

# Contents

Preface	xiii
Introduction	xv
<b>PART I:</b> Monogenic Diabetes: Genetic Defects of $\beta$ -Cell Development and Function	
Chapter 1: Monogenic Diabetes: Maturity-Onset Diabetes of the Young (MODY) and Neonatal Diabetes <i>R.N. Naylor, S.A.W. Greeley, D. Carmody, J.L. Hwang, M. Sanyoura, L.R. Letourneau, G.I. Bell, and L.H. Philipson</i>	3
<hr/>	
Part I Case Studies	37
Case 1: <i>KCNJ11</i> – DEND: Neonatal Diabetes <i>L.R. Letourneau, M. Sanyoura, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	41
<hr/>	
Case 2: <i>ABCC8</i> Neonatal Diabetes <i>L.R. Letourneau, M. Sanyoura, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	44
<hr/>	

Case 3:	<i>INS</i> – Insulin Gene Mutation in Neonatal Diabetes <i>L.R. Letourneau, M. Sanyoura, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	47
Case 4:	<i>STAT3</i> and Neonatal Diabetes <i>D.W. Hansen, J.C. Sanchez, and T.S. Hannon</i>	51
Case 5:	<i>GATA6</i> – Heart and Pancreas <i>T. Triolo and K. Simmons</i>	55
Case 6:	<i>KCNJ11</i> R201C – Neonatal Diabetes <i>L.M. Nally and D.M. Wilson</i>	61
Case 7:	<i>PDX1</i> – Pancreatic Agenesis <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	67
Case 8:	Neonatal Diabetes and Congenital Hypothyroidism: <i>GLIS3</i> <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	70
Case 9:	<i>EIF2AK3</i> : Wolcott-Rallison Syndrome <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.S. Ladsaria, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	73
Case 10:	Transient Neonatal Diabetes <i>E.O. Buschur and K. Dungan</i>	76
Case 11:	GCK-MODY <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	81

Case 12:	HNF1A – MODY <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	84
Case 13:	HNF4A – MODY <i>M. Sanyoura, L.R. Letourneau, L. Dickens, S.A.W. Greeley, R.N. Naylor, G.I. Bell, and L.H. Philipson</i>	87
Case 14:	MODY as a Possible Diagnosis <i>I. Jamil and J.L. Gilden</i>	90
Case 15:	Could This Be MODY vs. Type 2? <i>B.Z. Osmani, J.L. Gilden, A. Moid, and C. Harris</i>	95
Case 16:	Misclassification of MODY 3 as Type 1 <i>L. Song, J.N. McNulty, and I. Lingvay</i>	100
Case 17:	Wolfram Syndrome WFS1 <i>J. Sarhis-Avigdor and E. Cengiz</i>	105
Case 18:	Mitochondrial Diabetes <i>C. Sullivan and S. Alexanian</i>	109
Case 19:	Mitochondrial Diabetes: m.3243A>G Mutation <i>B.P. Galm, S. Sirrs, and R.O. Yeung</i>	115
Case 20:	mDNA3243A>G <i>A.L. Frederiksen and H. Beck-Nielsen</i>	120
Case 21:	Maternally Inherited Diabetes and Deafness: 23-Year Follow-Up <i>I. Ahmad and I.B. Hirsch</i>	125
Case 22:	Glycogen Storage Type 1A Presenting with Type 2 Diabetes <i>D. Sistla, M. Korytkowski, and S.Y. Mon</i>	130

<b>PART II:</b>	Insulin Resistance, Genetic Defects in Insulin Action, and Diseases of Exocrine Pancreas	
Chapter 2:	Insulin Resistance: Molecular Biology and Pathophysiology <i>C.C. Low Wang and B. Draznin</i>	135
Chapter 3:	The Lipodystrophy Syndromes: Clinical Features and Treatments <i>M. Mattingly, A. Valencia, E. Cochran, R.J. Brown, and P. Gorden</i>	155
Chapter 4:	Diabetes in Diseases of the Exocrine Pancreas <i>J. Abramowitz, U. Gunasekaran, X.F. Samaropoulos, A. Tumyan, and I. Lingvay</i>	181
Part II	Case Studies	215
Case 23:	Type A Insulin Resistance <i>A. Stears and R.K. Semple</i>	218
Case 24:	Donohue Syndrome (Leprechaunism) <i>C. Peters and R.K. Semple</i>	225
Case 25:	Rabson-Mendenhall Syndrome <i>R. Williams and R.K. Semple</i>	232
Case 26:	Familial Partial Lipodystrophy II (Dunnigan's Syndrome) <i>P.K. Cabandugama, S.A. Brietzke, M.J. Gardner, and J.R. Sowers</i>	239
Case 27:	Familial Partial Lipodystrophy <i>K.E. Kreider and M.N. Feinglos</i>	243
Case 28:	Severe Insulin Resistance and Lipodystrophy <i>K. Mekala and J. Aloï</i>	248

