

Contents

Part II: Connective Tissue and Purine Metabolism, and Endocrinology

For table of contents of Part I: Carbohydrate, Amino Acid and Lipid Metabolism,
published as Vol. 9 of 'Monographs in Human Genetics', see page VIII of this volume.

Diseases of Connective Tissue, Muscle and Bone

The Mucopolysaccharidoses

<i>Chester, M.A.; Lennartson, G.; Lundblad, A.; Lundsten, J.; Nordén, N.E.; Sjöblad, S.; Svensson, S., and Öckerman, P.-A.</i> (Lund): Isolation and Characterization of Oligosaccharides from Urine of Patients with Abnormal Glycoconjugate Metabolism	2
<i>Kohn, G.; Bach, G.; Lasch, E.; El Massri, M.; Legum, C., and Cohen, M.M.</i> (Jerusalem): A New Phenotypic Variant of α -L-Iduronidase Deficiency	7
<i>Liem, K.O. and Hooghwinkel, G.J.M.</i> (Amsterdam): Sanfilippo B Disease in Two Related Sibships. Biochemical Studies in Patients and Sibs	11
<i>Aula, P.; Raivio, K.; Autio, S.; Thoden, C.-E.; Rapola, J.; Koskela, S.-L., and Yamashina, I.</i> (Helsinki): Four Patients with a New Lysosomal Storage Disorder (Salla Disease)	16
<i>Borud, O.; Torp, K.H., and Dahl, T.</i> (Tromsø): Aspartylglycosaminuria, Urinary Excretion of Aspartylglycosamines Related to Mental Retardation	23

The Mucopolidoses and Fucosidosis

<i>Yabuuchi, H.; Okada, S.; Nishigaki, M., and Kobata, A.</i> (Osaka): Studies on the Pathogenesis of Mucopolidosis	27
<i>Ornøy, A.; Sekeles, E.; Cohen, R., and Kohn, G.</i> (Jerusalem): Electron Microscopy of Cultured Skin Fibroblasts and Amniotic Fluid Cells in the Diagnosis of Hereditary Storage Diseases	32
<i>Katzenstein, M.; Bach, G.; David, R.; Shochet, S.; Shoham, D., and Sadan, N.</i> (Kfar Sava): Three Cases of Mucopolidosis II in an Arab Kindred	40

<i>Sekeles, E.; Ornoy, A.; Cohen, R., and Kohn, G.</i> (Jerusalem): Mucopolipidosis IV: Fetal and Placental Pathology. A Report on Two Subsequent Interruptions of Pregnancy . . .	47
<i>Poenaru, L.; Dreyfus, J.C., and Boué, J.</i> (Paris): Prenatal Diagnosis of Fucosidosis . . .	51
<i>Staal, G.E.J.; Troost, J.; Van Der Heijden, M.C.M.; Borst-Eilers, E.; Schipper-Kester, G.; Moes, M., and Willemse, J.</i> (Utrecht): Two Different Families with α -L-Fucosidase Deficiency	56

Other Disorders

<i>Wang, P.; Clausen, T., and Ørskov, H.</i> (Copenhagen): Salbutamol Inhalations Suppress Attacks of Hyperkalemia in Familial Periodic Paralysis	62
<i>Sørensen, S.A.; Flodgaard, H., and Sørensen, E.</i> (Copenhagen): Serum Alkaline Phosphatase, Serum Pyrophosphatase, Phosphorylethanolamine and Inorganic Pyrophosphate in Plasma and Urine. A Genetic and Clinical Study of Hypophosphatasia	66
<i>Hösli, P. and Rudd, N.</i> (Paris): Alkaline Phosphatase Induction in Fibroblast Cultures: Prenatal Diagnosis and Carrier Detection in Hypophosphatasia	70
<i>Sengers, R.C.A.; Stadhouders, A.M., and Trijbels, J.M.F.</i> (Nijmegen): Metabolic Myopathies Associated with Stunted Growth	75

