish patients attending the NHS Tay Sachs screening clinic. BSGM 2014.

Sagoo G., S Mohammed, **G Norbury**, JW Ahn, CM Ogilvie, M Kroese. Cost-effectiveness of using array CGH for diagnosing learning disability. BSGM 2014.

Norbury G, Watts S, Izatt L, Jacobs, C, Kulkarni, Ruddy D, Shaw A, Chandler C, Sodha N, Keating S, Gill-Barman B. Four years of the integrated mismatch repair deficiency service at Guy's & St Thomas NHS Foundation Trust. BSHG 2014

Norbury G on behalf of the ACGS Quality Sub Committee & Audit Participants
Twenty years of the Clinical Molecular Genetics Society (CMGS) Activity Audit; past, present & future. ACGS 2014

Levene S, **G Norbury**, G Say, M Jackson, C Patch Experience of offering self-funded carrier screening for an expanded panel of recessive conditions to Ashkenazi Jewish patients attending the NHS Tay Sachs screening clinic - BSGM 2013

Chandler N, T Cullup, **G Norbury**, R Sarkany, A Lehmann, S Abbs, D Ellis, S Mohammed, H Fassihi. Xeroderma Pigmentosum (XP) - the next generation of testing. BSGM 2013

Mohammed S, N. Chandler, T. Cullup T, D. McGibbin, **G. Norbury**, R. Sarkany, A. Lehmann, S. Abbs, H. Fassihi. Clinical Genetics Society 2013. Journey of the XP patient: observations from a multidisciplinary clinic.

Norbury G, J. Hoyle, A. Devereau, P. Collinson Incorporating Genetics into the National Laboratory Medicine Catalogue.2012. J Med Genet 49: Suppl 1.S124

Slatter M, B. Angus, , K. Windebank, A. Taylor, C. Meaney, T. Lester **G. Norbury**, S. Hambleton M. Abinun T. Flood, A. Cant, and A. Gennery. 2011. Polymorphous lymphoproliferative disorder with Hodgkin like features in common gamma chain deficient severe combined immunodeficiency. J Allergy Clin Immunol. 127(2):533-5

Meaney C and **G. Nobury**. Non-invasive Prenatal Diagnosis. 2011. Methods Mol Biol. 688:155-72.

Norbury G, D. Ruddy, L. Izatt, H. Deere, J. Freeman, D. Moore, N. Trump, P. Ross, M. George, G. Pichert, A. Shaw, C. Jacobs. 2011. J Med Genet 48: Suppl 1.S98. Outcomes from the new integrated mismatch repair deficiency service at Guy's & St Thomas NHS Foundation Trust.

Stenhouse SAR, R. Butler, A. Curtis, J. Deller, K. Kelly, **G. Norbury**, R. Mountford. 2011. Eur J Hum Genet 19 Suppl 2.484. A robust measure of workload for molecular diagnostic laboratories.

Hill M, K. Finning, P. Martin, J. Hogg, C. Meaney, **G. Norbury**, G. Daniels and L. Chitty. 2010. Non-invasive prenatal determination of fetal sex: translating research into clinical practice. Clin Genet. Aug 19.

A Taylor, G. Bayly, K. Patel, L. Yarram, M. Williams, J. Hamilton-Shield, SE. Humphries and **G. Norbury**. 2010. A double heterozygote for familial hypercholesterolaemia and familial defective apolipoprotein B-100. Annals of Clinical Biochemistry 47(5):487-490

A Taylor, D. Wang, K. Patel, R. Whittall, G. Wood, M. Farrer, RDG. Neely, S. Fairgrieve, D. Nair, M. Barbir, JL. Jones, S. Egan, Y. Lolin, E. Hughes, JA Cooper, SG Hadfield, **G. Norbury**, SE. Humphries. 2009. Mutation Detection Rate and Spectrum in Familial Hypercholesterolaemia Patients in The UK Pilot Cascade Project. Clin Genet 77(6);572-580

Chitty LS, **G. Norbury**, H. White. 2009. Non-invasive prenatal diagnosis: The future of prenatal diagnosis? Reproductive Genetics, RCOG Press ISBN: 978-1-906985-16- pp159-172

Meaney C, **G. Norbury**. 2009. Non-invasive prenatal diagnosis of early onset primary dystonia I in maternal plasma. Prenat Diagn (13):1218-1221.

