

Training

Academic paediatrics

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Crisis or opportunity?

There has been a perception for some time that “academic paediatrics” is in a state of crisis.¹ University departments of paediatrics (child health, child life, etc) have been disappearing, some fusing with other departments and others absorbed into divisions, schools, or larger entities. Loss of clinical lecturers has been one consequence, as universities make high quality research their priority. The warning is raised that if academic departments vanish, the future leaders of paediatrics will be lost.² What is happening to academic paediatrics is not unique, but paediatricians have been surprised and distressed by it. Just when our new Royal College unites the specialty and gives it a stronger identity, we find the universities trying to do away with our academic departments. This has resulted in anguish, wringing of hands, and even shroud waving.

There are a number of reasons for this “crisis”. The demands of the research assessment exercise (RAE), the physical dislocation of “science” departments from clinical sites, and the drive by universities to concentrate on income generation through research to the neglect of the health problems of children, and undergraduate teaching, have impacted on academic staffing.² The pressures generated by shorter specialist training, consequent on “Calmanisation”, European training, and working hours directives, discourage young doctors from pursuing an academic career. The “new” student centred, problem based undergraduate curricula fail to equip medical students for a career in research. The lack of systematic teaching in the basic sciences is in part rectified by intercalated BSc courses, but most medical graduates have a weak grounding in the sciences that underpin modern clinical medicine. This has to be acquired post-graduation, and few young doctors experience research during their early clinical training.

BIOLOGY AND PATHOLOGY OF EARLY LIFE

Paediatricians have a proud and productive history of collaboration with non-clinical scientists, such as in neonatology, neuroscience, and nutrition. In vaccine

development and imaging, for example, they have been at the forefront, sharing in prestigious prizes and FRSS. The big questions in child health may have changed, but are clear enough: the early origins of health and disease, the genetic and molecular basis of conditions manifest first in infancy and childhood, and the aetiology, prevention, and treatment of specific childhood diseases.³ Modern medicine is increasingly applied biology and technology, and in a global research environment there are rich opportunities for fruitful collaborations, and plenty of funding. We need clinical scientists that are trained in appropriate scientific methods and understand the nature of childhood development and disease. They must be comfortable at bench and bedside.

There has long been a tension between the demands of “doing science” and “practising medicine” within those who choose to combine them.⁴ In the past it may have been possible for young paediatricians to learn to be clinical investigators or even laboratory based scientists, after their medical training. The old fashioned model of discrete departments, single handed clinical investigators, working alone or with little support in a clinical environment, has been replaced by interdisciplinary teams of researchers (some clinical, some non-clinical) using modern techniques, resourced to address big scientific questions effectively and efficiently, sometimes remote from a clinical base. Small departments in many universities have been slow and poorly equipped to react. Collaboration and critical mass is the name of the game. The changes that have come about over the last decade are not a process of disintegration, but of mould breaking. A new form of training that is more systematic and purposeful is demanded now, dictated by the nature of modern scientific methodology. The Academy of Medical Sciences has been a voice of the Royal Colleges and universities in debating these issues.⁵ The recent report “Modernising Medical Careers”⁶ now articulates them further, with stronger focus on the NHS and its role. A number of proposals are made, including integrated clinical pathways for training medical academics.

WHAT SHOULD WE DO?

We cannot and should not train future academic paediatricians exclusively in departments of paediatrics, unless they have a close and mutually supportive relationship with non-clinical scientific departments or research groups. The new academic training pathways offer the means, during the foundation years and in specialist training.⁶ We must establish clinician scientist trainees in multidisciplinary research groups within or allied to university researchers (for example, molecular, epidemiology, etc). This requires “letting go” of them, “embedding” them in research groups where they will acquire a scientific training in methods relevant to clinical questions, and then “bringing them back” into paediatrics. The onus is on academic paediatricians to establish cognate research groups within the portfolios of university research themes/divisions, which in large part are defined by their potential to succeed in the RAE. The onus is on the RCPCH and new postgraduate medical education training board (PMETB) to develop flexible competence based systems of assessment that accommodate the dual training of clinician scientists.

The dominance of molecular biology and the “new genetics” to medical research is waning as the challenge of understanding their relevance and implications, and applying novel findings to physiological and metabolic processes gets underway. The tide is turning in favour of “clinical”, whole body research, and intervention trials. This is being facilitated by the recognition of the potential of the health service to conduct large clinical trials, and convergence of NHS R&D agendas with those of university based clinical researchers. The testing of children’s medicines and vaccine development are welcome examples of new collaborative initiatives combining complementary expertise within clinical research facilities. Such partnerships between the universities and health service may also rationalise responsibilities and resources for undergraduate medical teaching.

SEIZE THE OPPORTUNITIES

Paediatrics is securely established as an independent medical specialty, with its own royal college. Preoccupation with the pressing issues of training, continuing professional development, revalidation, and governance should not lead to introspection. Paediatricians must not regard their calling as too holy or pure a vocation and forget that child health is the basis of adult health. They have not led the way in re-establishing this vital connection.³ To advance world class research into subjects that will benefit

both children and adults, they should get into bed with geneticists, epidemiologists, fetal medicine specialists, and “basic scientists” in molecular biology, immunology, neuroscience, pharmacology, medical physics, and nutrition, among other subjects. They can afford to relax their defence of the independent, special status of paediatrics, and embrace their colleagues in these related specialties in sharing the common goal of researching major questions that determine early health and affect the natural history of disease throughout life.

