

intercourse and maturation. Advice is often sought on participation in exercise,¹³ sexual activity, contraception, employment, and insurance. In many cases too few data are available to allow informed decisions. In a recent survey we found that prospective employers and insurers were often extremely cautious about applicants with congenital heart disease because of a perceived risk of premature incapacity and death. In some cases this caution is appropriate, but in people without symptoms or residual abnormalities this approach is unnecessarily harsh. One of the major roles of a specialised cardiac service for young adults would be to define the functional capability and long term survival of patients with congenital heart disease.

To care adequately for adults with congenital heart defects a specialised service must be available for referral or consultation. Efforts are being made to determine the provision available in Europe and to organise care.¹⁴ The service must be able to deal with both the cardiac problems and the wide range of other medical problems that occur in these patients, to provide expert surgery, and to offer psychosocial support and advice. Concentrating on this group of patients will provide education and opportunities for research for a new specialist group of cardiologists, surgeons, and health professionals. Long term follow up data from such clinics would allow services to be planned and also enable paediatric cardiologists to modify early management to optimise later outcome. With a high level of medical care and psychosocial advice each adult

with congenital heart disease would have the best chance of achieving his or her full potential.

DAVID S CELERMAJER
Honorary Senior Registrar
JOHN E DEANFIELD
Consultant Cardiologist

Cardiothoracic Unit,
Hospital for Sick Children,
Great Ormond Street,
London WC1N 3JH

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Understanding Marfan's syndrome

Abraham Lincoln revisited

The retrospective diagnosis of rare and exotic diseases in celebrities from the past has provided a rich source of speculation for medical historians, who must have often lamented their inability to call on contemporary medical evidence. High on the list of putative diagnoses comes Marfan's syndrome: Abraham Lincoln, Paganini, and Rachmaninov may have had it.¹ Events of the past few months suggest that for Marfan's syndrome, and possibly other single gene disorders, the medical historian may soon be able to call on the molecular biologist as an expert witness.

Even the most inattentive medical student is usually familiar with Marfan's syndrome. Although relatively rare, with an estimated prevalence of 1 in 10 000, the full blown clinical picture of tall stature, long limbs, and arachnodactyly is well known. A firm diagnosis should be based on well defined diagnostic criteria² with involvement of at least two and preferably all three of the major systems affected—that is, the eye, heart, and skeleton. To these can be added a fourth diagnostic criterion, a positive family history—for it has long been recognised that Marfan's syndrome shows autosomal dominant inheritance with close to complete penetrance (that is, everyone with the gene manifests it to some degree) but very variable expression even among affected members of the same family.

Despite extensive research, progress on the genetic front was disappointingly sluggish during a decade that witnessed remarkable progress in the understanding of most common single gene disorders. Then quite suddenly the situation changed dramatically with the almost simultaneous announcements that fibrillin, a component of the microfibrillar system associated with elastin, was lacking in skin from patients with Marfan's syndrome³ and that the disease

coregulated with polymorphic DNA markers on chromosome 15q.⁴ The attractive unifying hypothesis that the gene for fibrillin would be located on chromosome 15q proved to be valid. Further support for fibrillin as the basic defect has come from two further studies. One has confirmed tight linkage of polymorphisms in the fibrillin gene with the disease in multiplex families⁵; the other has found an identical arginine to proline point mutation in two unrelated patients.⁶

In most genetic disorders the molecular story has proved more complex than expected, with allelic (different mutations at the same locus) and locus heterogeneity (mutations at different loci) proving the rule rather than the exception. Marfan's syndrome will probably be no different, a point amply illustrated by the finding of the original arginine to proline mutation in only two of over 40 patients.⁶ Locus heterogeneity seems less likely given the very recent demonstration of linkage to chromosome 15q in 17 families from five different populations drawn from Europe and the United States.⁷ Healthy scepticism, however, is in order until a much larger volume of data has been accumulated.

What, if any, are the practical benefits of these observations for patients and their doctors? In the short term these will be diagnostic rather than therapeutic, although the two cannot be totally separated. In a family with more than one affected member, in which linkage to chromosome 15q has been shown, reliable preclinical and, if acceptable, prenatal diagnosis will now be available. Inevitably this will raise difficult issues at a time when the ethical aspects of presymptomatic diagnosis in children⁸ and associated problems of insurance⁹ are under active discussion. Given the potential for cardiac morbidity and mortality in young adults with Marfan's syndrome,¹⁰ the case for early diagnosis will be

overwhelming—particularly if prophylaxis with drugs such as propranolol is shown to be beneficial. Those who have not inherited the Marfan's gene will be spared unnecessary anxiety and investigation, whereas gene carriers could be entered into carefully planned surveillance programmes offering at the very least regular cardiac and ophthalmological assessment.

