IN MEMORIAM

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Professor Robin Michael Winter: 1950–2004. An appreciation

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Robin Winter, who made major contributions to Medical Genetics in Europe and beyond, has died at the age of 53 years of oesophageal cancer. Robin's main medical interest was Dysmorphology and he used his unique skills in clinical observation to delineate many previously unrecognised syndromes and, as technology developed, initiated programmes of research to identify the genes and pathways involved.

Robin came from a medical family in the Wirral in the north west of England and undertook his preclinical Studies at University College London. Before his clinical studies, he took an intercalated BSc Course in Genetics at the Galton Laboratory, University College London where his fascination with genetics and patterns of malformations began. Following 3 years postgraduate training in medicine and paediatrics, he spent a period as Post-Doctoral Fellow with Walter Nance at the Department of Human Genetics, Medical College of Virginia.

In 1978, he returned to the UK and took up one of the first three training posts in Clinical Genetics in the UK. Ian Young and myself were appointed to the other posts. Robin was based at the Kennedy-Galton Centre, Harperbury Hospital where Lionel Penrose did much of his work and at Northwick Park Hospital, Harrow. I remember we used to take slides of puzzling patients to the Clinical Genetics Society and gather round in a corner to discuss them. Out of these discussions came several papers describing rare entities. In time we arranged lunchtime, and then afternoon, and finally whole day meetings that evolved into the UK Dysmorphology Club. Robin was appointed Consultant Clinical Geneticist in 1981 and, with MRC funding, in 1988 head of the Dysmorphology Research Group at the Clinical Research Centre, Northwick Park Hospital. In 1992, he moved to the Institute of Child Health, and Hospitals for Sick Children, Great Ormond Street, London where he spent the rest of his professional life and in 1994 was appointed Professor of Dysmorphology and Clinical Genetics.

Robin published 280 papers in peer-reviewed journals and many reviews, chapters and books. Important reviews focussed on the interplay between clinical observations and the laboratory.^{1–3} He was as much 'at home' with scientists as clinicians and was respected by both groups. A major theme in his research was the homology between



mouse and human at clinical and molecular levels and his group has proved several of his hypotheses, for example, that Fraser syndrome results from mutations in a gene which is homologous to the genes which, when mutated in mice, cause the 'bleb' phenotype.^{4,5} His work on craniosynostosis syndromes was founded on large collections of patient samples gathered before the development of techniques to investigate these. With Sue Malcolm and Willie Reardon and in collaboration with Andrew Wilkie's and Max Muenke's groups major advances were made in elucidating the molecular basis of Crouzon, Apert and related syndromes.^{6–11}

The London Dysmorphology Database series that he developed with Michael Baraitser are essential tools for experts and trainees, particularly in Dysmorphology but increasingly for other disciplines. Both the Dysmorphology and the Neurogenetics databases have been through multiple editions since the mid-1980s and contain much really helpful information written by experienced clinicians for clinicians. These databases have received many awards including 1995 Royal Society of Medicine, Medical Book Awards and the 1998 BMA Medical Book Awards – electronic form categories. The London Medical Databases will continue to be updated, and developed (www.lmdatabases.com, info@lmdatabases.com).

Robin Winter wrote several books on dysmorphology and contributed numerous chapters to others. With Michael Baraitser and me he founded and edited the journal *Clinical Dysmorphology*.

Robin was liked and respected by colleagues from all over the world and contributed to many international meet-



ings. He was asked for an opinion hundreds of times every year, increasing in volume since the advent of email, and he replied promptly and patiently. He served on the Scientific Programme Committee of the European Society of Human Genetics and was honoured by the Society with the award of the Baschirotto Prize in 2001. He was also awarded the International Querci Prize for Paediatric Research for 1997–2000 and was elected a Fellow of the Academy of Medical Sciences in 2000. He was invited to give many prestigious lectures at meetings including the 1st Annual Meeting of the American College of Medical Genetics and the 1994 Distinguished Speakers Symposium at the American Society of Human Genetics.

Robin gave an outstanding service to the many families who consulted him. He was much in demand as a mentor; numerous visiting trainees from the UK and overseas benefited from spending time in his department. A large number of junior clinical geneticists from all over the world, attending the 'practical syndromology' sessions at the ESHG annual conferences, the 'curbstone consultations' at the ASHG annual meetings, or the 'Sesti Levante' school throughout the years, have been educated by Robins insightful teaching. The respect of his colleagues in the UK was reflected in his election as President of the Clinical Genetics Society from 2000 to 2002.

In spite of his many achievements, Robin Winter was a modest and, sometimes retiring, man with an incredible rather 'British' understated sense of humour. It was hard not to laugh out loud at his dry comments and asides at meetings and many of us have happy memories of his witty speeches at meetings and colleagues' retirement parties. It is difficult for many of us to imagine professional life without him but the messages sent from colleagues around the world following his death, bear testament to his enduring legacy for the profession.

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- 1 Winter RM: Recent molecular advances in dysmorphology. *Hum Mol Genet* 1995; **4**: 1699–1705.
- 2 Winter RM: What's in a face? Nat Genet 1996; 12: 124-129.
- 3 Winter RM: Analysing human developmental abnormalities. *BioEssays* 1996; 18: 965–971.
 4 Winter RM: Fraser syndrome and mouse 'bleb' mutants. *Clin*
- Genet 1990; 37: 494–495.
- 5 McGregor L, Makela V, Darling SM *et al*: Fraser syndrome and mouse blebbed phenotype caused by mutations in FRAS1/Fras1 encoding a putative extracellular matrix protein. *Nat Genet* 2003; **34**: 203–208.
- 6 Reardon W, Winter RM, Rutland P, Pulleyn LJ, Jones BM, Malcolm S: Mutations in the fibroblast growth factor receptor 2 gene cause Crouzon syndrome. *Nat Genet* 1994; **8**: 98–103.
- 7 Muenke M, Schell U, Hehr A *et al*: Mutations in the fibroblast growth factor receptor-1 gene in Pfeiffer syndrome. *Nat Genet* 1994; **8**: 269–274.
- 8 Wilkie AOM, Slaney SF, Oldridge M *et al*: Apert syndrome (craniosynostosis/syndactyly) results from localised mutations of FGFR2 and is allelic with Crouzon syndrome. *Nat Genet* 1995; 9: 165–172.
- 9 Rutland P, Pulleyn LJ, Reardon W *et al*: Identical mutations in the FGFR2 gene cause both Pfeiffer and Crouzon syndromes. *Nat Genet* 1995; **9**: 173–176.
- 10 Oldridge M, Wilkie AOM, Slaney SF *et al*: Mutations in the third immunoglobulin domain of the fibroblast growth factor receptor-2 gene in Crouzon syndrome. *Hum Mol Genet* 1995; 4: 1077–1082.
- 11 Reardon W, Wilkes D, Rutland P *et al*: Craniosynostosis associated with FGFR3 pro250arg mutation results in a range of the clinical presentations including unisutural sporadic craniosynostosis. *J Med Genet* 1997; **34**: 632–636.