

Clinical Outcomes, Physical Disorders (L-N)

- Labauge, P., Cavalier, L., Ichalalene, L. and Castelnovo, G. (1998). Friedreich's ataxia and hereditary vitamin E deficiency. Case study. *Revue Neurologique* **154**: 339-341 [In French].
- Labauge, P., Ouazzani, R., M'Rabet, A., Grid, D., Genton, P., Dravet, C., Chkili, T., Beck, C., *et al.* (1997). Allelic heterogeneity of Mediterranean myoclonus and the cystatin B gene. *Annals of Neurology* **41**: 686.
- Lachaux, A., Bouvier, R., Loras-Duclaux, I., Chappuis, J. P., Meneguzzi, G. and Ortonne, J. P. (1999). Isolated deficient alpha6beta4 integrin expression in the gut associated with intractable diarrhea. *Journal of Pediatric Gastroenterology and Nutrition* **29**: 395-401.
- Lafreniere, R. G., MacDonald, M. L., Dube, M. P., MacFarlane, J., O'Driscoll, M., Brais, B., Meilleur, S., Brinkman, R. R., *et al.* (2004). Identification of a novel gene (HSN2) causing hereditary sensory and autonomic neuropathy type II through the Study of Canadian Genetic Isolates. *American Journal of Human Genetics* **74**: 1064-1073.
- Lagier-Tourenne, C., Boltshauser, E., Breivik, N., Gribaa, M., Betard, C., Barbot, C. and Koenig, M. (2004). Homozygosity mapping of a third Joubert syndrome locus to 6q23. *Journal of Medical Genetics* **41**: 273-277.
- Lahat, H., Eldar, M., Levy-Nissenbaum, E., Bahan, T., Friedman, E., Khoury, A., Lorber, A., Kastner, D. L., *et al.* (2001). Autosomal recessive catecholamine- or exercise-induced polymorphic ventricular tachycardia: clinical features and assignment of the disease gene to chromosome 1p13-21. *Circulation* **103**: 2822-2827.
- Lahlou, N., Clement, K., Carel, J. C., Vaisse, C., Lotton, C., Le Bihan, Y., Basdevant, A., Lebouc, Y., *et al.* (2000). Soluble leptin receptor in serum of subjects with complete resistance to leptin: relation to fat mass. *Diabetes* **49**: 1347-1352.
- Lam, W., Chan, H. and Sillence, D. (2003). Desbuquois syndrome: Clinical and radiological report of the first two Chinese cases from a consanguineous family. *Journal of Paediatrics and Child Health* **39**: 707-712.
- Lammer, E. J., Scholes, T. and Abrams, L. (2001). Autosomal recessive tetralogy of Fallot, unusual facies, communicating hydrocephalus, and delayed language development: a new syndrome? *Clinical Dysmorphology* **10**: 9-13.
- Landau, D., Cohen, D., Shalev, H., Pinsk, V., Yerushalmi, B., Zeigler, M. and Birk, O. S. (2004). A novel mutation in the SLC17A5 gene causing both severe and mild phenotypes of free sialic acid storage disease in one inbred Bedouin kindred. *Molecular Genetics and Metabolism* **82**: 167-172.
- Langer, R., Al-Gazali, L., Haas, D., Raupp, P. and Varady, E. (2004). [Osseous abnormalities and CT findings in Stueve-Wiedemann-Syndrome (SWS)] [In

German]. *RoFo : Fortschritte auf dem Gebiete der Rontgenstrahlen und der Nuklearmedizin* **176**: 215-221.

- Larnaout, A., Belal, S., Ben Hamida, C., Ben Hamida, M. and Hentati, F. (1998). Atypical ataxia telangiectasia with early childhood lower motor neuron degeneration: a clinicopathological observation in three siblings. *Journal of Neurology* **245**: 231-235.
- Lasa, A., Piccolo, F., de Diego, C., Jeanpierre, M., Colomer, J., Rodriguez, M. J., Urtizberea, J. A., Baiget, M., *et al.* (1998). Severe limb girdle muscular dystrophy in Spanish gypsies: further evidence for a founder mutation in the gamma-sarcoglycan gene. *European Journal of Human Genetics* **6**: 396-399.