Case 29:	Lipodystrophy Due to PPAR- $\gamma$ Gene Mutation <i>A. Hodes, R. Brown, E. Cochran, and M. Zilbermint</i>	253
Case 30:	Congenital Generalized Lipodystrophy <i>H. Beck-Nielsen</i>	259
Case 31:	Pancreatogenic Diabetes <i>N. Zherdeva and B. Mankovsky</i>	263
Case 32:	A Case of Pancreatogenic Diabetes Due to Alcohol Abuse <i>B.Z. Osmani, J.L. Gilden, and A. Moid</i>	267
Case 33:	Post-Pancreatectomy Diabetes <i>E.D. Buras, R.Y. Gianchandani, and J. Wyckoff</i>	273
Case 34:	Diabetes and Hemochromatosis <i>D. Maggi, A. Aleo, L. Briatore, C. Mazzucchelli, L. Fontana, and R. Cordera</i>	277
Case 35:	Bronze Diabetes <i>E.K.P. Meyerhoff</i>	281
Case 36:	Type 2 Diabetes in Liver Cirrhosis and Hepatocellular Carcinoma <i>S. Ravi, A. Moid, J.L. Gilden, and D. Karam</i>	285
Case 37:	Poor Glycemic Control in Insulin-Treated Diabetes Due to Anti-Insulin Antibodies <i>D.S. Church and R.K. Semple</i>	290
Case 38:	Cystic Fibrosis–Related Diabetes <i>R. Iyengar, R.Y. Gianchandani, and J. Wyckoff</i>	296
Case 39:	Pembrolizumab and Diabetic Ketoacidosis <i>F. Cottini and K. Dungan</i>	300

Case 40:	Glycemic Control in Patient with Multiple Chronic Complications <i>A. Iqbal, J.L. Gilden, B.G. Theckedath, A. Moid, S. Rapaka, and D. Dai</i>	305
Case 41:	Diabetes and Exposure to Agent Orange <i>B.Z. Osmani, J.L. Gilden, and C. Harris</i>	309
Case 42:	Pasireotide-Induced Hyperglycemia <i>K. Brau and A. Drincic</i>	313
Case 43:	Hemoglobin A1C vs. Fructosamine <i>A. Iqbal, J.L. Gilden, B.G. Theckedath, S. Rapaka, and D. Dai</i>	318
<b>PART III:</b>	<b>Endocrinopathies, Immune-Mediated Diabetes, Diabetes of Unknown Cause, and Other Genetic Syndromes Sometimes Associated with Diabetes</b>	
Chapter 5:	Diabetes Secondary to Endocrinopathies <i>J.M. Silverstein, K. Sterl, J.L. Cartier, P. Cruz-Bravo, C.A. Davis, S. Sadiq, and J.B. McGill</i>	325
Chapter 6:	Immune-Mediated Diabetes <i>M.F. Salam, J.B. McGill, and J.W. Hughes</i>	357
Chapter 7:	Diabetes of Unknown Cause <i>Y. Azhar, C.A. Baker, and N. Rasouli</i>	383
Chapter 8:	Genetic Syndromes Associated with Diabetes <i>A.C.H. Wood, D. Abreu, A.L. Clark, M.A. Malbas, J.E. Sprague, and J.B. McGill</i>	395
Part III	Case Studies	423
Case 44:	Acromegaly and Diabetic Ketoacidosis <i>P. Akkireddy and A. Drincic</i>	426

Case 45:	Cushing's Syndrome and Diabetes <i>K. Peicher and L. Meneghini</i>	431
Case 46:	Pheochromocytoma and Diabetes <i>K. Modzelewski and D. Steenkamp</i>	436
Case 47:	Hypertension plus Hyperglycemia May Be Equal to Pheochromocytoma <i>L.L. Yanes Cardozo, V. Garla, and L.F. Lien</i>	442
Case 48:	Pheochromocytoma and Preexisting Diabetes <i>P. Akkireddy and A. Drincic</i>	447
Case 49:	Glucagonoma as a Cause of Diabetes <i>H. Beck-Nielsen</i>	452
Case 50:	Type 1 or Type 2 Diabetes vs. LADA <i>I. Jamil and J.L. Gilden</i>	455
Case 51:	LADA Masquerading as Type 2 Diabetes <i>N. Misaghian-Xanthos and L. Young</i>	459
Case 52:	LADA during the Obesity Epidemic <i>L. Viswanathan and C. Vaz</i>	462
Case 53:	LADA in Older Adults <i>L. Viswanathan and C. Vaz</i>	467
Case 54:	Stiff Person Syndrome <i>H.N. Kim and N. Mathioudakis</i>	472
Case 55:	Polyglandular Failure and Diabetes <i>B.Z. Osmani, J.L. Gilden, A. Moid, and D. Dai</i>	476
Case 56:	Type B Insulin Resistance Syndrome <i>S. Ahern, H. Nguyen, Y. Lyou, S. Singh, and P.H. Wang</i>	481

Case 57:	Remission of Type B Insulin Resistance <i>J. Giordano, M.S. Kalia-Reynolds, C. Houston, R.J. Tanenberg, R.J. Brown, R.K. Semple, and F.J. Cook</i>	486
Case 58:	Ketosis-Prone Type 2 Diabetes <i>J. Patel and C.M. Knight</i>	492
Case 59:	Ketosis-Prone Diabetes: Case Report <i>A. Milanesi and J.E. Weinreb</i>	497
Case 60:	Ketosis-Prone Diabetes <i>S. Ahmed and R.J. Tenenberg</i>	501
Case 61:	Prader-Willi Syndrome <i>J.L. Gilden and B.Z. Osmani</i>	506
Case 62:	Bardet-Biedl Syndrome <i>J.J. Iyengar, R.Y. Gianchandani, and J. Wyckoff</i>	512
Case 63:	Williams-Beuren Syndrome and Diabetes <i>M. Gaule and A. Drincic</i>	517
Case 64:	Spinal and Bulbar Muscular Atrophy and Diabetes <i>S. Ravi, J.L. Gilden, A. Moid, B.G. Theckedath, and J. Afranie-Sakyi</i>	522
Index		527