Taylor A, Patel K, Tsedeke J, Humphries SE, **Norbury G**. 2009. Mutation screening in patients for familial hypercholesterolaemia (ADH). Clin Genet 77(1):97-99.

Taylor A, Martin B, Patel B, Humphries S, **Norbury G**. 2009. Multiplex Ligation amplification analysis (MLPA) to look for deletions and duplications in the LDLR gene in patients with familial hypercholesterolemia. Clin Genet. 76(1):69-75.

Archer C, Emma Baple E, Anand Saggaer, Mann K, **Norbury G**. Case Report: Investigations to determine the inheritance of an apparently rare homozygous ITGB2 mutation in an infant with leukocyte adhesion deficiency (LAD) type II to unrelated parents. CMGS Spring meeting 2009

Taylor A, Patel K, Wang D, Tsedeke J, Martin B, Neely D, Nair D, Barbir M, Egan S, Lolin Y, R Whittall R, Hadfield G, Humphries S, **Norbury G**. Mutation Screening in patient

affected with autosomal dominant hypercholesterolemia – results from the Department of Health (DH) Pilot Project CMGS Spring meeting 2009

Norbury, G., and C. J. Norbury. 2008. Non-invasive prenatal diagnosis of single gene disorders: how close are we? *Semin Fetal Neonatal Med* 13:76-83.

Humphries, S. E., **G. Norbury**, S. Leigh, S. G. Hadfield, and D. Nair. 2008. What is the clinical utility of DNA testing in patients with familial hypercholesterolaemia? *Curr Opin Lipidol* 19:362-368.

Athanasiadis, A. P, M. Zafrakas, P. Polychronou, L. Florentin-Arar, P. Papasozomenou, **G. Norbury**, and J. N. Bontis. 2008. Apert Syndrome: The Current Role of Prenatal Ultrasound and Genetic Analysis in Diagnosis and Counselling. *Fetal Diagn Ther* 24:495-498.

Cregeen D, B.Martin, A.Kumar, S.Taffinder, W. McKenna, S.Jenkins, **G.Norbury**. Mutation screening for hypertrophic cardiomyopathy at the North East Thames Regional Molecular Genetics laboratory. BSHG Sept 2008. *J Med Genet* 2008; 45: suppl 1:2.37

Meaney C. L. Chitty, **G.Norbury** BSHG Sept 2008. Non-invasive prenatal diagnosis (NIPD) of single gene disorders in a Regional Genetics Laboratory *J Med Genet* 2008; 45: suppl 1:6.03

Meaney C., **G Norbury** Chitty L. Non-invasive prenatal genetic diagnosis of single gene disorders in a UK regional genetics service laboratory. ISPD 14th International Conference. 2008

SE Humphries, A Taylor, D Wang, R Whittall, D Neely, D Nair, M Barbir, S Egan, Y Lolin, G Hadfield, **G Norbury** Mutation Detection Rate and Spectrum in Definite (DFH) and Possible (PFH) Patients from the Department Of Health (DH) Pilot Project . *Atherosclerosis* 2008;199:233

S. M. Whitten · S. Clayton · **G. Norbury** · L. S. Chitty. 2007. Etiology of hyperechogenic bowel within a tertiary fetal medicine unit *Ultrasound in Obstetrics and Gynecology* 2007. 30(4):398

Taylor, A., S. Tabrah, D. Wang, M. Sozen, N. Duxbury, R. Whittall, S. E. Humphries, and **G. Norbury**. 2007. Multiplex ARMS analysis to detect 13 common mutations in familial hypercholesterolaemia. *Clin Genet* 71(6):561-568.