- Lau, I. F., Soardi, F. C., Lemos-Marini, S. H., Guerra Jr, G., Jr., Baptista, M. T. and De Mello, M. P. (2001). H28+C insertion in the CYP21 gene: a novel frameshift mutation in a Brazilian patient with the classical form of 21-hydroxylase deficiency. *Journal of Clinical Endocrinology and Metabolism* **86**: 5877-5880.
- Leal, A., Morera, B., Del Valle, G., Heuss, D., Kayser, C., Berghoff, M., Villegas, R., Hernandez, E., *et al.* (2001). A second locus for an axonal form of autosomal recessive Charcot-Marie-Tooth disease maps to chromosome 19q13.3. *American Journal of Human Genetics* **68**: 269-274.
- Leal, G. F. and Da-Silva, E. O. (1999). Limb-girdle muscular dystrophy with apparently different clinical courses within sexes in a large inbred kindred. *Journal of Medical Genetics* **36**: 714-718.
- Leal, S. M., Apaydin, F., Barnwell, C., Iber, M., Kandogan, T., Pfister, M., Braendle, U., Cura, O., *et al.* (1998). A second Middle Eastern kindred with autosomal recessive non-syndromic hearing loss segregates DFNB9. *European Journal of Human Genetics* **6**: 341-344.
- Lebel, R. R. and Gallagher, W. B. (1989). Wisconsin consanguinity studies. II. Familial adenocarcinomatosis. *American Journal of Medical Genetics* **33**: 1-6.
- Lecube, A., Hernandez, C., Oriola, J., Galard, R., Gemar, E., Mesa, J. and Simo, R. (2002). V804M RET mutation and familial medullary thyroid carcinoma: report of a large family with expression of the disease only in the homozygous gene carriers. *Surgery* **131**: 509-514.
- Lee, W. S., Chong, L. A., Begum, S., Abdullah, W. A., Koh, M. T. and Lim, E. J. (2001). Factor V inhibitor in neonatal intracranial hemorrhage secondary to severe congenital factor V deficiency. *Journal of Pediatric Hematology/Oncology* **23**: 244-246.
- Leegwater, P. A., Konst, A. A., Kuyt, B., Sandkuijl, L. A., Naidu, S., Oudejans, C. B., Schutgens, R. B., Pronk, J. C., *et al.* (1999). The gene for leukoencephalopathy with vanishing white matter is located on chromosome 3q27. *American Journal of Human Genetics* **65**: 728-734.

- Leeners, B., Funk, A., Cotarelo, C. L. and Sauer, I. (2004). Two sibs with fibrochondrogenesis. *American Journal of Medical Genetics* **127A**: 318-320.
- Lefevre, C., Audebert, S., Jobard, F., Bouadjar, B., Lakhdar, H., Boughdene-Stambouli, O., Blanchet-Bardon, C., Heilig, R., *et al.* (2003). Mutations in the transporter ABCA12 are associated with lamellar ichthyosis type 2. *Human Molecular Genetics* **12**: 2369-2378.
- Lehmann, O. J., El-ashry, M. F., Ebenezer, N. D., Ocaka, L., Francis, P. J., Wilkie, S. E., Patel, R. J., Ficker, L., *et al.* (2001). A novel keratocan mutation causing autosomal recessive cornea plana. *Investigative Ophthalmology & Visual Science* **42**: 3118-3122.
- Leiberman, E., Pesler, D., Parvari, R., Elbedour, K., Abdul-Latif, H., Brown, M. R., Parks, J. S. and Carmi, R. (2000). Short stature in carriers of recessive mutation causing familial isolated growth hormone deficiency. *American Journal of Medical Genetics* **90**: 188-192.
- Lemire, E., Hildes-Ripstein, G., Reed, M. and Chudley, A. (1998). SAMS: provisionally unique multiple congenital anomalies syndrome consisting of short stature, auditory canal atresia, mandibular hypoplasia, and skeletal abnormalities. *American Journal of Medical Genetics* **75**: 256-260.
