

MEDICAL GENETICS

PROCEEDINGS OF THE SYMPOSIUM AT
DEBRECEN-HAJDÚSZOBOSZLÓ, HUNGARY
27-29 APRIL, 1976

Edited by

G. Szabó, M. D., C. Sc., D. Sc.

and

Z. Papp, M. D., C. Sc.

MEDICAL GENETICS

PROCEEDINGS OF THE SYMPOSIUM AT
DEBRECEN-HAJDÚSZOBOSZLÓ, HUNGARY
27-29 APRIL, 1976

Edited by

G. Szabó, M. D., C. Sc., D. Sc.

and

Z. Papp, M. D., C. Sc.

© AKADÉMIAI KIADÓ, BUDAPEST 1977

All rights reserved. No part of this publication may be reproduced, stored in a retrieval system or transmitted, in any form or by any means, electronic, mechanical, photocopying, recording or otherwise, without permission in writing from the copyright owner.

INTERNATIONAL CONGRESS SERIES . 428

Distribution of this publication is being handled by
EXCERPTA MEDICA
305-311 Keizersgracht
Amsterdam
P.O. Box 1126

for the U.S.A. and Canada
ELSEVIER NORTH-HOLLAND INC.
52 Vanderbilt Avenue
New York, N.Y. 10017

for the socialist countries
AKADÉMIAI KIADÓ
Alkotmány u. 21.
H-1363 Budapest
P.O. Box 24

Library of Congress Cataloging in Publication Data

Symposium on Medical Genetics, Debrecen, Hungary, and Hajdúszoboszló, Hungary, 1976.
Medical Genetics.

(International congress series; No. 428)

Includes Indexes.

1. Medical genetics—Congresses. I. Szabó, G. II. Papp, Z. III. Title. IV. Series
[DNLM: 1. Genetics, human—Congresses. 2. Hereditary diseases—Congresses.

QZ50 S992m 1976]

RB155. S944 1976 616'.042 77-21503

ISBN 0-444-90002-0

Joint edition published by Excerpta Medica, Amsterdam and Akadémiai Kiadó, Budapest

Printed in Hungary
77.4050.66-14-1 Alföldi Nyomda, Debrecen



**Debrecen-Hajdúszoboszló, Hungary
27-29 April, 1976**

PRESIDENT OF THE SYMPOSIUM

Prof. G. SZABÓ, M. D., C. Sc., D. Sc. (Debrecen)

Corresponding Member of the Hungarian Academy of Sciences

VICE-PRESIDENTS OF THE SYMPOSIUM

Prof. G. KISZELY, M. D., C. Sc. (Szeged)

President of the Hungarian Society of Human Genetics

Prof. D. SCHULER, M. D., C. Sc., D. Sc. (Budapest)

Secretary General of the Hungarian Society of Human Genetics

SECRETARY GENERAL OF THE SYMPOSIUM

Z. PAPP, M. D., C. Sc. (Debrecen)

MEMBERS OF THE SCIENTIFIC COMMITTEE

E. CZEIZEL, M. D., C. Sc. (Budapest)

Prof. S. R. HOLLÁN, M. D., C. Sc., D. Sc. (Budapest)

Corresponding Member of the Hungarian Academy of Sciences

Prof. J. LÁSZLÓ, M. D., C. Sc., D. Sc. (Budapest)

Prof. Gy. LENÁRT, M. D., C. Sc. (Budapest)

Prof. J. NEMESKÉRI, M. D., C. Sc. (Budapest)

MEMBERS OF THE ORGANIZING COMMITTEE

G. HERPAY, M. D. (Debrecen)

K. HORVÁTH, M. D. (Debrecen)

Gy. JUHÁSZ, M. D. (Debrecen)

L. SZABÓ, M. D. (Debrecen)

É. VÁRADÍ, M. D. (Debrecen)