The “crisis in academic paediatrics” may be perceived as a threat to departments of child health. But it is not a threat to child health itself. Crossing the boundaries that surround paediatrics and separate it from other allied specialties, and forging alliances with them in the common interest in research into the biology, pathology, and public health of early life must be the future.

The opportunities for funding are there; new models for the training of aspiring clinical researchers, and for postdoctoral support are emerging.⁷ European and global research networks are easily established, through email and cheap travel. Facilitated by the EU, specialist medical societies, and industry, the “added value” of sharing expertise, pooling patients, and interchanging trainees can be exploited. In this respect Europe may be ahead of North America. The old mould is broken. The new structures will reinvigorate paediatrics through cross fertilisation and put it at the heart of research into the early genesis of adult ill health, the developmental basis of rare congenital disease, and many common health problems.

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REFERENCES

- 1 Anon. UK paediatric clinical research under threat. *Arch Dis Child* 1997;**76**:1–8.
- 2 Levene M, Olver R. A survey of clinical academic staffing in paediatrics and child health in the UK. *Arch Dis Child* 2005;**90**:450–3.
- 3 Weaver LT. The child is father of the man; paediatricians be more interested in adult disease. *Clin Med* 2001;**1**:38–43.
- 4 Weaver LT. A good doctor and/or a good scientist? The medical academic’s mid-life crisis. *J R Soc Med* 1993;**27**:444–6.
- 5 Academy of Medical Sciences. www.academicsci.ac.uk.
- 6 UK Clinical Research Collaboration. *Modernising medical careers*. NHS. 2005.
- 7 Savill J. Academic paediatrics: Easter Island or Easter Sunday? *Arch Dis Child* 2005;**90**:441.

Genetics

A monogenetic disorder yet multiple and varied clinical manifestations

L I Landau

Commentary on the paper by Callaghan *et al* (see page 1029)

It was thought that identification of the cystic fibrosis gene—the cystic fibrosis transmembrane conductance regulator—15 years ago would lead to the solution for many of the serious consequences of this most common inherited fatal disorder. Instead, over 1000 mutations of this single gene have been reported with varied disease manifestations for each of these different mutations for cystic fibrosis. Substantial variations in the disease within the same CFTR genotype have been found. The impact of CFTR on other conditions such as infertility, diarrhoeal diseases (cholera), and asthma, has been described. This highlights the need for further investigation to better understand the mechanisms for the varied phenotypic expression of these numerous polymorphisms of CFTR.

In this issue, Callaghan and colleagues¹ report on growth and lung function in Asian patients in the United Kingdom with cystic fibrosis. They found that Asian girls had lower FEV₁

and FVC, but isolation of *Pseudomonas aeruginosa* at a later age (on average 3 years later) than a matched control group. These findings are different to those reported by Bowler *et al* from Leeds,² who found that the Asian patients had lower respiratory function results, lower BMI (body mass index), and isolated *P aeruginosa* on average 3 years earlier than the control group.

These observations raise interesting issues that are essential to our understanding of the impact of the environment on phenotypic expression of most genetic disorders. Asians have a low incidence of cystic fibrosis and lower prevalence of the ΔF508 mutation.

Kabra and colleagues³ reported 17% prevalence of ΔF508 in cystic fibrosis children in India and Pakistan compared with over 70% in most Caucasian cystic fibrosis cohorts. Although diagnosis was delayed in these patients, the clinical presentation was otherwise described as classical. Wang and colleagues⁴ found no ΔF508 mutations in 100

Japanese children with cystic fibrosis. Just as the pattern of mutations varies throughout Caucasian societies, especially as one moves from the Mediterranean to Northern Europe, similar variations appear to be present in different Asian societies.

Wu and colleagues⁵ found that the frequency of CFTR mutant alleles in Taiwanese men with congenital bilateral absence of the vas deferens (CBAVD) was 36%, lower than published frequencies in other ethnic CBAVD patients (ranging from 50% to 74%). As well the mutation spectrum of CFTR in CBAVD patients did not overlap with the Caucasian CFTR mutation spectrum in this condition.

The environmental impact on phenotypic expression can relate to social factors such as recognition affecting the age of diagnosis, access to medical care, compliance with recommended care, and relative social disadvantage, especially in migrant communities.

Gene expression can be influenced more directly by epigenetic factors such as diet and toxins or by epigenetic inheritance of modifier genes co-inherited with the candidate gene. Promoter sequences elsewhere in the genes, independent of CFTR, may exert considerable influence on the outcomes of CF. However, a definitive modifier gene for CF remains to be identified.⁶ The expression of particular alleles in other conditions may also be influenced by whether the particular allele was inherited from the father or the mother and this may even be applicable to CF.⁷