Disorders of Purine and Pyrimidine Metabolism

<i>Zöllner, N.</i> (Munich): Nutritional Aspects in the Treatment of Inborn Errors	82
<i>Seegmiller, J.E.</i> (La Jolla, Calif.): Progress in Understanding the Mechanism of Immunodeficiency Disease Associated with Defects in Purine Metabolism	88
<i>Willis, R.C. and Seegmiller, J.E.</i> (La Jolla, Calif.): Defining Serum Requirements of Culture Media for Use in Studies of Physiological Effects of Various Genetic Diseases	92
<i>Zoref, E.; Vries, A. de, and Sperling, O.</i> (Petah Tiqva): Kinetic Aspects of Purine Metabolism in Cultured Fibroblasts. A Comparative Study of Cells from Patients Overproducing Purines Due to HGPRT Deficiency and PRPP Synthetase Superactivity	96
<i>Thompson, L.F.; Willis, R.C.; Stoop, J.W., and Seegmiller, J.E.</i> (La Jolla, Calif.): Purine Metabolism in Cultured Human Fibroblasts Derived from Patients Deficient in Enzymes of the Purine Salvage Pathway	100
<i>Staal, G.E.J.; Stoop, J.W.; Zegers, B.J.M.; Vlist, M.J.M. van der; Bruyn, C.H.M. de, and Wadman, S.K.</i> (Utrecht): Red Cell Metabolism in Purine Nucleoside Phosphorylase Deficiency after Enzyme Replacement Therapy	104
<i>Spector, E.B.; Hershfield, M.S., and Seegmiller, J.E.</i> (Los Angeles, Calif.): Purine Reutilization and Synthesis <i>de novo</i> in APRT Deficient Long-Term Lymphocyte Lines	108
<i>Zöllner, N.; Goebel, F.D., and Gröbner, W.</i> (Munich): Partial HGPRT Deficiency: Persistence of Tophi after 12 Years of Therapeutic Normouricemia and Development of a Pheochromocytoma	112
<i>Müller, M.M.; Kraupp, M.; Falkner, G., and Bruyn, C.H.M.M. de</i> (Vienna): Uptake of Purine Bases by HGPRT Deficient Erythrocytes	116
<i>Sperling, O.; Weinberger, A.; Benjamin, D.; Pinkhas, J., and de Vries, A.</i> (Petah Tiqva): Hereditary Renal Hypouricemia: Heterogeneity of Tubular Abnormality	122
<i>Bakay, B.; Nissinen, E.A.; Sweetman, L., and Nyhan, W.L.</i> (La Jolla, Calif.): Analysis of Radioactive and Nonradioactive Purine Bases, Purine Nucleosides and Purine Nucleotides by High-Speed Chromatography on a Single Column	127

<i>Brown, P.R.; Krstulović, A.M., and Hartwick, R.A. (Kingston, R.I.): Selective Analysis of Adenosine, 1-Methylinosine and N²-Methylguanosine by High Pressure Liquid Chromatography</i>	135
<i>Harley, E.H. (Cape Town): Erythrocyte Pyrimidine Metabolism in Pyrimidine 5'-Nucleotidase Deficiency</i>	141

Miscellaneous

<i>Goka, T.J. and Howell, R.R. (Houston, Tex.): Copper Metabolism in Menkes Disease</i>	148
<i>Kopelovich, L. (New York, N.Y.): Cutaneous Manifestations Occurring Systemically in Heritable Forms of Colon Cancer</i>	156
<i>Wöhler, W.; Bartram, C.R.; Schürer, C.C., and Rüdiger, H.W. (Hamburg): Benzpyrene Metabolism in Human Diploid Fibroblasts</i>	165
<i>Rosenfeld, G.B.; Lebenthal, E.; Branski, D.; Mosovich, L., and Baliah, T. (Buffalo, N.Y.): Cholestatic Hepatitis with Renal Tubular Acidosis, Vitamin D Resistant Rickets and Growth Failure</i>	171