E. BEDE (Debrecen)

J. HAJDÚ (Debrecen)

I. KALMÁR (Debrecen)

I. KEREKES (Debrecen)

Contents

Preface	13
-------------------	----

I. CHROMOSOME MAPPING AND CLINICAL CYTOGENETICS

FRÉZAL, J. and NGUYEN VAN CONG: Human chromosome mapping, results obtained by cell hybridization	17
PFEIFFER, R. A.: Current aspects of the banding phenomena	27
LAURITSEN, J. G. and THERKELSEN, A. J.: Banding studies on spontaneous and threatened abortion	39
ENDRES, M., STENGEL-RUTKOWSKI, S. and MURKEN, J. D.: Analysis of factors influencing G-band formation, using the trypsin-free method of Walther	49
Osztovics, M., Kiss, P. and Ivády, Gy.: Structural variations of chromosome 9	61
MICHALOVÁ, K., MÁLKOVÁ, J., ČINÁTL, J. and CHRŽ, R.: Differential staining of sister chromatids in human metaphase chromosomes	71
KUČEROVÁ, M. and POLÍVKOVÁ, Z.: Relationship of human heterochromatin and congenital malformations	83
MIKKELSEN, M.: Down's syndrome. Current stage of cytogenetical research	89
MATTEI, J. F., MATTEI, M. G., AYME, S., GOUVERNET, J. and GIRAUD, F.: Inheritance of acrocentric association patterns	107
SERRA, A., MASTROIACOVO, P., SINGH-KAHLON, P. D. and BOVA, R.: Karyotype-phenotype correlation in a girl with r(21) chromosome	117
KOSZTOLÁNYI, G., BÜHLER, E. M. and STALDER, G. R.: Proposed list of the leading clinical features in trisomy 8 mosaicism	127
MAZURCZAK, T., STOLARKA, A., SITO, A. and MATASZEWSKA, K.: Familial [5:12] translocation	135
NAGY, Z.: A case of Marfan's syndrome associated with G mosaic monosomy	143
BOCKOWSKI, K.: Genetics of sex determination and differentiation in man	147
BÜHLER, E. M. and STALDER, G. R.: The human Y chromosome	155
FRACCARO, M., TIEPOLO, L., ZUFFARDI, O., GIAROLA, A. and CHIARA, F.: The role of chromosome imbalance in male subfertility	173
RAPOSA, T. and SRÉTER, L.: Reassociation kinetics of the human Y chromosome studied by Hoechst 33,258	181

LÁSZLÓ, J. and GAÁL, M.: Analysis of cases with isochromosomes for the long arm of the X chromosome	187
GAÁL, M. and LÁSZLÓ, J.: Gonadal dysgenesis and X-autosomal translocation	201
KÖRNER, H. and LISSE, K.: Structural aberrations on the X chromosome	209
POLGÁR, V. and SÁNDOR, Gy.: Familial D/D translocation and its role in fertility	223
MIDRO, A., KOWALSKA-WOCHNA, E., WIŚNIEWSKI, L., TREMBACZOWSKA, M. and MUSIATOWICZ, B.: Report on a case of a 46,XX male	227
BARON, J. and WARENİK-SZYMANKIEWICZ, A.: Cytogenetic and autoradiographic investigations in gonadal dysgenesis	235
FEKETE, G., GÁL, N., DOBOS, M., POLGÁR, V., BŐSZE, P. and LÁSZLÓ, J.: Psychological gender identification in patients with gonosomal aberrations	241
BŐSZE, P., BOGNÁR, B. and LÁSZLÓ, J.: Skeletal abnormalities in gonadal dysgenesis and their relation to the gonad and the karyotype	247
SCHULER, D., DOBOS, M., FEKETE, G., MILTÉNYI, M., KALMÁR, L. and IGALI, S.: "Spontaneous" and in vitro induced chromosome mutations in children treated with immunosuppressive drugs (according to different schedules)	259
SZEMERE, G.: Meiotic and post-meiotic studies in the male mouse exposed to X-rays and their human implications	269
MÜLLER, R., WALLER, M. and WALLER, H.: Chromosomal investigations of fetal tissue and of lymphocyte cultures from healthy women under influence of hormonal contraceptives	277
TILL, G. and CZEIZEL, E.: Chromosome tests with patients working with benzene	289
SZENTESI, I., HORNYÁK, É., and CZEIZEL, E.: Chromosome analysis in PVC workers	293
GEORGIEVA, V. L.: Cytogenetic investigations in agricultural workers in occupational contact with pesticides	297
RASKÓ, I., WENT, M. and JOHNSON, R. T.: Ultraviolet light induced changes in the organization of mitotic chromosomes in porphyria cutanea tarda	301
SZENDE, B., SELLYEI, M., FERENCZ, G. and LAPIS, K.: Cytogenetic studies on CBA mouse fibroblast cultures after spontaneous and methylcholanthrene induced transformation	307
KULCSÁR, G., FEKETE, G., DÁN, P., NÁSZ, I., SCHULER, D. and DOBOS, M.: Lymphoblastic transformation by viruses in Down's syndrome	315
KIRCHNER, M.: Correlations between professional contact with leukaemias and the ability of the peripheral lymphocytes to be stimulated by phytohaemagglutinin	319
SZOLLÁR, J.: Investigation of X-ray-induced chromosome aberrations in "preleukaemic" mammalian cells	327
TZONEVA, M. T., VALKOVA, G., KRACHUNOVA, M., TOZEEVA, A., LUCANOV, L., YORDANOVA, L., GEORGIEVA, V. L., LOZANOVA, T., STOYKOVA, M. and KAZAROVA, M.: Population cytogenetic investigation in Bulgaria	339