- Lench, N. J., Markham, A. F., Mueller, R. F., Kelsell, D. P., Smith, R. J., Willems, P. J., Schatteman, I., Capon, H., *et al.* (1998). A Moroccan family with autosomal recessive sensorineural hearing loss caused by a mutation in the gap junction protein gene connexin 26 (GJB2). *Journal of Medical Genetics* **35**: 151-152.
- Leroy, J. G., Leroy, B. P., Emmery, L. V., Messiaen, L. and Spranger, J. W. (2004). A new type of autosomal recessive spondyloepiphyseal dysplasia tarda. *American Journal of Medical Genetics* **125A**: 49-56.
- Leutelt, J., Oehlmann, R., Younus, F., van den Born, L. I., Weber, J. L., Denton, M. J., Mehdi, S. Q. and Gal, A. (1995). Autosomal recessive retinitis pigmentosa locus maps on chromosome 1q in a large consanguineous family from Pakistan. *Clinical Genetics* **47**: 122-124.
- Leuzzi, V., Rinna, A., Gallucci, M., Di Capua, M., Dionisi-Vici, C., Longo, D. and Bertini, E. (2000). Ataxia, deafness, leukodystrophy: inherited disorder of the white matter in three related patients. *Neurology* **54**: 2325-2328.
- Levadoux, M., Michel, G., Gadea, J., Jouve, J. L., Jacquemier, M. and Bollini, G. (1999). Osteopetrosis: diagnostic and therapeutic management. Apropos of 5 cases. *Revue de Chirurgie Orthopedique et Reparatrice de L'appareil Moteur* **85**: 627-631.
- Li, A., Tedde, R., Krozowski, Z. S., Pala, A., Li, K. X., Shackleton, C. H., Mantero, F., Palermo, M., *et al.* (1998). Molecular basis for hypertension in the "type II variant" of apparent mineralocoid excess. *American Journal of Human Genetics* **63**: 370-379.

- Liang, Y., Wang, A., Probst, F. J., Arhya, I. N., Barber, T. D., Chen, K. S., Deshmukh, D., Dolan, D. F., *et al.* (1998). Genetic mapping refines DFNB3 to 17p11.2, suggests multiple alleles of DFNB3, and supports homology to the mouse model shaker-2. *American Journal of Human Genetics* **62**: 904-915.
- Liburd, N., Ghosh, M., Riazuddin, S., Naz, S., Khan, S., Ahmed, Z., Liang, Y., Menon, P. S., *et al.* (2001). Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith-Magenis syndrome. *Human Genetics* **109**: 535-541.
- Lie, R. T., Wilcox, A. J. and Skjærven, R. (1994). A population-based study of the risk of recurrence of birth defects. *The New England Journal of Medicine* **331**: 1-4.
- Liede, A., Malik, I. A., Aziz, Z., Rios Pd Pde, L., Kwan, E. and Narod, S. A. (2002). Contribution of BRCA1 and BRCA2 mutations to breast and ovarian cancer in Pakistan. *American Journal Human Genetics* **71**: 595-606.
- Lin, Z., deMello, D. E., Batanian, J. R., Khammash, H. M., DiAngelo, S., Luo, J. and Floros, J. (2000). Aberrant SP-B mRNA in lung tissue of patients with congenital alveolar proteinosis (CAP). *Clinical Genetics* **57**: 359-369.
- Lin-Su, K., Zhou, P., Arora, N., Betensky, B. P., New, M. I. and Wilson, R. C. (2004). In vitro expression studies of a novel mutation delta299 in a patient affected with apparent mineralocorticoid excess. *Journal of Clinical Endocrinology and Metabism* **89**: 2024-2027.
- Lipson, A. H. (1995). Amelia of the arms and femur/fibula deficiency with splenogonadal fusion in a child born to a consanguineous couple. *American Journal of Medical Genetics* **55**: 265-268.
- Liu, T. T., Chiang, S. H., Wu, S. J. and Hsiao, K. J. (2001). Tetrahydrobiopterin-deficient hyperphenylalaninemia in the Chinese. *Clinica Chimica Acta* **313**: 157-169.
- Liu, X., Xu, L., Zhang, S. and Xu, Y. (1994). Epidemiological and genetic studies of congenital profound deafness in general population of Sichuan, China. *American Journal of Medical Genetics* **53**: 192-195.