Endocrinological Errors

<i>Oelsner, G.; Kraiem, Z.; Lunenfeld, B.; Akstein, E.; Goldman, B., and Serr, D.M. (Tel Hashomer): Congenital Adrenal Hyperplasia. I. Problems of Management Associated with Late Diagnosis. II. Response to Gonadotrophin Releasing Hormone</i>	178
<i>Sack, J.; Katznelson, D.; Kraiem, Z., and Lunenfeld, B. (Tel Hashomer): Management of Congenital Hypoadosteronism</i>	181
<i>Sack J.; Katznelson, D.; Czerniak, P., and Lunenfeld, B. (Tel Hashomer): Early Detection of Thyroid Dyshormonogenesis in Breast-Fed Infant Using a Filter Paper Screening Method</i>	185
<i>Landau, H.; Lasch, E.; Spitz, I.M.; Abu Amara, I., and Russell, A. (Jerusalem): Hypothalamic-Pituitary Axis in Total Lipodystrophy</i>	188
<i>Krieger, I. and Hart, Z. (Detroit, Mich.): Hypometabolism of Nonthyroid Origin in Oculocraniosomatic Neuromuscular Disease</i>	192

Acatalasemia

<i>Aebi, H. and Wyss, S.R. (Bern): Molecular Hybridization in Heterozygotes for Swiss-Type Acatalasemia</i>	200
<i>Takahara, S. and Ogata, M. (Kurashiki-Shi): Erythrocyte Metabolism against Oxidation in Japanese Acatalasemia</i>	205

The Porphyrias

<i>Van Steveninck, J.; Goetj, A.F.P.M. de, and Dubbelman, T.M.A.R. (Leiden): Biochemical Studies on Erythropoietic Protoporphyria</i>	212
<i>Nordmann, Y. and Grandchamp, B. (Colombes): Hereditary Coproporphyria: Demonstration of a Genetic Defect in Coproporphyrinogen Metabolism</i>	217
<i>Douer, D.; Schoenfeld, N.; Weinberger, A.; Pinkhas, J., and Atsmon, A. (Petah Tiqva): Favourable Effect of Intravenous Propranolol in Acute Intermittent Porphyria</i>	223
<i>Kreimer-Birnbaum, M.; Franco-Saenz, R., and White, P. (Toledo, Ohio): Acute Intermittent Porphyria: Detection of Carriers</i>	227

Author Index	235
Subject Index	237

Part I: Carbohydrate, Amino Acid and Lipid Metabolism

Published as Vol. 9 of 'Monographs in Human Genetics'.

Disorders of Carbohydrate Metabolism

<i>Schapira, F.; Hatzfeld, A., and Gregori, C. (Paris): Structural Mutation of Aldolase B in Hereditary Fructose Intolerance. Electrofocusing Results</i>	2
<i>Koster, J.F.; Slee, R.G., and Fernandes, J. (Rotterdam): Lactic Acidosis Due to a Defect in the Pyruvate Dehydrogenase Complex. A Possible Brain Pyruvate Dehydrogenase Phosphatase Deficiency</i>	7
<i>Blass, J.P.; Cederbaum, S.D., Kark, R.A.P., and Rodriguez-Budelli, M. (Los Angeles, Calif.): Pyruvate Dehydrogenase Deficiency in 35 Patients</i>	12
<i>Wasserzug, O.; Szeinberg, A., and Sperling, O. (Petah Tiqva): Erythrocyte Glutathione Reductase in Gout and in Glucose-6-Phosphate Dehydrogenase Deficiency</i>	16
<i>Saddi, R.; Feingold, J., Fagard, R., and Eschwege, E. (Paris): Idiopathic Haemochromatosis Diabetes: Assessment of Serum Iron and Transferrin Saturation in High and Low Risk Populations for Diabetes Mellitus</i>	20
<i>Staal, G.E.J.; Van Biervliet, J.; Vlug, A.M.C., and Akkerman, J.W.N. (Utrecht): Three Variants of Glucose-Phosphate Isomerase Deficiency</i>	25

Glycogen Storage Diseases

<i>Davidson, A.G.F.; Wong, L.T.K.; Kirby, L.; Tze, W.J.; Rigg, J.M., and Applegarth, D.A. (Vancouver): Glycogen Storage Disease Type I: Effect of Continuous Nocturnal Nasogastric Feeding</i>	29
<i>Slonim, A.E.; Terry, A.; Greene, H.L.; Lacy, W.W., and Burr, I.M. (Nashville, Tenn.): Amino Acid and Hormonal Response to Long-Term Nocturnal Nasogastric Feeding Therapy of Glycogen Storage Disease Type I (GSD-I)</i>	37
<i>Tarui, S.; Kono, N.; Kuwajima, M., and Ikura, Y. (Osaka): Type VII Glycogenosis (Muscle and Erythrocyte Phosphofructokinase Deficiency)</i>	42

