

anomalies, lower thoracic vertebral scoliosis, two sacral segments (partial sacral agenesis), abnormal pelvis with abnormal right ischium and pubis and overlapping of pubic bones, and questionable dislocated left hip. There was no bone in the presacral appendage. Both parents had normal skeletal surveys.

In addition to this new patient, a patient reported previously as having VATER association³ may represent another example of a disorganisation-like gene. The 31 week old male had multiple vertebral anomalies, imperforate anus, distal tracheo-oesophageal atresia with partial proximal oesophageal atresia, bilateral renal dysplasia, single umbilical artery, bilateral cleft lip and palate, and dysplastic ears. Although VATER 'association'⁴ might describe this patient (and would also characterise our patient), the presence of additional unusual anomalies (agenesis of the bladder, urethra, and penis with rudimentary scrotum, sacral caudal skin appendage, and right sided 'lobster claw' foot) suggests that the diagnosis of disorganisation homologue is more accurate.

I agree with Winter and Donnai¹ that patients with "extra limbs, appendages, or hamartomatous structures, in association with polydactyly or partial duplication/reduction of limbs and apparently distinct malformations, such as urogenital, body wall, and craniofacial abnormalities" may be the result of a disorganisation-like gene, especially when such patients are atypical examples of their diagnoses. Although this new patient lacks duplicated digits or limbs, several malformations (phallus-like sacral structure, rudimentary perineum, left sided foot-like appendage, absent right sided radius and thumb, ectopic renal, adrenal, and thymic tissues) resemble patients with the disorganisation-like complex.

Although the partial sacral agenesis, absent kidneys, abnormal testes, imperforate anus, and shortened lower segment and lower extremities could be attributed to the caudal dysplasia sequence (caudal regression syndrome),⁵ the type and extent of non-caudal anomalies suggest a more widespread condition. These entities are not mutually exclusive. If there is indeed a single gene disorder in humans resembling the mouse mutant disorganisation, then the caudal dys-

plasia and Potter oligohydramnios sequences may occur as part of that disorganisation-like syndrome.⁶ Similarly, VATER association, which describes many features of our patient and that of Dusmet *et al.*,³ may also be found within the broader context of that syndrome.

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- 1 Winter RM, Donnai D. A possible human homologue for the mouse mutant disorganisation. *J Med Genet* 1989;26:417-20.
- 2 Donnai D, Winter RM. Disorganisation: a model for 'early amnion rupture'? *J Med Genet* 1989;26:421-5.
- 3 Dusmet M, Fete F, Crusi A, Cox JN. VATER association: report of a case with three unreported malformations. *J Med Genet* 1988;25:57-60.
- 4 Weaver DD, Mapstone CL, Yu PL. The VATER association. Analysis of 46 patients. *Am J Dis Child* 1986;140:225-9.
- 5 Jones KL. *Smith's recognizable patterns of human malformation*. Philadelphia: Saunders, 1988:575.
- 6 Spranger J, Benirschke K, Hall JG, *et al.* Errors of morphogenesis: concepts and terms. *J Pediatr* 1982;100:160-5.

BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add £2 per item 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 your full name.

Genomic Imprinting. Ed M Monk, A Surani. (Pp 155; £60.00.) Cambridge: The Company of Biologists. 1990.

Genomic imprinting is attracting increasing attention as a possible explanation for some of the unusual or non-classical inheritance patterns seen in human genetic disease. There-

fore this collection of papers from an international symposium on genomic imprinting held in Manchester in April 1990 is both topical and potentially of great interest to many clinicians and scientists working in medical genetics. The range of topics covered is extremely wide including aspects of genomic imprinting in plants, yeast, insects, and mammals. Only two of the 18 papers are directly concerned with clinical genetics: Angus Clarke (Cardiff) and Judith Hall (Vancouver) each discuss the relevance of genomic imprinting to human genetic disease. Although there is some overlap between papers, together both papers document the clinical disorders in which there is strong evidence for parental genome effects, and also speculate on those disorders which appear to be candidates for imprinting but for which so far there is little evidence. The recent experimental evidence in support of the Laird hypothesis for fragile X is encouraging, but in many human genetic diseases the molecular evidence for imprinting is less than the number of hypotheses and models proposed. Such models are based on extrapolations from processes studied in invertebrates and small mammals, such as position effect variegation in *Drosophila*, and transgenes, gene methylation, and imprinted regions in the mouse genome. All of these (and other relevant) topics are the subject of individual contributions so that this volume represents a convenient starting point from which to explore many of the diverse genetic phenomena which are encompassed in the term 'genomic imprinting'.

Inevitably, as with all symposia proceedings, this volume does not provide a completely comprehensive and coordinated account of the subject. Nevertheless, the distinguished contributors cover a wide area to a high standard. I found this collection of papers informative and provocative and would recommend it to the many clinicians and scientists with an interest in this rapidly advancing field.

E R MAHER

Chromosome Banding. A T Sumner. (Pp 434; £60.00.) Glasgow: Harper Collins. 1990.

Chromosome banding refers to the patterns of bands which may be