Aplastic anaemia and telomerase RNA mutations

Sir—Tom Vulliamy and colleagues (June 22; p 2168) report the association between mutations in the telomerase RNA gene (hTR) and aplastic anaemia.

In a previous study by these authors, a rare autosomal dominant type of dyskeratosis congenita (an inherited type of aplastic anaemia) was associated with very short telomeres and mutations in the hTR gene. Patients with aplastic anaemia also present shorter telomere length than age-matched controls.

We have assessed 42 unselected patients with acquired aplastic anaemia (aged 2–57 years, median 26 years) for mutations in the hTR gene. Of these patients, ten were classified as having moderate aplastic anaemia, 20 as having severe aplastic anaemia, and 12 with very severe aplastic anaemia. In 17 patients, aplastic anaemia was attributed to drug or chemical exposure, two to post-hepatitis aplastic anaemia, and in 12, a paroxysmal nocturnal haemoglobinuria (PNH) clone was detected by flow cytometry.

None of the three hTR mutations described by Vulliamy and colleagues was noted in our series. This discrepancy is intriguing. However, as pointed out by the authors, their study was done in patients from one institution, which is the only referral centre for the treatment of aplastic anaemia in an area covering five million of the population of southeast Brazil. Moreover, our patients’ population included variable disease presentation, such as drug-induced, hepatitis-associated, and PNH-associated aplastic anaemia.

In congenital aplastic anaemia, especially dyskeratosis congenita, short telomere length has a role in the pathophysiology of the disease, and some cases are linked to hTR gene mutations. However, although telomeres are also shorter in acquired aplastic anaemia, our data indicate that mutations in the hTR gene are not involved in the pathogenesis of the disease, in which bone-marrow failure is the result of immune-mediated destruction of haematopoiesis.

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Time to do the right things right: Nigerian health care

Sir—The June 8 Reportage from Sally Hargreaves is most welcome. As a Nigerian who trained at the nation’s oldest medical school (formerly University College London, Ibadan Campus; now, University of Ibadan), I could not help identifying with most of her observations.

During my own stint at the Lagos University Teaching Hospital, I came face to face with the appalling man-made decay that had befallen the Nigerian health-care system. I was prompted to pursue postgraduate studies in health services and systems research.

As a country that accounts for about 50% of the west African economy, Nigeria cannot afford to drown the rest of the region. Bearing in mind that health and wealth are highly correlated, it becomes urgent that Nigeria addresses its abysmal health and social services to jump start its non-petroleum dependent economy and create a robust society of healthy people who can contribute to nation building.

Like the rest of Africa, Nigeria presents a fertile ground for research and seemingly miraculous interventions if the right initiatives are taken in a coordinated way. However, currently, there are no up-to-date or accurate data on the state of the health of Nigerians. Research and interventions are, therefore, made difficult. This situation complicates and is further confounded by the near chaotic, inconsistent, foreign aids and programmes aimed at tackling the difficulties.

The Nigerian leadership must rise to the occasion and get its priorities right: restoration of public confidence; increase in investment in health, education, research, and related sectors; creation of sound health and demographic databases; proper needs assessment; the abandoning of inappropriate projects for the identified needs; stimulation of a development of a