Smith, A. J., F. Ahmed, D. Nair, R. Whittall, D. Wang, A. Taylor, **G. Norbury**, and S. E. Humphries. 2007. A functional mutation in the LDLR promoter (-139C>G) in a patient with familial hypercholesterolemia. *Eur J Hum Genet* 15:1186-1189.

Sanchez, J. J., G. Monaghan, C. Borsting, **G. Norbury**, N. Morling, and H. B. Gaspar. 2007. Carrier frequency of a nonsense mutation in the adenosine deaminase (ADA) gene implies a high incidence of ADA-deficient severe combined immunodeficiency (SCID) in Somalia and a single, common haplotype indicates common ancestry. *Ann Hum Genet* 71:336-347.

Chitty L, G Daniels, K Finning, J Hogg, B Ladd, P Martin, C Meaney, **G Norbury**, L Thomasson. Prospective Register of Outcomes of Free-fetal DNA testing (PROOF) – results of first year's audit. *J Med Genet* 2007; 44: suppl 1:S28

Chitty L, J Hogg, P.G Martin, C Meaney, G Norbury. Effectiveness of non-invasive prenatal diagnosis using free fetal DNA in the maternal circulation. *J Med Genet* 2007; 44: suppl 1:S28

Wallace A, D Barton, PA van Bunderen, J Duncan, J Dunlop, S Ma5, J MacPherson11, G Monaghan, J McLuskey, **G Norbury**, Y Patel, H Powell, V Race, M Sweeney, E Thompson, R Treacy, MM Weiss, N Williams, HE White, B Wymer. A Multi -centre Technology Assessment of the Abbott Fragile X Assay *J Med Genet* 2007; 44: suppl 1:S86

Taylor A, D. Neely, S. Fairgrieve, D. Wang, S. Humphries, **G. Norbury**. Is assessment of lipid phenotype needed for cascade genetic screening in Familial Hypercholesterolaemia families? *J Med Genet* 2007; 44: suppl 1S67

Martin B., D.Wang, A.Taylor, S. Humphries **G. Norbury**. J MLPA analysis to detect large deletions and duplications in Familial Hypercholesterolaemia *Med Genet* 2007; 44: suppl 1:S81

Archer C, Quen Mok & **Gail Norbury**. Developing a Diagnostic Service for screening ABCA3, SFTPB and SFTPC genes in patients with Hereditary Pulmonary Surfactant Deficiency *J Med Genet* 2007; 44: suppl 1:S85

Amin N, Farrer M, Robertson L, Mikhalidos DP, **Norbury G**, Humphries S, Nair D. Phenotype or genotype for diagnosis of FH? *Atherosclerosis* September 2007;194 :1:280

Taylor A D Neely S Fairgrieve, Wang D, **Norbury G**. Are both lipid phenotype and genotype needed for cascade genetic screening in FH families? *Atherosclerosis* 2007 ;194 : 1: 280

Taylor A, S.Tabrah, D. Wang, M. Sozen, N.Duxbury, R. Whittall, S.Humphries, **G. Norbury**. Multiplex ARMS analysis to detect 13 common mutations in familial hypercholesterolemia. *CMGS* 2007

Meaney C, B.Mistry, A. Turner, L. Chitty, **G.Norbury**. Diagnostic experience of ffDNA analysis for prenatal sex determination and testing for Achondroplasia. *CMGS* Spring 2007

Norbury, G., and C. J. Norbury. 2006. DNA analysis: what and when to request? *Arch Dis Child* 91:357-60.

G. Norbury C. Meaney B.Ladd. Technical challenges in non-invasive prenatal diagnosis. New and Developing Technologies for Genetic Diagnostics 2006. NGRL(Wessex). http://www.ngrl.org.uk/Wessex/downloads/tm06/TM2%20S4_4_Norbury.pdf

Chitty LS, J Hogg, C Meaney, **G Norbury**. Non-invasive prenatal diagnosis of fetal sex using free fetal DNA in the maternal circulation – effect on obstetric management. 16th World Congress on Ultrasound in Obstetrics and Gynaecology. London 2006.

