

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 seems too finely sculpted to 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 constant 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 an extra 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 that of case 1, it will be seen 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 this case 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 recessive malformation syndrome with some resemblance to Robinow syndrome.

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- 1 Sabry MA, Ismail EAR, Al-Naggar RL, *et al.* Unusual traits associated with Robinow syndrome. *J Med Genet* 1997;34:736-40.
- 2 Bain MD, Winter EM, Burn J. Robinow syndrome without mesomelic "brachymelia": a report of five cases. *J Med Genet* 1986;23:350-4.

This letter was shown 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. In family 1 of our report, we described a female patient with many of the constant traits of 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 diagnosis of 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 Brunner 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

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Human Cytogenetic Cancer Markers. Editors Sandra R Wolman, Stewart Sell. (\$125.00.) New Jersey, USA: Humana Press. 1997. ISBN 0-896-03357-0.

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 fur-

thering 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 a good 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 observed in organ 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 as 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 describes special techniques in cytogenetics, with emphasis on microdissection, which would perhaps have been better placed earlier in the volume along with the other methodologies. Although the colour plates are replicated as black and white photographs within the chapters, their placement within the centre 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 WILLATT

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 where pancreatic cancer has occurred as part of a familial cancer syndrome. This collabo-