

- Kitamura, K. and Hirako, T. (1955). Über zwei japanische Fälle einer eigenartigen retikulären Pigmentierung. *Dermatologica*, **110**, 97-107.
- Koszewski, B. J. and Hubbard, T. F. (1956). Congenital anemia in hereditary ectodermal dysplasia. *Archives of Dermatology*, **74**, 159-166.
- Kubicz, J. (1970). Syndroma Zinsser-Engman-Cole cum dyschromia extensiva corporis. *Przegląd Dermatologiczny*, **57**, 239-242.
- Lobo, J. (1964). Doença de Zinsser et Engman-Cole. *Anais Brasileiros de Dermatologia*, **39**, (3), 33.
- McDonald, R. and Goldschmidt, B. (1960). Pancytopenia with congenital defects (Fanconi's anaemia). *Archives of Disease in Childhood*, **35**, 367-372.
- McGregor, D. D. (1968). Bone marrow origin of immunologically competent lymphocytes in the rats. *Journal of Experimental Medicine*, **127**, 953-966.
- Merigan, T. C. and Stevens, D. A. (1971). Viral infections in man associated with acquired immunological deficiency states. *Federation Proceedings*, **30**, 1858-1864.
- Milgrom, H., Stoll, H. L., Jr., and Crissey, J. T. (1964). Dyskeratosis congenita: a case with new features. *Archives of Dermatology*, **89**, 345-349.
- Moon-Adams, D. and Slatkin, M. H. (1955). Familial pigmentation with dystrophy of the nails. *Archives of Dermatology*, **71**, 591-598.
- Morrison, J. G. L. (1974). Dyskeratosis congenita: two extremes. *South African Medical Journal*, **48**, 223-225.
- Nazzaro, P., Argentieri, R., Bassetti, F., Leonetti, F., Topi, G., and Valenzano, L. (1972). Dyskeratose congénitale de Zinsser-Cole-Engmann. *Bulletin de la Société Française de Dermatologie et de Syphiligraphie*, **79**, 242-244.
- Orfanos, C. and Gafmann, H. (1966). Leukoplakien, Pigmentverschiebungen und Nageldystrophie: Zinsser-Cole-Engman-Syndrom-sog. Dyskeratosis congenita. *Medizinische Welt*, **2**, 2589-2594.
- Ortega, J. A., Swanson, V. L., Landing, B. H., and Mammond, G. D. (1972). Congenital dyskeratosis. Zinsser-Engman-Cole syndrome with thymic dysplasia and aplastic anemia. *American Journal of Diseases of Children*, **124**, 701-704.
- Pastinszky, I., Vánkos, J., and Rác, I. (1957). Ein Beitrag zur Pathologie der 'Dyskeratosis congenita' Cole-Rauschkolb-Toomey. *Dermatologische Wochenschrift*, **135**, 587-593.
- Peterson, R. D. A., Kelly, W. D., and Good, R. A. (1964). Ataxia-telangiectasia. Its association with a defective thymus, immunological-deficiency disease, and malignancy. *Lancet*, **1**, 1189-1193.
- Reich, H. (1973). Zinsser-Cole-Engman Syndrom. *Medizinische Klinik*, **68**, 283-292.
- Robbins, J. H., Kraemer, K. H., Lutzner, M. A., Festoff, B. W., and Coon, H. G. (1974). Xeroderma pigmentosum. An inherited disease with sun sensitivity, multiple cutaneous neoplasms, and abnormal DNA repair. *Annals of Internal Medicine*, **80**, 221-248.
- Rollier, M. R., Rollier, M., and Prost, A. (1966). Sur un cas de syndrome de Zinsser-Engman-Cole. *Bulletin de la Société Française de Dermatologie et de Syphiligraphie*, **73**, 383-386.
- Sandberg, A. A. and Hossfeld, D. K. (1970). Chromosomal abnormalities in human neoplasia. *Annual Review of Medicine*, **21**, 379.
- Sato, S. I. and Hannibal, J. E., Jr. (1962). Ectodermal defect: dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris. *Archives of Dermatology*, **86**, 114-115.
- Schamberg, I. L. (1960). Dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris. *Archives of Dermatology*, **81**, 266.
- Scoggins, R. B., Prescott, K. J., Asher, G. H., Blaylock, W. K., and Bright, R. W. (1971). Dyskeratosis congenita with Fanconi-type anemia: investigations of immunologic and other defects. *Clinical Research*, **19**, 409.
- Silva, J. R. E. (1966). Syndrome de Zinsser-Fanconi. *Annales de Dermatologie et Syphiligraphie*, **93**, 497-502.
- Sorrow, J. M., Jr. and Hitch, J. M. (1963). Dyskeratosis congenita: first report of its occurrence in a female and a review of the literature. *Archives of Dermatology*, **88**, 340-347.
- Steier, W., Van Voolen, G. A., and Selmanowitz, V. J. (1972). Dyskeratosis congenita: relationship to Fanconi's anemia. *Blood*, **39**, 510-521.
- Swift, M., Zimmerman, D., and McDonough, E. R. (1971). Squamous cell carcinomas in Fanconi's anemia. *Journal of the American Medical Association*, **216**, 325-326.
- Waldmann, T. A., Strober, W., and Blasse, R. M. (1972). Immunodeficiency disease and malignancy. Various immunologic deficiencies of man and the role of immune processes in the control of malignant disease. *Annals of Internal Medicine*, **77**, 605-628.
- Walzer, P. D., Perl, D. P., Krogstad, D. J., Rawson, P. G., and Schultz, M. G. (1974). *Pneumocystis carinii* pneumonia in the United States. Epidemiologic, diagnostic, and clinical features. *Annals of Internal Medicine*, **80**, 83-93.
- Wise, F. (1943). Dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris. *Archives of Dermatology and Syphilology*, **48**, 560.
- Zinsser, F. (1910). Atrophia cutis reticularis cum pigmentatione, dystrophia unguis et leukoplakia oris. *Ikonographia Dermatologica (Hyoto)*, **5**, 219-223.