For isolated cases, accounting for perhaps one quarter to one third of all patients with Marfan's syndrome, and patients from families too small for linkage studies, preclinical and prenatal diagnosis will have to await elucidation of their specific gene defect. If the identification of point mutations and subtle rearrangements in the fibrillin gene proves to be as difficult as pinpointing the individual collagen mutations in osteogenesis imperfecta then specific molecular diagnosis is likely to be a long way off. This is particularly unfortunate given the existing problems in diagnosing apparently mild cases of Marfan's syndrome and allied conditions such as contractural arachnodactyly, which may be due to mutations in a fibrillin gene on chromosome 5⁵; the familial mitral valve prolapse syndrome; and other less certain entities such as the Marfanoid hypermobility syndrome.

Ultimately the discovery of the underlying basic genetic and protein defect must raise hope for the development of more effective therapeutic strategies. This alone would seem

to justify the excitement generated by recent events. If attempts to identify a mutation in blood stains, bone, and hair from Abraham Lincoln are successful¹¹ then the medical historians can justly take pride in their diagnostic acumen. Meanwhile patients with Marfan's syndrome can console themselves with the notion that they are in distinguished company.

Consultant Clinical Geneticist,
City Hospital,
Nottingham NG1 6HA

IAN YOUNG

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Doctors in the making

How to resolve problems in the preregistration year

The publication of *Doctors in the Making*¹ is an important contribution to what is becoming an impressive body of published work on the problems of junior doctors. Having conducted a series of structured interviews with house officers, consultants, nurses, and managers, the authors present their results and base a comprehensive list of recommendations on them.

The interviews confirm previous research. House officers become disillusioned,² are stressed and unsupported,³ and receive little formal training.⁴ They feel undervalued by the hospital, and their perception is correct. Managers and consultants avoid or deny the implications of this and the punitive conditions in which house officers work.⁵ A worrying finding from this report is that consultants seem to be largely ignorant of the recommendations of the General Medical Council on the training and supervision of house officers.⁶

Possibly the most refreshing aspect of *Doctors in the Making* is that it appreciates there is a complex relationship within the organisation that is dynamic and involves several professional groups. It then uses this relationship to formulate a series of linked, multidimensional proposals aimed at the resolution of house officers' problems. The past is littered with well intentioned initiatives aimed at one component rather than at the problems as a whole. Hours of work, education, and support are all important topics, but the key to improving the lot of house officers lies in enhancing their job satisfaction by a combination of diminishing their workload, improving the appropriateness of their work and how much control they have over it, recognising their contribution and giving them feedback on their performance, enabling their active participation in the management of their work, and giving them an educational experience that they will perceive as valuable.

None of this need be complex or expensive or require

centralised direction. A striking feature is how the perceptions of house officers changed in response to simple interventions, such as consultants appearing interested in their life outside work.

The report contains a new analysis of the role of house officers. Instead of the traditional model, which makes all aspects of a preregistration house officer's work the responsibility of consultants, different lines of accountability are suggested for different parts of the work. For some parts house officers would be accountable to nurses and managers—a long overdue acknowledgment of a reality that should be taken on board by doctors, rather than responded to defensively.

The important and informal contribution made to the education of house officers by senior nursing staff is recognised, and the report suggests that it should be formalised. The authors acknowledge that in doing this there could be difficulties in definitions and responsibilities. Given the undoubted overlap in care between house officers and nurses, it is surprising that the concept of partnership in the provision of care, at least at ward level, is avoided. If there is a genuine mutuality between the nursing and medical professions it must be logical to integrate them rather than preserve the traditional model. The relationship between house officer and ward sister must be a sensible place to start.

The main difficulty with a multidimensional approach will be how to drive it. Professional defences can be overcome, and finding financial and human resources is often an overrated problem. Essentially the authors advocate changing the culture according to the tenets of good personnel management. National bodies, such as the General Medical Council and the Junior Doctors' Committee, have a part to play, mainly in enabling change. The real power and possibilities,