Farrer M, D. Nair, A.Taylor, R. Whittall, **G. Norbury**, S. Humphries. Genetic Test Results In FH Need Careful Interpretation. *HEART* UK 2006

Sozen M, R. Whittall, N.J.Duxbury, M.B.T.Webb, **G. Norbury**, A. Taylor, S.E. Humphries. An ARMS-Based Diagnostic Kit For Genetic Testing For Familial Hypercholesterolemia In the UK XIV International Symposium on "ATHEROSCLEROSIS" (Rome, June 18-22, 2006)

Differ A, T.Linton-Willoughby, **G.Norbury**. Mutation screening of the GNAS1 gene *CMGS* Spring 2006

Cregeen D, S.Tabrah, **G.Norbury** Mutation screening of the IRF6 gene for Van der Woude and Popliteal Pterygium syndromes. CMGS Spring 2006

McCann, E., S. B. Kaye, W. Newman, **G. Norbury**, G. C. Black, and I. H. Ellis. 2005. Novel phenotype of craniosynostosis and ocular anterior chamber dysgenesis with a fibroblast growth factor receptor 2 mutation. *Am J Med Genet A* 138A:278-281.

Gilmour KC, R. Peraj, J.Hobbs, L. Haddad, C. Meaney, V. Aldridge, **G.Norbury**, H. Bobby Gaspar and C.M. Cale. Familial Hemophagocytic Lymphohistiocytosis: Protein based screening of perforin, munc 13-4 and SAP. Primary immunodeficiency Network meeting, York. 2005.

Tabrah S, A Taylor, S Humphries, **G Norbury**. A strategy for molecular genetic mutation screening for familial hypercholesterolaemia. *J Med Genet* 2005; 42: suppl 1:S110

Norbury G, C Meaney, B Lad, T Stojilkovic-Mikic, M Whitten, J Hyett, L Chitty. Reliability of prenatal sexing by non-invasive diagnosis using real time PCR. *J Med Genet* 2005; 42: suppl 1:S100

Aldridge VL, K.C. Gilmour, B. Gaspar, A. Thrasher, A. Coffey, **G. Norbury**. Polyadenylation Signal Site Mutation in a Boy with X-Linked Severe Combined Immunodeficiency ESHG vol 13 2005; suppl 1

Tabrah S, A Taylor, S Humphries, **G Norbury**. Utility of genetic testing for Familial Hypercholesterolaemia. CMGS Spring Meeting 2005

Farrer JM, D Nair, A Taylor, **G Norbury**, SE Humphries. Does DNA information compliment lipid measures in cascade testing in familial hypercholesterolaemia Heart UK 19th annual meeting. Atherosclerosis 2005

Hendriksz CJ, **G. Norbury**, S. Tabrah, A. Taylor, and S. E. Humphries. 2004. Homozygous hypercholesterolaemia and Ezetimibe: a case report. *Acta Paediatr* 93:280-282.

Haddad L, G. Monaghan, C. Cale, K. Gilmour, **G. Norbury** Development of a UK diagnostic service for familial hemophagocytic lymphohistiocytosis (FHL). BSHG 2004 *J Med Genet* 41 2004 Suppl no 1

Cregeen D, **G. Norbury**. Impact of pre-screening Ashkenazi Jewish BRCA1/2 mutations on patient service and laboratory workload in North East Thames. BSHG 2004 *J Med Genet* 2004 41 Suppl no 1

Taylor, A E. Green, K. Pearce, S. Tabrah, E. Young, **G.Norbury**. Mutation Screening of the ARSA gene in Metachromatic Leukodystrophy BSHG 2004 *J Med Genet* 2004 41: Suppl 1:

Stopps K, Q. Mok, L. Nogee, **G. Norbury**. Diagnostic testing for Hereditary Surfactant Protein B Deficiency. BSHG 2004 *J Med Genet* 2004 Suppl no 1

Rossi M, R. L. Jones, **G. Norbury**, A. Bloch-Zupan, and R. M. Winter. 2003. The appearance of the feet in Pfeiffer syndrome caused by FGFR1 P252R mutation. *Clin Dysmorphol* 12:269-274.

