Molecular genetic tests are used increasingly for accurate diagnosis of single gene disorders or to subcategorise polygenic or multifactorial diseases according to their genetic basis. It is likely that the focus of genetic testing will change in the near future. While family-based screening for relatively rare genetic conditions remains important, it seems inevitable that genetic testing will be increasingly performed to identify mutations of low penetrance that occur relatively frequently in the general population. This is due to the fact that nutrition and/or other lifestyle factors could determine whether persons with genetic risk factors will develop the associated disease.

A network of dieticians trained in nutritional genetics has been established countrywide (www.genecare.co.za/HealthGen.html) to provide a support base to medical practitioners who would refer patients for dietary intervention based on test results. A question-answer service is also available and several genetic counselling facilities have been established (www.genecare.co.za/counsel.html) to assist health professionals who offer genetic testing in their practices.

What does the future hold? Genetic discoveries have led to the development of commercially available genetic tests for more than 1000 clinically-useful DNA alterations (www.geneclinics.org). As this number is expected to double over the next five years, multiplex genetic testing will replace many single gene tests. Such strip-assay tests are already available in South Africa for genetically heterogeneous diseases and complex conditions such as hereditary haemochromatosis and cardiovascular disease. Ultimately, new micro-array technology that allows simultaneous analysis of hundreds or thousands of genes will be applied in clinical practice. The increasing number of genetic tests that move from the research setting into clinical practice will require health care providers without specialty training in genetics to become “practitioners of genomic medicine.” Utilisation of the available technology will be driven by patient demand for genetic testing, which can be expected to increase significantly as test results are linked directly to key intervention strategies. By relating a person’s genetic make-up with the overall risk profile a highly individualised intervention strategy becomes possible as opposed to a generalised one-size-fits-all approach. The growing support for establishment of clinical-genetic specialist networks promises to increasingly change the health system from disease management to health management. Close collaboration between molecular geneticists and primary care providers will lead to the development of educational tools and practice guidelines required for the era of genomic healthcare.

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