- Liu, X. Z., Blanton, S. H., Bitner-Glindzicz, M., Pandya, A., Landa, B., MacArdle, B., Rajput, K., Bellman, S., *et al.* (2001). Haplotype analysis of the USH1D locus and genotype-phenotype correlations. *Clinical Genetics* **60**: 58-62.
- Lo, B., Faiyaz-Ul-Haque, M., Banwell, B., Blaser, S., Paterson, A. D., Tsui, L. C. and Teebi, A. S. (2004). The locus responsible for horizontal gaze palsy/progressive scoliosis and brainstem hypoplasia is refined to a 9-cM region on chromosome 11q23. *Clinical Genetics* **65**: 137-142.

- Loeffen, J., Elpeleg, O., Smeitink, J., Smeets, R., Stockler-Ipsiroglu, S., Mandel, H., Sengers, R., Trijbels, F., *et al.* (2001). Mutations in the complex I *NDUFS2* gene of patients with cardiomyopathy and encephalomyopathy. *Annals of Neurology* **49**: 195-201.
- Loeys, B., Van Maldergem, L., Mortier, G., Coucke, P., Gerniers, S., Naeyaert, J. M. and De Paepe, A. (2002). Homozygosity for a missense mutation in fibulin-5 (*FBLN5*) results in a severe form of cutis laxa. *Human Molecular Genetics* **11**: 2113-2118.
- Longman, C., Sewry, C. A. and Muntoni, F. (2004). Muscle involvement in the cerebro-oculo-facio-skeletal syndrome. *Pediatric Neurology* **30**: 125-128.
- Longo, N., Langley, S. D., Still, M. J. and Elsas, L. J. (1995). Prenatal analysis of the insulin receptor gene in a family with leprechaunism. *Prenatal Diagnosis* **15**: 1070-1074.
- López, N. J. (1992). Clinical, laboratory, and immunological studies of a family with a high prevalence of generalized prepubertal and juvenile periodontitis. *Journal of Periodontology* **63**: 457-468.
- Lopez-Arlandis, J. M., Vílchez, J. J., Palau, F. and Sevilla, T. (1995). Friedreich's ataxia: an epidemiological study in Valencia, Spain, based on consanguinity analysis. *Neuroepidemiology* **14**: 14-19.
- Lowry, R. B., Machin, G. A., Morgan, K., Mayock, D. and Marx, L. (1985). Congenital contractures, edema, hyperkeratosis, and intrauterine growth retardation: A fatal syndrome in Hutterite and Mennonite kindreds. *American Journal of Medical Genetics* **22**: 531-543.
- Lowry, R. B., Morgan, K., Holmes, T. M. and Gilroy, S. W. M. (1985). Congenital anomalies in the Hutterite population: A preliminary survey and hypothesis. *American Journal of Medical Genetics* **22**: 545-552.
- Lucien, N., Celton, J. L., Le Pennec, P. Y., Cartron, J. P. and Bailly, P. (2002). Short deletion within the blood group Dombrock locus causing a Do(null) phenotype. *Blood* **100**: 1063-1064.
- Lucotte, G., Barre, E. and Berriche, S. (1991). Frequency of the cystic fibrosis delta-F508 in Algeria. *Human Genetics* **87**: 759.
- Ludwig, M., Beck, A., Wickert, L., Bolkenius, U., Tittel, B., Hinkel, K. and Bidlingmaier, F. (1998). Female pseudohermaphroditism associated with a novel homozygous G-to-A (V370-to-M) substitution in the P-450 aromatase gene. *Journal of Pediatric Endocrinology and Metabolism* **11**: 657-664.
- Maatouk, F., Laamiri, D., Argoubi, K. and Ghedira, H. (1995). Dental manifestations of inbreeding. *The Journal of Clinical Pediatric Dentistry* **19**: 305-306.

- Macari, F., Lautier, C., Girardet, A., Dadoun, P., Dutour, A., Renard, E., Bouvagnet, P., Claustres, M., *et al.* (1998). Refinement of genetic localization of the Alstrom syndrome on chromosome 2p12-13 by linkage analysis in a North African family. *Human Genetics* **103**: 658-661.
- Maccluer, J. W. (1980). Inbreeding and human fetal death. In, *Human Embryonic and Fetal Death*. (Academic Press, New York). 241-260.