II. POPULATION GENETICS AND CONGENITAL MALFORMATIONS

BOCHKOV, N. P.: Medical genetic studies on isolates	347
JACQUARD, A.: The need for precise concepts in genetics	361
NEMESKÉRI, J.: The inbreeding tendencies of the Ivády clan and their effect on the population history of the village of Ivád	365
FERÁK, V. and KROUPOVÁ, Z.: Changes in the population structure of Slovakia and their genetic significance	383
AKHMETELI, M. A.: The role of genetic factors in mycotoxicoses	399
CARTER, C. O.: The aetiology of the common congenital malformations	401
CZEIZEL, E.: Multifactorial aetiology of common congenital malformations	411
EMERY, A. E. H.: Prevention of genetic disease in the population	431
EDWARDS, J. H.: Genetic screening	443
KUČERA, J.: Relation between population genetics and population teratology	449
DENİZ, E.: Morphological aspects of congenital anophthalmia in the calf from the viewpoint of comparative teratology	475
MÉHES, K.: A follow-up study of infants with minor malformations	483
KISS, P., OSZTOVICS, M., PAZONYI, I. and CZEIZEL, E.: Clinical and cytogenetical study of congenital multiple malformations registered in Hungary (1970-1974)	487
BOCHKOVÁ, D. N. and KUANDYKOV, E. U.: Clinical-genealogical analysis of mucoviscidosis	495
LÁSZLÓ, A., PÉNZES, P. and GYURKOVITS, K.: Total amylase activities of serum and mixed saliva samples in homozygous and heterozygous cases with mucoviscidosis	499
TAKÁCS, Ö., SOHÁR, I., LÁSZLÓ, A., PÉNZES, P. and GYURKOVITS, K.: Distribution of serum amylase isoenzymes in homo- and heterozygotes with mucoviscidosis	503
KALAYDJIEVA, L. V. and KREMENSKY, I. M.: Galactose-1-phosphate uridyl-transferase. Normal values and variant forms	511
TASNÁDY, Zs., EMBER, I., JUHÁSZ, E., KARSAI, T. and ELÖDI, P.: Demonstration of heterozygosity in amino-acidopathies	517
KNAPP, A. and SCHLENZKA, K.: The prevalence of varicosity of veins in heterozygotes for phenylketonuria (PKU)	523
TZONEVA, M. and MAVRUDIEVA, M.: Frequency of glucose-6-phosphate dehydrogenase deficiency in some areas of Bulgaria	527
SRŠEŇ, Š. and KOSKA, Ľ.: The prevalence of alkaptonuria in Slovakia	531
SIMONOVITS, I. and KERÉNYI, S.: Sex ratio of Rh(D)-positive offsprings of Rh(D)-negative women	537
ZAHÁLKOVÁ, M. and KUBIKOVÁ, A.: Selection in ABO blood groups. Twin study and sib study	541
GAL, E.: Representation of dermatoglyphic information in computers (structured medical data)	549
JUHÁSZ, Gy., SZILÁGYI, K., SZABÓ, L. and PAPP, Z.: Dermatoglyphic analysis in Turner's syndrome	561

KORCHMÁROS, I., SZALAY, E., FODOR, M. and JÁBLONSZKY, É.: Incidence and heredity of the non-opened lacrimal pathways	567
PAP, K.: Seasonal distribution of the congenital dislocation of the hip	577
MAROSFI, S., UNGVÁRI, G. and PETHŐ, B.: Clinical-genetic studies on schizophrenic patients using electroencephalography	583
KRACHUNOVA, M., MILENKOV, K. and DOBRINOVA, O.: Clinical-genetic polymorphism in manic-depressive psychosis	587
KELEMEN, A.: Influence of parental age at conception in endogenous psychoses	593
KONOTEY-AHULU, F. I. D.: Male procreative superiority in African populations: The fact established and quantified	599
BONNEY, G. E. and KONOTEY-AHULU, F. I. D.: The role of polygamy in the maintenance of genetic variability	609
EIBEN, O. G.: Some genetic aspects of human growth	615
JUVANČ, I., HEVÉR, Ö. and HAJPÁL, A.: Chances to prove non-paternity	621

