

(The genetics of) breast cancer

Big news recently was Angeline Jolie's 'medical choice' to have a preventive double mastectomy.^[1] Co-incidentally the *SAMJ* had received articles dealing with breast cancer, including its genetic diagnosis, and several relating to maternal and neonatal health. We opted to make this August issue a Women's Month edition.

The incidence of breast cancer in sub-Saharan Africa (SSA) is reportedly low at 20 cases per 100 000 population, compared with 90/100 000 in the West. However, this undoubtedly reflects under-diagnosis, especially since breast cancer is closing in on cervical cancer as the most common malignancy affecting women, and the incidence rates appear to be rising.

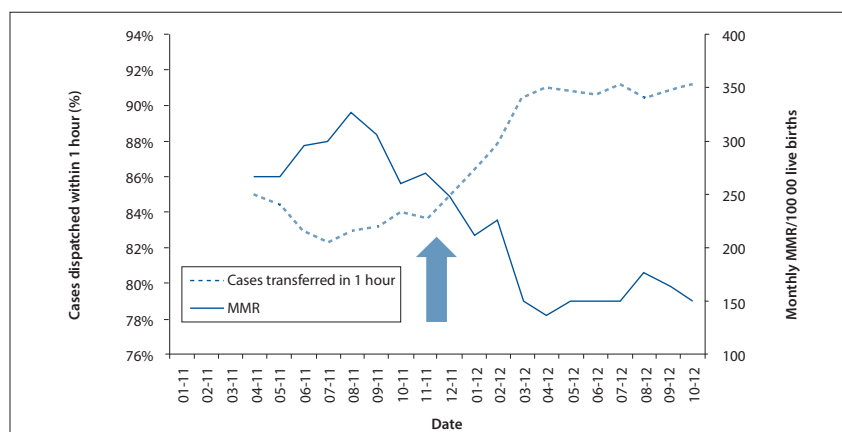
In developed countries, breast cancer is a disease of older women. However, in SSA, more than 50% of patients are premenopausal. In developing and resource-poor countries women present very late – in the Eritrean study in this issue,^[2] over 70% of patients sought consultation more than 1 year after noticing symptoms. Not surprisingly 1 in 5 of such women had advanced stage disease and distant organ metastasis. Many women had resorted to 'traditional' therapy (e.g. herbal medicine, holy water, soil), which further exacerbated their poor outcome and prognosis.

The situation is happier in our own country – as our vivid guest editorial notes. We are fortunate to have available modern diagnostic technology, including sophisticated genetic testing.^[3]

An editorial from Urban^[4] contains advice for the generalist on how to avoid ordering genetic tests that, without appropriate genetic counselling^[5] of patient and family members, would prove frivolous – 'recreational' is his term – and even potentially harmful.

Mother and child health – outlook for South Africa

South Africa (SA) is a long way off from meeting Millennium Development Goals 4 and 5 by 2015. However, an editorial from Burton^[6] offers a glimmer of hope, and one from Lloyd and de Witt,^[7] shows – based on their experience at the Steve Biko Academic Hospital – how simple measures undertaken by midwives, could improve outcomes. On a related note, Schoon,^[8] writing of the experience in the Free State, where maternal mortality rates are among the highest in South Africa, provides a valuable insight: by the simple expedient of arranging that there be dedicated inter-facility transport for pregnant mums in trouble, maternal mortality was dramatically reduced (see figure above).



Inter-facility vehicles dispatched v. maternal mortality (from Schoon, in this issue).^[8]

HIV and maternal mortality

The HIV pandemic has become increasingly feminised, with HIV prevalence among antenatal clinic attendees at 30%, (even higher in the Pietermaritzburg area, at 42%). HIV/AIDS is thus the major cause of maternal deaths. Given the scarcity of intensive care unit (ICU) facilities in our public sector environment, it seems reasonable to ask, as Ngene *et al.*^[9] do, **whether HIV-positive pregnant women should receive ICU care or not.** In a prospective study, they determined the maternal and fetal outcomes of HIV-positive and -negative patients, whether pregnant or postpartum, and constituting almost 10% of all admissions to ICUs. The chief pre-ICU admission diagnosis was pre-eclampsia/eclampsia syndrome, more common among HIV-negative than HIV-positive patients. Among HIV-positive patients, pneumonia, reflecting immune suppression, was the most common diagnosis. All maternal and fetal outcomes showed a worse trend with HIV-positive v. HIV-negative patients. These findings are likely to favour HIV-negative pregnant women, with their better-expected outcomes, over their HIV-positive counterparts for admission to ICU. Larger studies are urgently needed to investigate these trends more completely. The authors believe that, until such studies are done, HIV sero-status should **not** be used as an isolated determinant of admission to ICU.