- Madani, K., Otoukesh, H., Rastegar, A. and Van Why, S. (2001). Chronic renal failure in Iranian children. *Pediatric Nephrology* **16**: 140-144.
- Magen, D., Adler, L., Mandel, H., Efrati, E. and Zelikovic, I. (2004). Autosomal recessive renal proximal tubulopathy and hypercalciuria: a new syndrome. *American Journal of Kidney Disease* **43**: 600-606.
- Magnus, P., Berg, K. and Bjerkedal, T. (1985). Association of parental consanguinity with decreased birth weight and increased rate of early death and congenital malformations. *Clinical Genetics* **28**: 335-342.
- Mahdi, A. H. (1991). Genetically determined neurodegenerative disorders: experiences in Saudi Arabia. *Annals of Tropical Paediatrics* **11**: 17-23.
- Maheshwari, H. G., Silverman, B. L., Dupuis, J. and Baumann, G. (1998). Phenotype and genetic analysis of a syndrome caused by an inactivating mutation in the growth hormone-releasing hormone receptor: Dwarfism of Sindh. *Journal of Clinical Endocrinology and Metabolism* **83**: 4065-4074.
- Mahjneh, I., Bushby, K., Anderson, L., Muntoni, F., Tolvanen-Mahjneh, H. and Bashir, R. (1999). Merosin-positive congenital muscular dystrophy: a large inbred family. *Neuropediatrics* **30**: 22-28.
- Mailänder, V., Heine, R., Deymeer, F. and Lehmann-Horn, F. (1996). Novel muscle chloride channel mutations and their effects on heterozygous carriers. *American Journal of Human Genetics* **58**: 317-324.
- Majeed, H. A., Al-Tarawna, M., El-Shanti, H., Kamel, B. and Al-Khalaileh, F. (2001). The syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anaemia. Report of a new family and a review. *European Journal of Pediatrics* **160**: 705-710.
- Mamedova, R. A., Moshkina, I. S., Lozlova, S. I., Galkina, V. A., Rudenskaia, G. E., Khlebnikova, O. V., Starkov, I. V., Zhigacheva, A. V., *et al.* (1997). Medico-genetic study of residents of Marii El republic: burden of hereditary diseases in four regions of the republic. *Genetika* **33**: 1697-1702 [In Russian].
- Mancini, G. M., Stojanov, L., Willemsen, R., Kleijer, W. J., Huijmans, J. G., van Diggelen, O. P., de Klerk, J. B., Vuzevski, V. D., *et al.* (1999). Juvenile hyaline fibromatosis: clinical heterogeneity in three patients. *Dermatology* **198**: 18-25.

- Mancuso, M., Filosto, M., Tsujino, S., Lamperti, C., Shanske, S., Coquet, M., Desnuelle, C. and DiMauro, S. (2003). Muscle glycogenesis and mitochondrial hepatopathy in an infant with mutations in both the myophosphorylase and deoxyguanosine kinase genes. *Archives of Neurology* **60**: 1445-1447.
- Manzur, A. Y., Sewry, C. A., Ziprin, J., Dubowitz, V. and Muntoni, F. (1998). A severe clinical and pathological variant of central core disease with possible autosomal recessive inheritance. *Neuromuscular Disorders* **8**: 467-473.
- Marcus, P. and Verp, M. S. (1993). Menetrier disease in a child of a consanguineous union. *American Journal of Medical Genetics* **47**: 1231-1232.
- Margaglione, M., Santacroce, R., Colaizzo, D., Seripa, D., Vecchione, G., Lupone, M. R., De Lucia, D., Fortina, P., *et al.* (2000). A G-to-A mutation in IVS-3 of the human gamma fibrinogen gene causing afibrinogenemia due to abnormal RNA splicing. *Blood* **96**: 2501-2505.
- Marsden, D., Nyhan, W. L. and Sakati, N. O. (1994). Syndrome of hypoparathyroidism, growth hormone deficiency, and multiple minor anomalies. *American Journal of Medical Genetics* **52**: 334-338.
