If the majority of active X chromosomes in the brain are those with the mutation, intellectual disability is likely to occur.

A woman who carries a fragile X premutation could have inherited the mutation from her mother or her father. For men with a premutation all their daughters, but none of their sons, will be carriers.

in the next generation is a little lower, depending on the number of CGG repeats.

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Can premutation 'carriers' have symptoms?

A number of men, who have an increase in the CGG repeat number in the FMR1 gene in the premutation range, have developed a late-onset neurological disorder characterised by tremor, ataxia and cognitive decline. A small number of female premutation carriers have been described with similar symptoms but without dementia. Women who carry premutations are also at increased risk of developing premature ovarian failure.

How is fragile X syndrome diagnosed?

The clinical features of fragile X syndrome can be subtle, particularly in young children and in girls. The diagnosis should be considered in all individuals with intellectual disability or developmental delay, especially if there is a compatible family history.

Cytogenetic demonstration of a fragile site is diagnostic but less sensitive than molecular testing. Reliable DNA testing is available for diagnosis, carrier detection and prenatal diagnosis.

Genetic counselling in fragile X families

Diagnosing an individual with fragile X syndrome is only the beginning. The family should be evaluated and family members at risk identified for genetic counselling and testing where appropriate. Genetic counselling in this disorder is complex, but carries the hope of reducing the burden of this condition for families and communities.

MEDICAL INFORMATICS AND BIO-INFORMATICS: THE INFORMATION REVOLUTION AND MEDICINE

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As knowledge of the human genome brings us nearer to realising the dream of understanding the underlying causes of many disorders, ranging from straightforward genetic conditions such as cystic fibrosis (CF)[1] and Tay-Sachs disease (TSD),[2] through to the complex interaction of genes and environment that contribute to the individualised risk of heart disease or asthma, it is becoming increasingly important that we are aware of the resources making it easier to handle the overwhelming array of data available on the Internet.

Similarly, advances in medical informatics are set to change the practice of medicine at all levels. Advances such as personal databases for the compilation and analysis of patient records and related information, practice management software,[3] online libraries of drug interactions,[4,5] disease-specific support sites,[6-8] and continuing medical education,[9-10] to name but a few, completely refashion the toolset available to the GP in the practice of medicine.

The importance of information technology (IT) in medicine is nowhere more evident than in our medical schools. The University of Cape Town has made IT skills one of the cornerstone abilities of its new medical students. In fact, the first evaluation of new MB ChB students is an IT literacy evaluation on day one of their first year. The underlying principle is not only that it is important to know which resources are available now (such as the list of support sites for individuals afflicted with rare genetic conditions),[6-8] but also to equip the GP with the know-how to track down new resources as they become available. In this regard search engines like Google[11-13] and a range of others are becoming some of the most used electronic tools. One correctly placed query can save endless hours of wading through old journals, textbooks or antique lecture notes (heaven forbid).

As an example try to find the contact details of the South African Inherited Disorders Association'. Then try 'South African Inherited Disorders Association as a Google[11-13] search. (Note that entering SAIDA is not nearly as efficient, although the link is still on the first page of results.)

An important caveat needs to be placed on the use of Internet resources for clinically important information: remember that unless the information is from a proven (that means proven to your standards of proven, not anyone else's) source, the information available from the majority of sites should be regarded as having back-of-breakfast-box reliability. This means that it may sound right, and it may even be right, but it cannot be accepted as scientific fact simply because some person has decided to place it on a website. This does not mean that useful and factual information is not available; it only requires that you apply the same level of judgement that you

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would apply to anything a stranger might say. Reliable sites, such as Online Mendelian Inheritance in Man (OMIM[14]) that have been carefully curated tend to be strong on scientific fact and have a reliable academic pedigree, whereas personal websites dedicated to pet topics are more likely to be no more valuable than graffiti, and one would normally not regard graffiti as a good source of information — philosophical or medical.
The potential benefits of embracing the IT revolution in the GP practice are varied, but one of the best applications has to be practice management software, where one system can be used to replace patient record cards, the booking system, the medicines formulary, and the prescription pad (along with the associated risk of reading/writing errors in the dispensing of drugs). It will then be possible to record allergies, chronic medication and other relevant information to support medical decision making. With the promise of personalised medicine coming out of the genomic revolution, it will be increasingly important for practitioners to be able to use and interpret the new data, such as drug sensitivity profiles and disease risk factors, and to incorporate this information into their service to patients. A progressive practice management system should be able to do this and adapt to future, unforeseen needs.

The informatics explosion has changed the way biological research is performed. It is currently not advisable to undertake a research project in human molecular genetics without having first carried out a detailed bio-informatic analysis. Medical informatics is set to change GP practice in much the same way. It will be impractical to expect a GP to have a full working knowledge of all the possible drug interactions, contraindications, and adverse reactions. To simultaneously be able to process individual drug responses, treatment profiles, and allergies is a task no-one should attempt without adequate IT support.

We should be looking to our continuing medical education and professional development programmes to advance the skill levels needed to utilise these new tools of the art efficiently.

Universal Resource Locators (URLs) for Web-based links

1. **http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=219700**
2. **http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=272800**
3. **http://www.hipaa.org/pmsdirectory/**
4. **http://www.drugs.com/drug_interactions.html**
5. **http://www.rxlist.com/**
6. **http://www.kumc.edu/gec/support**
7. **http://www.rpsa.org.za**
8. **http://sunsite.wits.ac.za/saida/**
9. **http://www.cmelist.com/list.htm**
10. **http://www.medicalcomputingtoday.com/0listcme.html**
11. **http://www.google.com**
12. **http://www.ananzi.co.za**
13. **http://www.yahoo.com**
14. **http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM**

SINGLE SUTURE

DON'T OVERDO IT

Physicians are being warned that they are 'overperforming' surveillance colonoscopy in patients found to have hyperplastic polyps. Published guidelines say that repeat colonoscopy is not advised, but half of the gastroenterologists questioned recommended repeat colonoscopy every 3 years or more often. The report concludes that unjustified overuse of surveillance colonoscopy after polypectomy will deplete resources that could be better used elsewhere.

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