flat nasal bridge with a small snub nose, but the overall pattern of the face does not seem to fit Robinow syndrome as I have seen it in a total of seven cases. First, the frontal bossing is not impressive and actually looks more like a metopic ridge. Infants with Robinow syndrome usually have pronounced frontal bossing and more obvious macrocephaly than case 1 of Sabry et al. Her head also may seem large as a result of the obvious dystrophic condition she is in. Second, the lower face is too fine and the chin too pointed for Robinow syndrome. The mouth does not look like the typical thin lipped “carp mouth”. The overall facial structure is too finely sculptured for me. The face in Robinow syndrome tends to be rather coarse, Greig hypertelorism-like, in contrast, with a rounded lower half. The coarseness can be so pronounced that sometimes metabolic investigations are initiated because a mucopolysaccharidosis is suspected (own observation).

Third, eyelid hypoplasia giving the impression of exophthalmos seems to be quite a common feature. I do not see it in this patient.

Fourth, the mesomelia in case 1 is certainly not impressive. It is a highly variable feature and actually not of much use in the diagnosis (see, for instance, Bain et al.), but if present it is a strong argument for the diagnosis, so some measurements of bone length would have been helpful here.

Finally, stating that the labia minora and clitoris were “slightly hypoplastic” seems a bit vague. What is “slightly”? A photograph would have been helpful.

The photograph of subject 2 in fig 7 poses some difficulties. Though it is true that the facial abnormalities tend to become somewhat less obvious with age, some anomalies remain quite obvious: the snub nose, the hypertelorism, and the thick alveolar ridges. The face also remains rather square and coarse. Subject 2 has a large nose compared to some of my patients of the same age and his hypertelorism is rather modest. His face seems too fine, much like his sister’s. His alveolar ridges can, of course, not be judged from the picture. In my opinion, a diagnosis of Robinow syndrome is not certain in his case either.

As far as case 2 is concerned, her photograph (fig 8) is more convincing. Particularly when comparing the lower half of her face with case 1, it will be possible that there is a clear difference between the two. In my opinion the face of case 2 is definitely more “Greig-like”. Though there is no mesomelia in this patient, this feature can be variable, as stated. I feel that in case 2, the diagnosis of Robinow syndrome is probably correct. This patient is not related to the other patients and, considering this, I think it is possible that the authors have in fact encountered, in case 1 and subject 2, a new, previously unreported malformation syndrome with some resemblance to Robinow syndrome.

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This letter was sent to Dr Sabry et al, who reply as follows.

We read the comments of Dr van Steensel concerning our report of unusual traits associated with Robinow syndrome, who showed other unusual traits in addition. We also observed variable expression of some of the traits of Robinow syndrome in healthy sibs/cousins of the proband in this consanguineous family. Like many syndromes for which no molecular/ biochemical/cytogenetic markers have been identified, the Robinow syndrome remains solely dependent on the clinical phenotype of the patients. Naturally, this gives wide scope for different subjective views to argue for or against a given diagnosis. This is particularly true for Robinow syndrome with its wide spectrum of inter/intrafamilial phenotypic heterogeneity that would be expected to reflect a corresponding degree of molecular variability. Of course, the profile in the proband of family 1 does not show a straightforward Robinow phenotype, or it would have been of little interest to the genetics community. Although we bear in mind the possibility of a new Robinow-like malformation syndrome in family 1 of the report, we are reluctant to designate it a new syndrome until all available possibilities are exhausted. Incidentally, we have recently received a letter from Dr H G Brummer from the Department of Genetics, University Hospital, Nijmegen, expressing interest in our Robinow syndrome cases and requesting our collaboration in their ongoing molecular study to map and clone the gene(s) responsible, which we are now considering.

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BOOK REVIEW

If you wish to order or require further information regarding the titles reviewed here, please write to or telephone the BMJ Bookshop, PO Box 295, London WC1H 9JX. Tel 0171 383 6606. Fax 0171 383 6602. Books are supplied post free in the UK and for BFPO addresses. Overseas customers should add 15% for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name. (The price and availability are occasionally subject to revision by the Publishers.)


This book reviews the genetic changes observed in solid tumours with particular emphasis on the practical issues of diagnosis, prognosis, and monitoring therapy. The book provides comprehensive coverage of the impact of the new genetic technology in furthering the understanding of the mechanisms underlying tumour development. The opening chapter illustrates the increasing relevance of genetic markers in tumour diagnosis and prognosis and provides an excellent introduction to the subject. Part 1 of the book, consisting of several chapters, covers the application of the major techniques, including flow cytometry, in situ hybridisation, CGH, and nucleic acid amplification. The relevance and applications of these techniques are well described. Part 2 of the book comprises comprehensive reviews of the current knowledge of the cytogenetic and molecular genetic changes, this book provides specific tumour types/subtypes. Each of these chapters is contributed by acknowledged experts in the field.

There is, almost inevitably, some variability in the apparent quality of the reviews and advances in this field are taking place continually a book of this nature is always going to be a little behind hand. With the exception of an excellent chapter on the morphological, antibody, and chromosomal classification of haematological malignancies, this book does not cover leukaemias and lymphomas. One chapter at the very end of the book is disappointing. This necessitates frequent page turning, as the colour is fundamental to the illustration in some instances! However, this book provides excellent background information and an overview from which it would be possible to delve deeper using the cited references, although a quick scan for the new publications would also be wise in some instances.

A certain level of knowledge of solid tumours, cytogenetics, and molecular biology is assumed. This should be a useful book for the interested pathologist and clinician as well as students in these areas. Those in research will find the book an easy introduction to a topic and useful in the process of formulating ideas and methodological approaches before embarking on conducting their own investigations. Selected areas of cancer cytogenetics represented in this book are also areas of expanding interest for cytogeneticists and genetic technologists who, working in league with pathologists, may ultimately be able to provide more information of practical use for patients.

LIONEL WILATT
JANET SHIPLEY

NOTICE

Call for patients with familial pancreatic disease: the EUROPAC Register

We are establishing a European register (EUROPAC) of families with hereditary pancreatitis, familial pancreatic cancer, and familial atypical pancreatic cancer as part of a familial cancer syndrome. This collabo-