- Martignetti, J. A., Aqeel, A. A., Sewairi, W. A., Boumah, C. E., Kambouris, M., Mayouf, S. A., Sheth, K. V., Eid, W. A., *et al.* (2001). Mutation of the matrix metalloproteinase 2 gene (MMP2) causes a multicentric osteolysis and arthritis syndrome. *Nature Genetics* **28**: 261-265.
- Martin, S. N., Sutherland, J., Levin, A. V., Klose, R., Priston, M. and Heon, E. (2000). Molecular characterisation of congenital glaucoma in a consanguineous Canadian community: a step towards preventing glaucoma related blindness. *Journal of Medical Genetics* **37**: 422-427.
- Martinez-Frias, M. L. (1998). Analysis of the risk of congenital defects in different ethnic groups in Spain. *Anales Español Pediatrica* **48**: 395-400 [In Spanish].
- Martinez-Frias, M. L. and Bermejo, E. (1992). Prevalence of congenital anomaly syndromes in a Spanish Gypsy population. *Journal of Medical Genetics* **29**: 483-486.
- Martinez-Frias, M. L., Bermejo, E., Rodriguez-Pinilla, E. and Frias, J. L. (1999). Maternal and fetal factors related to abnormal amniotic fluid. *Journal of Perinatology* **19**: 514-520.
- Martinez-Frias, M. L., Castilla, E. E., Bermejo, E., Prieto, L. and Orioli, I. M. (2000). Isolated small intestinal atresias in Latin America and Spain: epidemiological analysis. *American Journal of Medical Genetics* **93**: 355-359.
- Martinez-Frias, M. L., Frias, J. L., Rodriguez-Pinilla, E., Urioste, M., Bermejo, E. and Cereijo, A. (1991). Value of clinical analysis in epidemiological research: the Spanish registry experience. *American Journal of Medical Genetics* **41**: 192-195.

- Mashima, Y., Konishi, M., Nakamura, Y., Imamura, Y., Yamada, M., Ogata, T., Kudoh, J. and Shimizu, N. (1998). Severe form of juvenile corneal stromal dystrophy with homozygous R124H mutation in the keratoepithelin gene in five Japanese patients. *British Journal of Ophthalmology* **82**: 1280-1284.
- Masmoudi, S., Antonarakis, S. E., Schwede, T., Ghorbel, A. M., Gratri, M., Pappasavas, M. P., Drira, M., Elgaied-Boulila, A., *et al.* (2001). Novel missense mutations of Tmprss3 in two consanguineous Tunisian families with non-syndromic autosomal recessive deafness. *Human Mutation* **18**: 101-108.
- Masmoudi, S., Charfedine, I., Hmani, M., Grati, M., Ghorbel, A. M., Elgaied-Boulila, A., Drira, M., Hardelin, J. P., *et al.* (2000). Pendred syndrome: phenotypic variability in two families carrying the same PDS missense mutation. *American Journal of Medical Genetics* **90**: 38-44.
- Masmoudi, S., Tlili, A., Majava, M., Ghorbel, A. M., Chardenoux, S., Lemainque, A., Zina, Z. B., Moala, J., *et al.* (2003). Mapping of a new autosomal recessive nonsyndromic hearing loss locus (DFNB32) to chromosome 1p13.3-22.1. *European Journal of Human Genetics* **11**: 185-188.
- Matthiesen, G., Pedersen, V. F., Helin, P., Jacobsen, G. K. and Nielsen, N. S. (2001). Winchester syndrome. *International Orthopaedics* **25**: 331-333.
- Mazarib, A., Xiong, L., Neufeld, M. Y., Birnbaum, M., Korczyn, A. D., Pandolfo, M. and Berkovic, S. F. (2001). Unverricht-Lundborg disease in a five-generation Arab family: instability of dodecamer repeats. *Neurology* **57**: 1050-1054.
- Mazigh M'Rad, S., Sghaier, K., Boukthir, S., Barbouch, R. and Barsaoui, S. (2000). [Chronic septic granulomatosis in a young girl manifesting as invasive pulmonary aspergillosis and flaccid paraplegia]. *La Tunisie Medicale* **78**: 210-215 [In French].