Gilmour KC., D. Walshe, S. Heath, G. Monaghan, S. Loughlin, T. Lester, **G. Norbury**, C. M. Cale. 2003. Immunological and genetic analysis of 65 patients with a clinical suspicion of X linked hyper-IgM. *Mol Pathol* 56:256-262.

Hoskins B, Holden M, Taffinder S, **Norbury G**, Jones R, Vant Hoff W, Pandya P, Winter R, Scrambler P, Beales P. Molecular disease confirmation in a case of Bardet-Biedl syndrome, prenatal diagnosis in a sibling and segregation of genitourinary malformations in unaffected relatives with heterozygous mutation. *AJHG*2003; 73(S) 5:169

Jones RL, McMahon C, Craig E, Stopps K, Moxer M, Winter RM, **Norbury G**. Five years' experience of a diagnostic service for craniofacial syndromes. *J Med Genet* 2003; 40: suppl 1:S62, *AJHG*2003; 73(S) 5:403

Jones RL, McMahon C, Craig E, Monaghan G, Stopps K, Boxer M, Winter RM, **Norbury G**. Findings of a molecular genetic diagnostic service for skeletal dysplasia syndromes. *J Med Genet* 2003; 40: suppl 1:S71

Stopps K, King W, **Norbury G**. Taylor R, Flinter F. A rare bimodal distribution of expanded alleles in a patient with myotonic dystrophy type 1. *J Med Genet* 2003; 40: suppl 1:S71

Loughlin S, Rickard S, Jenkins L, Bitner-Glindzicz M, **Norbury G**. Molecular genetic testing for brachio-oto-renal syndrome at NE Thames Regional Molecular Genetics. *J Med Genet* 2003; 40: suppl 1:S80

Jones RL, Craig E, McMahon C, Monaghan G, Barber R, Boxer M, Winter R, **Norbury G**. Five years' experience of a diagnostic service for craniofacial and skeletal dysplasia syndromes. *CMGS* Spring 2003.

Taylor AE, Attard M, Tabrah S, **Norbury G**, Van't Hoff W, Town M. Mutation analysis of the CTNS gene for cystinosis. *ESHG* 2003;11 supp1

Cregeen D, **Norbury G**, Logan R, Jenkins L. Fragile X syndrome; An expansion/contraction mosaic *ESHG* 2003;11 supp1

Aldridge V, Jenkins L, Bitner-Glindzicz, **Norbury G**. Normal live born of offspring of an Angelman syndrome mother with Prader Willi syndrome phenotype *ESHG* 2003;11 supp1

Mort O, Craig E, Hyett J, Chitty L, **Norbury G**. Use of free fetal DNA for non-invasive fetal sex determination. *ESHG* 2003;11 supp1

Porter, T. R., F. M. Richards, R. S. Houlston, D. G. Evans, J. A. Jankowski, F. Macdonald, **G. Norbury**, S. J. Payne, S. A. Fisher, I. Tomlinson, and E. R. Maher. 2002. Contribution of cyclin d1 (CCND1) and E-cadherin (CDH1) polymorphisms to familial and sporadic colorectal cancer. *Oncogene* 21:1928-33.

Deshpande C, Jenkins L, **Norbury G**, Middleton-Price M, Bitner-Glindzicz M. Significance of a single connexin 26 mutation in a deaf child. *J Med Genet* 2002;39: suppl 1:1.45

Taylor A, Tabrah S, Young E, Mitchison M, **Norbury G**. Mutation screening of the CLN1 gene for Batten's disease *J Med Genet* 2002;39: suppl 1:3.81

Taylor A, Tabrah S, Cranston T, **Norbury G**. Mutation Screening for autosomal recessive malignant osteopetrosis *J Med Genet* 2002;39:suppl 1:3.08

White, S. M., A. Lucassen, and **G. Norbury**. 2001. Cystic fibrosis: a further case of an asymptomatic compound heterozygote. *Am J Med Genet* 103:342-343.