High-risk patients, expertly guided, can benefit from PrEPs

When it comes to treatment, very few HIV clinicians dispute the overall efficacy of increasing the CD4⁺ count threshold from 350 to 500 in initiating antiretroviral therapy (ART) (a probability from 1 April 2014), or the value of fixed-dose combinations. The same goes for prevention (e.g. circumcision, vaginal-based microbicides best applied

before and after predictable sex, or condoms/femidoms). However, when it comes to treatment-as-prevention, ranging from ART at diagnosis of HIV to pre-exposure prophylactics (PrEPs), the debate heats up, especially with the latter. The PrEP issue was thoroughly vented at the June/July national AIDS conference in Durban. *Izindaba* reports^[10] from experts at the coalface on just how, where and for whom it makes the most sense, and most critically, which high-risk groups might be most adherent. Much of it is based on pragmatic common sense, e.g. (generally) why would a discordant couple use PrEPs when antiretrovirals with condoms pretty much do the trick? Field studies show appalling adherence to PrEPs, but in the real world, sex workers are already showing 95% adherence to ART (and they are hyper-aware of the risks of their trade). Used highly selectively, it is a handy addition to our self-defence weaponry.

JS

1. Jolie A. My Medical Choice. The New York Times. 14 May 2013. http://www.nytimes.com/2013/05/14/opinion/my-medical-choice.html?_r=0 (accessed 9 Jul 2013).
2. Tesfamariam A, Gebremichael A, Mufunda J. Breast cancer clinicopathological presentation, gravity and challenges in Eritrea, East Africa: Management practice in a resource-poor setting. *S Afr Med J* 2013;103(8):526-528. [<http://dx.doi.org/10.7196/SAMJ.6829>]
3. Grant KA, Apffelstaedt JP, Wright C, et al. MammaPrint Pre-screen Algorithm (MPA) reduces chemotherapy in patients with early-stage breast cancer. *S Afr Med J* 2013;103(8):522-526. [<http://dx.doi.org/10.7196/SAMJ.7223>]
4. Urban M. Clinical issues in genetic testing for multifactorial diseases. *S Afr Med J* 2013;103(8):517. [<http://dx.doi.org/10.7196/SAMJ.7232>]
5. Dandara C, Greenberg J, Lambie L, et al. Direct-to-consumer genetic testing: To test or not to test, that is the question. *S Afr Med J* 2013;103(8):510-512. [<http://dx.doi.org/10.7196/SAMJ.7049>]
6. Burton R. Maternal Health: There is cause for optimism. *S Afr Med J* 2013;103(8):520-521. [<http://dx.doi.org/10.7196/SAMJ.7237>]
7. Lloyd LG, de Witt TW. Neonatal mortality in South Africa: How are we doing and can we do better? *S Afr Med J* 2013;103(8):518-519. [<http://dx.doi.org/10.7196/SAMJ.7200>]
8. Schoon MG. Impact of inter-facility transport on maternal mortality in the Free State Province. *S Afr Med J* 2013;103(8):534-537. [<http://dx.doi.org/10.7196/SAMJ.6828>]
9. Ngene NC, Moodley J, Songca P, et al. Maternal and fetal outcomes of HIV-infected and non-infected pregnant women admitted to two intensive care units in Pietermaritzburg, South Africa. *S Afr Med J* 2013;103(8):543-548. [<http://dx.doi.org/10.7196/SAMJ.6590>]
10. Bateman C. Neglected, high-risk groups a top priority in AIDS prevention/treatment. *S Afr Med J* 2013;103(8):503-505. [<http://dx.doi.org/10.7196/SAMJ.7201>]