Disorders of Amino Acid Metabolism

<i>Irving, C.S.; Nissim, I., and Lapidot, A. (Rehovot): Whole Body Amino Acid Pools and Turnover Rates Determined by a Stable Isotope Gas Chromatographic Mass Spectrometric Methodology</i>	50
<i>Von Wendt, L.; Simila, S.; Hirvasniemi, A., and Suvanto, E. (Oulu): Nonketotic Hyperglycinemia. A Clinical Analysis of 19 Finnish Patients</i>	58
<i>Russell, A.; Statter, M., and Abzug-Horowitz, S. (Jerusalem): Methionine Dependent Glutamic Acid Formiminotransferase Deficiency. Human and Experimental Studies in its Therapy</i>	65
<i>Teijema, H.L.; Brubakk, A.M.; Gelderen, H.H. van, and Ruys, J.H. (Leiden): Glutamic Acidemia</i>	75
<i>Cespedes, C. de; Loria, A.R., Estrada, Y.; Sweetman, L., and Nyhan, W.L. (San José): The Diagnosis and Management of Propionic Acidemia</i>	80

<i>Hammersen, G.; Wille, L.; Schmidt, H.; Lutz, P., and Bickel, H. (Heidelberg): Maple Syrup Urine Disease: Emergency Treatment of the Neonate</i>	84
<i>Spielberg, S.P.; Boxer, L.A.; Oliver, J.M.; Butler, E.J., and Schulman, J.D. (Indianapolis, Ind.): Altered Phagocytosis and Microtubule Function in Leukocytes from a Patient with Severe Glutathione Synthase Deficiency (5-Oxoprolinuria)</i>	90

Phenylketonuria

<i>Cohen, B.E.; Szeinberg, A.; Levine, Y.; Peled, I.; Pollack, S.; Crispin, M., and Normand, M. (Tel Aviv): Phenylketonuria (PKU) in Israel</i>	95
<i>Tischler, B. and Lowry, R.B. (New Westminster, B.C.): Phenylketonuria in British Columbia, Canada</i>	102
<i>Trefz, F.K.; Bartholomé, K.; Bickel, H.; Lutz, P., and Schmidt, H. (Heidelberg): In vivo Determination of Phenylalanine Hydroxylase Activity Using Heptadeutero-Phenylalanine and Comparison to the <i>in vitro</i> Assay Values</i>	108
<i>Lombeck, I.; Kasperek, K.; Feinendegen, L.E., and Bremer, H.J. (Düsseldorf): Trace Element Disturbances in Dietetically Treated Patients with Phenylketonuria and Maple Syrup Urine Disease</i>	114

Disorders of Tyrosine Metabolism

<i>Cohen, B.E.; Keren, G.; Crispin, M.; Horwitz, A.; Szeinberg, A., and Legum, C. (Tel Hashomer): Congenital Tyrosinemia</i>	118
<i>Modilevsky, T.; Manor, E., and Marmor, A. (Be'er Ya'acov): Alkaptonuria: Prevalence among Bedouins Related by Consanguinity</i>	123
<i>Hösli, P.; Griscelli, C., and Good, R.A. (Paris): Chediak-Higashi Syndrome I: Biochemical Abnormality and Prospects for Prenatal Diagnosis</i>	126

Sulphur Containing Amino Acids

<i>States, B.; Harris, D.; Hummeler, K., and Segal, S. (Philadelphia, Pa.): Amino Acid Disorders in Tissue Culture Cells: Effects of Mycoplasma and Antibiotics on Cystine Metabolism</i>	131
<i>Griffiths, R. (St. Andrews): Cystathionine β-Synthase Deficiency: Observations on the Biochemical Lesion in a Vitamin B₆ Non-Responsive Patient</i>	135
<i>Wada, Y.; Narisawa, K., and Arakawa, T. (Nagoya): Infantile Type of Homocystinuria with 5,10-Methylenetetrahydrofolate Reductase Deficiency</i>	140

