John MacMillan, M.D. (1959–2014): an inspiring example of a community clinical geneticist

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John Crichton MacMillan was a determined and outspoken person. By the time he was four, after having accompanied his father Allan to work, where he was a charge nurse, John announced his intention to become a doctor. He was also passionate and humorous, a person of many interests and a wonderer, who found his later and deepest passion in serving the Aboriginal communities in Australia, providing genetics education and counselling to the families afflicted with Machado-Joseph disease (MJD).

Born in Aberdeen, where he graduated from the University School of Medicine, he did his medical training between his native Scotland and England. He completed an intercalated

degree in pathology and started working as a doctor in Harrogate Yorkshire, in 1983. From 1985 to 1995, he moved between Invercargill and Dunedin, in New Zealand (where he began his training in neurology), and Cardiff, at the University Hospital of Wales (where he qualified in medical genetics and did research in neurogenetics, with Peter Harper and Angus Clarke). He was first author on the *Lancet* paper that established the correlation between the larger expansions in Huntington disease and the more severe juvenile forms. In July 1995, he moved to Brisbane, Australia, where he has been working since, as a clinical geneticist and professor, and founded the largest medical genetics service in the country.

Many will remember him as the host of the International Congress of Human Genetics, August 2006, in which he printed his remarkable and joyful personality (he had been trying to take it to Australia, since the 1990s). I believe that was where I first met him, and we talked extensively about MJD, which was known to affect several Aboriginal families in Arnhem Land, Northern Territories.

John then became involved with the MJD Foundation (MJDF) since its founding in 2008, being part of its Research Advisory Board and medical team. I recall very vividly his genetic counselling sessions under the mango trees (Fig. 1), at the time of the launch of the MJDF at Angurugu, Groote Eylandt. Since then, he kept going several times every year to various parts of Arnhem land, but also to central Australia, to follow MJD families.

He worked within these Aboriginal communities to provide genetic education and counselling to affected families, helped training care workers and family caregivers, organizing workshops and information sessions to local general practitioners and other healthcare professionals, and in the production of educating materials about genetics and MJD, in a culturally appropriate manner.

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Fig. 1 John MacMillan talking to a group of women from MJD families, at Angurugu, Groote Eylandt, at the time of the launch of the MJD Foundation, 2008 (photo by Kate Freestone)

His commitment to the Aboriginal communities went as deep as him graduating in Yolngu Language and Culture, from the Charles Darwin University, in 2013. Before, in 2009, he had graduated also in Nutrition Medicine.

John met Sarah at a Guy Fawkes party, November 1979, just a few days before he turned 20. They had three children, Ewen, Liusadh (Lucy) and Kieran, born in Invercargill (New Zealand), Cardiff (Wales) and Brisbane (Queensland), in that order. John often joked he could not afford moving to still another country, as a new baby seemed to arrive each time that happened.

John MacMillan touched many lives before losing his battle to cancer, aged only 55, on 21 December 2014. The MJDF will continue his genetic education work, under the “John MacMillan Indigenous Australian Genetic Education Program”.

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