## Notice

### Symposium on the Pathology of Pregnancy

The Royal College of Pathologists are holding a symposium on the pathology of pregnancy on 12 and 13 February 1976, at the Royal College of Physicians. The following subjects will be covered in the two-day meeting. Fetoplacental function: its nature and assessment; haematological problems; hypertension and renal disease; infections; trophoblastic tumours; and congenital abnormalities.

The symposium is open to workers in all disciplines connected with the subject. The registration fee is £12 (sterling), which includes coffee, lunch, and tea on the two days and a copy of the papers to be published as a special supplement to the *Journal of Clinical Pathology*. Those wishing to attend the symposium dinner on Thursday evening, 12 February, should send with their application form the additional fee of £8 (sterling) which includes drinks before and with the dinner. The final programme will be sent out in February 1976. Application forms can be obtained from The Royal College of Pathologists, 2 Carlton House Terrace, London SW1Y 5AF.

of non-chromosomal genetic topics: cleft lip and palate; the present status of treatment of the mucopolysaccharidoses; new studies on mucopolipidosis III; Menkes syndrome; the prune belly syndrome; familial cardiac lipidoses.

While an individual volume of these Boston conferences is perhaps not worth buying, the series may provide a useful continuous summary of advances in clinical genetics.

C. O. CARTER

**Progress in Medical Genetics, Volume X.** Edited by Arthur G. Steinberg and Alexander G. Bearn. (Pp. 269; illustrated. £10.80.) New York: Grune and Stratton. 1974.

Volume X in the series is an excellent indicator of the continuing expansion of medical genetics and its increasing practical importance. There are seven chapters whose subjects include the mutation rate in man (John Edwards), biochemical polymorphisms in animals (M. J. Siciliano, D. A. Wright, and Charles R. Shaw), inborn errors of the thyroid (John B. Stanbury), mucopolysaccharidoses (Elizabeth F. Neufeld), control of Tay Sachs' disease (M. M. Kaback, R. S. Zeiger, L. W. Reynolds and Marguerite Sonneborn), the XYY conundrum (D. S. Borgaonkar and S. A. Shah), and finally the evergreen topic of ethical issues in genetics (Robert M. Veatch). It is perhaps invidious to select from among

consistently good articles, and one's own interests naturally colour one's selection. The chapter by Siciliano, Wright, and Shaw describing biochemical polymorphisms in animals is particularly fascinating. Having kept tropical fish for a number of years and observed at considerable expense their frequent mortality, I was greatly enlightened by the account of a single gene model for melanoma occurring in F1 hybrids of certain swordtails and platyfish. The descriptions of biochemical markers in these chromosomally undistinguished fish show the way that massive and apparently selectively neutral polymorphism can provide convenient markers for mapping. As the authors correctly point out animal models may vary in their relevance to man from being totally meaningless to being completely satisfying. For the most part their values lie in suggesting mechanisms or experiments which may be relevant in man and provide experimental systems which cannot be directly approached for ethical reasons. Borgaonkar and Shah have produced a nicely balanced account of the XYY chromosome male and discussed its merits as a syndrome. Though they are unable to resolve the issue, they provide a most valuable review and bibliography. Elizabeth Neufeld discusses the mucopolysaccharidoses and mucopolipidoses storage disease with particular clarity. The other chapters are excellent, as I indicated earlier, and the whole volume deserves to be read from cover to cover.

R. HARRIS

### Announcement

The Dr. Heinz Karger Memorial Foundation invites the submission of papers on the following subjects: 1976—an original research paper on 'Methods for the early diagnosis of genetic disorders'. 1977—an original research paper on 'Molecular biology of metabolic diseases'.

**Conditions:** Manuscripts shall not exceed 20 typewritten pages, including illustrations, tables, and bibliography. Manuscripts marked 'Competition' must reach the publishers, S. KARGER AG, Arnold-Böcklin-Strasse 25, CH-4011 Basle (Switzerland), not later than 28 February 1976 and 1977. The manuscript must be typewritten on one side only, double-spaced, and is to be submitted in quadruplicate, and in accordance with the instructions contained in *The Manuscript* (Rules for the preparation of manuscripts and bibliographies of scientific papers). This booklet can be obtained free of charge from the publishers if the request is marked 'Competition'. Language: English, German, or French. Publication: The winning papers will be published in English in one of the Karger journals. The award for the prizes will be SFr. 7000.00 each. The Council of the Foundation will judge the papers and confer the prizes.

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