Disorders of Amino Acid Transport

<i>Segal, S. (Philadelphia, Pa.): Inherited Disorders of Transport: Aminoacidurias</i>	147
<i>Law, E.A. and Sardharwalla, I.B. (Aberdeen): A New Type of Heterozygote of Familial Renal Iminoglycinuria</i>	152

The Lipidoses

Gluco- and Galactoside Storage Diseases

<i>Blonder, E. and Klibansky, C. (Petah Tiqva): Inhibition of Human Leukocyte Glucocerebrosidase by Series of Tweens. Dependence on Interaction with Other Detergents</i>	156
<i>Silverstein, E.; Friedland, J., and Vuletin, J.C. (Brooklyn, N.Y.): Elevated Angiotensin-</i>	

Converting Enzyme in Type 1 and 2 Gaucher's Disease, and Elevated Serum Lysozyme in Type 1; Association of Hepatic Fibrosis with Long-Spacing Collagen Fibrils in Type 2	163
--	-----

Ganglioside Storage Diseases

<i>Padeh, B.; Shachar, S.; Modan, M., and Goldman, B.</i> (Tel Hashomer): Screening and Prevention of Tay-Sachs Disease in Israel	170
<i>Dreyfus, J.C.; Poenaru, L., and Boue, J.</i> (Paris): Characterization of a Variant of β -Hexosaminidase	176
<i>Wood, S. and Applegarth, D.A.</i> (Vancouver): Juvenile Sandhoff Disease. Report of an Additional Case with Family Studies	182
<i>Navon, R.; Wiselter, J., and Modan, M.</i> (Tel Hashomer): A Modified Method for Prenatal Diagnosis of Tay-Sachs Disease in Cell-Free Amniotic Fluid	186

Neutral Lipid Storage Disease

<i>Arbisser, A.; Brothers Arbisser, L.; Garcia, C.A.; Akhtar, M.; Morizot Moore, C., and Howell, R.R.</i> (Houston, Tex.): Ocular Findings in Acid Lipase Deficiency	193
--	-----

Sphingomyelin Lipidoses

<i>Mazière, J.C.; Mazière, C., and Hösli, P.</i> (Paris): An Ultramicrochemical Assay for Sphingomyelinase. Rapid Prenatal Diagnosis of a Fetus at Risk for Niemann-Pick Disease	198
--	-----

General Subjects

Screening and Prenatal Diagnosis

<i>Watts, R.W.E.</i> (Harrow): Recent Advances in Screening for Inborn Errors of Metabolism	204
<i>Reinecke, C.J.</i> (Potchefstroom): Screening for Inborn Errors of Metabolism: A Multiple Test Program	212
<i>Kleijer, W.J.; Niermeijer, M.F., and Galjaard, H.</i> (Rotterdam): Prenatal Diagnosis of Genetic Metabolic Diseases in 118 Pregnancies at Risk	217
<i>Golbus, M.S.</i> (San Francisco, Calif.): The Use of Fetal Blood for the Prenatal Diagnosis of Genetic Defects	222
<i>Schapira, G. and Dreyfus, J.C.</i> (Paris): Serum Enzymes and Inborn Errors of Metabolism in Man	227

Mutant Cell Repository

<i>Coriell, L.L.</i> (Camden, N.J.): Mutant Cell Repository Workshop: Opening Remarks ..	231
<i>Greene, A.E.</i> (Camden, N.J.): Programs of the Human Genetic Mutant Cell Repository	233
<i>Coriell, L.L.</i> (Camden, N.J.): Quality Control	239
<i>Seegmiller, J.E.</i> (La Jolla, Calif.): Collection and Uses of Lymphocyte Cultures	244
<i>Greene, A.E.; Manduka, M., and Coriell, L.L.</i> (Camden, N.J.): SV40 Virus Transformation of Genetic Mutant Cell Cultures	248

Author Index	253
Subject Index	255