III. PRENATAL DIAGNOSIS AND GENETIC COUNSELLING

VALENTI, C.: Prenatal genetic diagnosis	629
SCHMID, W.: Prenatal chromosome studies: in situ preparation and analysis of clonally grown amniotic fluid cells. Technique and results in 314 cases	633
BROCK, D. J. H. and SCRIMGEOUR, J. B.: Alphafetoprotein and the prenatal diagnosis of neural tube defects	649
ALKEMA, F. M. J., HUISJES, H. J., DE BRUIJN, H. W. A. and FRIESE, S.: Alpha-1-fetoprotein in amniotic fluid and the antenatal diagnosis of anencephaly and spina bifida	651
PAPP, Z.: Prenatal genetic diagnosis in Debrecen from 1967 to 1976	659
VAMOS-HURWITZ, E., RODESCH, F., PETIT, P., LIEBAERS, I., MATHY, M., HAYEZ, F. and MANDELBAUM, I. M.: Prenatal diagnosis of congenital diseases. A report on 234 amniotic cell cultures	669
ZWINGER, A., JIRÁSEK, J. E., MACEK, M. and SEEMANOVÁ, E.: Prenatal diagnosis of birth defects. Results obtained from examinations of 200 pregnant women	679
SZABÓ, L., JUHÁSZ, Gy. and PAPP, Z.: Fetal sex determination by amniocentesis	685
KLUJBER, L.: Glycosaminoglycans in pregnancy	693
GAL, I.: Vitamin-A in human reproduction	699
JUHÁSZ, E., EMBER, I., TASNÁDY, Zs., KARSAI, T., HAUCK, M. and PAPP, Z.: Amino acid composition of amniotic fluid in early pregnancy	707
EMBER, I., JUHÁSZ, E., TASNÁDY, Zs., KARSAI, T. and PAPP, Z.: Acid soluble glycoproteins in amniotic fluid	713
WIŚNIEWSKI, L., BOGDANIKOWA, B., DROZD, J. and CZOKAŁO, M.: Proteins of the amniotic fluid in 10-12-week-old normal and pathologic pregnancy	719
BLANK, C. E.: Genetic counselling in a British hospital practice	727
FRASER, G. R.: Genetic counselling in the case of non-familial deafness	739

CZEIZEL, E., OSZTOVICS, M. and KISS, P.: Data of three years of genetic counselling	741
WITTWER, B. B.: Epidemiology and counselling in medical genetics. Aims and problems	745
WITKOWSKI, R. and DIETL, H. M.: Genetic counselling and adoption	755
KISS, I., MÁNDI, A. and SZAPPANOS, L.: Familial hypoplasia of the patella	759

IV. HAEMOGLOBINOPATHIES AND IMMUNOGENETICS

LEHMANN, H.: The human haemoglobin molecule	769
HOLLÁN, S. R., DANG HUU LANH, FÖLDI, J., HORÁNYI, M. and SZELÉNYI, J. G.: Duplication of the haemoglobin α -gene	771
RINGELHANN, B., KONOTEY-AHULU, F. I. D., YAWSON, G., BRUCE-TAGOE, A. A., MILLER, A. and HUISMAN, T. H.: The presence of an α -thalassaemia gene in the population of West Africa	783
KONOTEY-AHULU, F. I. D.: The usual and unusual in clinical haemoglobinopathy	791
CEPELLINI, R.: Biological and clinical significance of the so-called transplantation antigens	801
BULANOV, A. G. and CEPELLINI, R.: Genetic markers and disease associations	803
KLEIN, G.: Malignant behaviour and antigen expression in hybrids derived from the fusion of normal with malignant mouse cells	811
PETRÁNYI, G. Gy.: The association of immune responsiveness and disease susceptibility to histocompatibility system	817
GERGELY, J., MEDGYESI, G. A. and RAJNAVÖLGYI, É.: The genetic control of antibody variability	825
SZIGETI, R., RÉVÉSZ, T. and SCHULER, D.: In vitro delayed hypersensitivity to leukaemia associated antigens in Down's syndrome and other conditions	831
ÓNODY, K., BOJÁRSZKY, K. and PETRÁNYI, G. Gy.: Genetic aspects of phytohaemagglutinin stimulation in human lymphocytes	837
STENSZKY, V., SZEGEDI, Gy., ROCHLITZ, Sz. and PETRÁNYI, Gy.: The occurrence of HLA-A1, B8 haplotype in autoimmune disease	843
PIETRZYK, J. J.: Study of the HLA system in children with congenital malformations	851
BACH, K., KRIZSA, F. and KAISER, G. I.: Frequency of HLA antigens in patients with malignant lymphoma	859
KAISER, G. I., LÁSZLÓ, A. and GYURKOVITS, K.: HLA antigens in cystic fibrosis. An association of B18 with the disease	863
HERNÁDI, E., PAÁL, M., AMBRUS, M. and BAJTAI, G.: Investigation of HLA antigens in primary immune deficiency syndrome	865
VARGA, M., BENCZUR, M., GyÓDI, É., ÓNODY, K., PETRÁNYI, G. Gy. and HOLLÁN, S. R.: Correlation between spontaneous cytotoxic activity of peripheral lymphocytes and the HLA system	869

