Editorial

Journal of Medical Genetics 1988, **25,** 433

Dysmorphology is the clinical activity concerned with the diagnosis and study of congenital anomalies. The diagnosis of multiple congenital anomaly syndromes has always been a specialised activity with some practitioners seemingly able to store large amounts of information about exceedingly rare conditions, and to identify instantly a characteristic facial appearance. This sometimes leads to the accusation of 'stamp collecting', or of making arbitrary subjective diagnoses. However, as this issue of the *Journal of Medical Genetics* shows, dysmorphology has much greater depth and breadth.

At a clinical level a correct diagnosis is essential for proper management, including prevention of complications, and for assessment of prognosis and genetic risks. The study of the natural history of multiple congenital anomaly syndromes is important both for the provision of information to clinicians and families and for the understanding of the underlying causes of the disorder.

Dysmorphology is now moving out of the purely 'descriptive' phase into an era where the molecular, cytogenetic, biochemical, and embryological mechanisms involved in the causation of congenital anomalies can be explored and elucidated. As many more dysmorphic syndromes are delineated, the process of diagnosis must be examined and explained. Several groups are using computers to classify and diagnose children with multiple congenital anomaly syndromes. These approaches range from attempts at numerical taxonomy to computer assisted diagnostic systems. Any successful, that is, clinically useful, computerised system for diagnosis will involve steps similar to those taken by an experienced clinician, so that analysis of the design and development of these systems should tell us something about the diagnostic process itself. The organisation of diagnostic categories within these computerised systems will also help to improve the classification of birth defects.

A request for lymphocyte chromosome analysis is an automatic reaction for any clinician faced with the investigation of a dysmorphic child. However, there are now many recognised chromosome microdeletion and mosaic syndromes and a more sophisticated assessment of which cytogenetic investigations are appropriate is required. The diagnosis of these conditions may require detailed prometaphase chromosome studies, with suggestions from an experienced clinician to the laboratory as to where to look, as well as sampling of tissues other than blood lymphocytes.

The clinical phenotypes associated with many lysosomal storage disorders and aminoacidopathies are well known. The dysmorphologist must now also consider further groups of metabolic disorders, such as organic acidurias and peroxisomal disorders in the investigation of the dysmorphic child. There can no longer be an artificial distinction between inborn errors of metabolism and the multiple congenital anomaly syndromes and a return to the biochemistry text books should be encouraged.

Ultimately, all developmental defects must be explained at the molecular level. Molecular biologists are now focusing their attention on aspects of developmental biology. They need suitable models to study and initially these will include rare human single gene malformation syndromes and appropriate animal models. The dysmorphologist must be an integral part of this investigative team, providing both clinical information and samples and insight into animal models.

Dysmorphology has become an important sub-speciality of clinical genetics and is an essential part of the training of clinical geneticists. In addition to their clinical knowledge, dysmorphologists should be acquainted with comparative studies in laboratory animals, computerised classification, and the rapidly advancing techniques of cytogenetics, biochemistry, and molecular biology. The current edition of the Journal contains reviews and papers which emphasise this joint clinical and scientific approach and point the way ahead.

ROBIN WINTER AND DIAN DONNAI

Separate copies of this issue of the journal may be obtained from the publisher on request.