Lipton, L., H. J. Thomas, R. A. Eeles, R. S. Houlston, M. Longmuir, R. Davison, S. V. Hodgson,

V. A. Murday, **C. G. Norbury**, C. Taylor, and I. P. Tomlinson. 2001. Apparent Mendelian inheritance of breast and colorectal cancer: chance, genetic heterogeneity or a new gene? *Fam Cancer* 1:189-195.

Norbury G, Jones DC, Lucassen L, Crawford G, Jones L, Tomlinson IP. A practical approach to the diagnosis of hereditary non-polyposis colon cancer. *J Med Genet* 2001;38: supp 1:3.07

King W, Seller A, **Norbury G**, Canham N, Sachdev R. Mutation scanning in UBE3A using DGGE (2001). CMGS Spring meeting.

Martindale JE, **Norbury G**, Seller A. Mutation analysis of the BRCA1 gene by DGGE (2001). CMGS Spring meeting.

Woodford-Richens, K., S. Bevan, M. Churchman, B. Dowling, D. Jones, **C. G. Norbury**, S. V. Hodgson, D. Desai, K. Neale, R. K. Phillips, J. Young, B. Leggett, M. Dunlop, P. Rozen, C. Eng, D. Markie, M. A. Rodriguez-Bigas, E. Sheridan, T. Iwama, D. Eccles, G. T. Smith, J. C. Kim, K. M. Kim, J. R. Sampson, G. Evans, S. Tejpar, W. F. Bodmer, I. P. Tomlinson, and R. S. Houlston. 2000. Analysis of genetic and phenotypic heterogeneity in juvenile polyposis. *Gut* 46:656-660.

White, S. M., S. S. Gubbay, **C. G. Norbury**, E. M. Rosser, and S. M. Huson. 2000. Diagnostic re-evaluation of a case of 'cerebellar atrophy with Huntington's disease'. *J Neurol Sci* 174:47-8.

Boyd P, **Norbury G**. Echogenic bowel and cystic fibrosis. *J Med Genet* 1999;36: supp1:SP2 (joint oral presentation).

Wang, Z. J., F. Taylor, M. Churchman, **G. Norbury**, and I. Tomlinson. 1998. Genetic pathways of colorectal carcinogenesis rarely involve the PTEN and LKB1 genes outside the inherited hamartoma syndromes. *Am J Pathol* 153:363-366.

Norbury G, Boyd P, Pironne P, Chamberlain P. Isolated fetal echogenic bowel as an indication for molecular screening for cystic fibrosis. *J Med Genet* 1998;35 Supp 1:S55.

Rubinsztein, D. C., J. Leggo, M. Chiano, S. Korn, A. Dodge, **G. Norbury**, E. Rosser, and D. Craufurd. 1997. Homozygotes and heterozygotes for ciliary neurotrophic factor null alleles do not show earlier onset of Huntington's disease. *Neurology* 49:890-892.

Rubinsztein, D. C., J. Leggo, M. Chiano, A. Dodge, **G. Norbury**, E. Rosser, and D. Craufurd. 1997. Genotypes at the GluR6 kainate receptor locus are associated with variation in the age of onset of Huntington disease. *Proc Natl Acad Sci U S A* 94:3872-6.

Hindley NJ, **Norbury CG**, Jobst KA, Rosser EM, Huson SM, Pearce MJ, King EM. Late-onset Huntington's disease as a cause of dementia: where should the clinician's index of suspicion lie? *Int J Geriatric Psychiatry* 1996;11:729-733.

Rosser EM, McDonald B, **Norbury G**, Huson SM. Huntington's disease after the gene - the diagnostic difficulties remain. *J Med Genet* 1996;33 Supp1 S30.

Norbury G, Lunt P, Tyrrell J, Ng WC, Moss TH, Davis M, Stevens D. Gene dosage effect at the Dentatorubral-pallidoluysian atrophy gene locus. *European J Human Genetics* 1996;4, Suppl 1 15 (oral presentation by Norbury).