FACHET, J. and ANDÓ, I.: Genetic control of contact sensitivity to oxazolone in inbred and congenitally athymic nude mice	873
ANDÓ, I. and FACHET, J.: Genetic control of IgG response to sheep red blood cells in mice. Studies on inbred, congenic and recombinant lines	879
<i>List of participants</i>	885
<i>Index of authors</i>	901
<i>Subject index</i>	905

Preface

As is known but perhaps not always fully appreciated, human genetics was in a difficult situation after the Second World War in quite a number of countries. Among those who helped to make up for the deficiencies were Prof. M. Hauge and his colleagues in the Institute of Human Genetics, Copenhagen, who, with the active support of the World Health Organization (WHO), held courses on Human Genetics both in Copenhagen and abroad. The participants became not only teachers in medical schools but several of them became active research workers and contributed to the development of genetics.

By 1976 there were already quite a few scientists interested and it seemed to us that further progress in developing medical genetics in Hungary and in the neighbouring countries could best be served not by a new course on medical genetics but by collecting well-known experts, from home and abroad, who could survey the most quickly progressing fields in medical genetics and by creating an opportunity for all the interested researchers to show their own results on posters and discuss them.

We are indebted to WHO, the Hungarian Society of Human Genetics, the Postgraduate Medical School of Budapest, the University Medical School of Debrecen and the sponsors of the Symposium. With this help a Symposium on Medical Genetics was decided on by the Hungarian Society of Human Genetics. Scientists from 22 countries came to take part in the work of the meeting on the following four topics: chromosome mapping and clinical cytogenetics; population genetics and congenital malformations; prenatal diagnosis and genetic counselling; haemoglobinopathies and immunogenetics. There were 59 lectures and 97 contributions as posters. A book of Abstracts had been published by the time of the opening of the Symposium. The Symposium on Medical Genetics took place at Debrecen-Hajdúszoboszló (Hungary), on April 27-29, 1976. Participants were also requested to submit their delivered papers. In the present book 109 papers are included in the same form as they were submitted by the authors, with a few exceptions of corrections of a technical nature.

We hope that the papers of this volume will serve the advancement of medical genetics and help teachers and students in their daily work.

The Editors

I. CHROMOSOME MAPPING AND CLINICAL CYTOGENETICS

Chromosome mapping and clinical cytogenetics are two major areas of research in the field of medical genetics. Chromosome mapping involves the physical localization of genes or genetic markers on chromosomes, while clinical cytogenetics involves the analysis of chromosomal abnormalities in human cells. These two fields are closely related and often work together to improve our understanding of genetic diseases.

Chromosome mapping has been used to identify the chromosomal location of many genes, including those responsible for common diseases such as cancer, heart disease, and mental retardation. This information can be used to develop more effective treatments for these conditions. Clinical cytogenetics is used to diagnose chromosomal abnormalities in patients, such as Down syndrome, Turner syndrome, and Klinefelter syndrome. It can also be used to predict the risk of inheritance of certain genetic disorders.

The combination of chromosome mapping and clinical cytogenetics has led to significant advances in the diagnosis and treatment of genetic disorders. For example, the identification of specific chromosomal rearrangements in cancer patients has led to the development of targeted therapies that are more effective than traditional treatments. Similarly, the use of clinical cytogenetics in conjunction with chromosome mapping has allowed for the identification of new genes and genetic markers that can be used to predict the risk of developing certain diseases.

Overall, the integration of chromosome mapping and clinical cytogenetics is essential for advancing our understanding of genetic diseases and improving patient care. As new technologies continue to emerge, we can expect even more exciting developments in this field in the future.