Woods, G., G. Black, and **G. Norbury**. 1995. Male neonatal death and progressive weakness and immune deficiency in females: an unknown X linked condition. *J Med Genet*

32:191-196.

Norbury CG, Hindley NJ, Jobst KA, Rosser EM, Huson SM, Pearce MJ, King EM. Late-onset Huntington's disease as a cause of dementia: where should the clinician's index of suspicion lie? (1995) 16th World Federation for Neurology HD Meeting, Leuven and British Medical Genetics Conference.

Shiwach, R. S., and **C. G. Norbury**. 1994. A controlled psychiatric study of individuals at risk for Huntington's disease. *Br J Psychiatry* 165:500-505.

Rosser, E., S. M. Huson, and **G. Norbury**. 1994. Prenatal, presymptomatic, and diagnostic testing with direct mutation analysis in Huntington's disease. *Lancet* 343:487-488.

Barnes, P. R., D. Hilton-Jones, **G. Norbury**, A. Roberts, and S. M. Huson. 1994. Incorrect diagnosis of myotonic dystrophy and its potential consequences revealed by subsequent direct genetic analysis. *J Neurol Neurosurg Psychiatry* 57:662.

Norbury G, Seller A, Huson S. Diagnostic experience of analysis for myotonic dystrophy (1994). *J Med Genet*. 31. 171-172.

Rosser E, **Norbury G**, Glew R, Jones L, Huson S. Direct mutation analysis for Huntington's disease (HD) – the Oxford experience (1994). *J Med Genet* 1994:148.

Norbury CG. Operational computer system for a service DNA laboratory (1993). CMGS, Manchester. (oral presentation)

Rosser E, **Norbury G**, Galliard A, Glew R, Seller A, Huson S. Research and Service - are they compatible? (1993). 15th International World Federation of Neurology, Boston.

Norbury G, Seller A, Roberts A, Huson S. Experience of DNA analysis for myotonic dystrophy (1993). CGS, Leeds.

Myring, J., A. L. Meredith, H. G. Harley, G. Kohn, **G. Norbury**, P. S. Harper, and D. J. Shaw. 1992. Specific molecular prenatal diagnosis for the CTG mutation in myotonic dystrophy. *J Med Genet* 29:785-788.

Norbury CG, Kiernan E, Miciak A. Analysis of the expansion associated with myotonic dystrophy from fetuses predicted to be at high risk (1992). CGS, Nottingham.

Kiernan E, Wilcocks T, Craig I, Mackenzie A, MacDonald F, Dixon J, **Norbury G**, Edwards J. Use of tandem repeats in two loci near to the locus for myotonic dystrophy (1991). CGS, Bristol.

Lindenbaum RH, Woods CG, **Norbury CG**, Povey S, Ryslecki G. A case report of an individual with maternal disomy of chromosome 4 and isochromosomes 4p and 4q. *Am J Hum Genet* 1991;49; supp 285,1582. (oral presentation by Norbury)

Miciak A, Keene A, **Norbury G**, Hopkin J, Edwards J. The distribution of deletions in boys with Duchenne and Becker muscular dystrophy and the application of pulsed field gel electrophoresis to carrier detection in females (1990). ACC, Manchester.

Norbury CG, Miciak A, Lindenbaum RH. Practical considerations in carrier detection and prenatal diagnosis of DMD using conventional techniques (1988). *Molecular Probes: Technology and Medical Applications Meeting*. Florence, Italy.

Norbury C.G. 1987. Simplified method for the determination of plasma cotinine using gas chromatography-mass spectrometry. *J Chromatogr* 414:449-453.

Norbury CG, Fry DE. The effect of smoking on prolactin levels in subfertile women *Annal. Clin. Biochem Suppl.* (1987) (oral presentation by Norbury).

Cummings, B., M. R. Kaser, **G. Wiggins**, M. G. Ord, and L. A. Stocken. 1982. Beryllium toxicity. The selective inhibition of casein kinase 1. *Biochem J* 208:141-146.