- McConville, C. M., Byrd, P. J., Ambrose, P. J. and Taylor, A. M. R. (1994). Genetic and physical mapping of the ataxia-telangiectasia locus on chromosome 11q22-q23. *International Journal of Radiation Biology* **66**: S45-S56.
- McEntagart, M., Kamel, H., Lebon, P. and King, M. D. (1998). Aicardi-Goutières syndrome: an expanding phenotype. *Neuropediatrics* **29**: 163-167.
- McHale, D. P., Jackson, A. P., Campbell, D. A., Levene, M. I., Corry, P., Woods, C. G., Lench, N. J., Mueller, R. F., *et al.* (2000). A gene for ataxic cerebral palsy maps to chromosome 9p12-q12. *European Journal of Human Genetics* **8**: 267-272.
- McHale, D. P., Mitchell, S., Bunday, S., Moynihan, L., Campbell, D. A., Woods, C. G., Lench, N. J., Mueller, R. F., *et al.* (1999). A gene for autosomal recessive symmetrical spastic cerebral palsy maps to chromosome 2q24-25. *American Journal of Human Genetics* **64**: 526-532.

- McKee, S. A., Barnicoat, A., Fryer, A., Flinter, F., McCormick, D. and McKeown, C. (2001). Joint and skin laxity with Dandy-Walker malformation and contractures: a distinct recessive syndrome? *Clinical Dysmorphology* **10**: 177-180.
- McKusick, V. A. (1980). Medical genetic studies of the Amish, with comparison to other populations, In *Population Structure and Genetic Disorders* (eds. Eriksson, A. W.) 291-300 (Academic Press, London).
- McLean, W. H., Irvine, A. D., Hamill, K. J., Whittock, N. V., Coleman-Campbell, C. M., Mellerio, J. E., Ashton, G. S., Dopping-Hepenstal, P. J., *et al.* (2003). An unusual N-terminal deletion of the laminin alpha3a isoform leads to the chronic granulation tissue disorder laryngo-onycho-cutaneous syndrome. *Human Molecular Genetics* **12**: 2395-2409.
- McMahon, C., Will, A., Hu, P., Shah, G. N., Sly, W. S. and Smith, O. P. (2001). Bone marrow transplantation corrects osteopetrosis in the carbonic anhydrase II deficiency syndrome. *Blood* **97**: 1947-1950.
- Megarbane, A. (2003). Unknown diagnosis in two male cousins with facial abnormalities, optic atrophy, abnormal EEG, and severe psychomotor retardation. *American Journal of Medical Genetics* **116A**: 381-384.
- Megarbane, A., Choueiri, R., Bleik, J., Mezzina, M. and Caillaud, C. (1999). Microcephaly, microphthalmia, congenital cataract, optic atrophy, short stature, hypotonia, severe psychomotor retardation, and cerebral malformations: a second family with micro syndrome or a new syndrome? *Journal of Medical Genetics* **36**: 637-640.
- Megarbane, A., Desguerres, I., Rizkallah, E., Delague, V., Nabbout, R., Barois, A. and Urtizbereia, A. (2000). Brown-Vialetto-Van Laere syndrome in a large inbred Lebanese family: confirmation of autosomal recessive inheritance? *American Journal of Medical Genetics* **92**: 117-121.
- Megarbane, A., Haddad-Zebouni, S., Nabbout, R., Khoury, A. H. and Traboulsi, E. I. (1999). Microcephaly, colobomatous microphthalmia, short stature, and severe psychomotor retardation in two male cousins: a new MCA/MR syndrome? *American Journal of Medical Genetics* **83**: 82-87.
- Megarbane, A., Khalil, G., Waked, N., Rotig, A., Caillaud, C. and Loiselet, J. (1999). Two sibs with myoclonic epilepsy, congenital deafness, macular dystrophy, and psychiatric disorders. *American Journal of Medical Genetics* **87**: 289-293.
- Megarbane, A., Kharrat, K. and Kreichati, G. (1998). Four sibs with dislocated elbows, bowed tibiae, scoliosis, deafness, cataract, microcephaly, and mental retardation: a new MCA/MR syndrome. *Journal of Medical Genetics* **35**: 755-758.
- Megarbane, A., Mustapha, M., Bleik, J., Waked, N., Delague, V. and Loiselet, J. (2000). Exclusion of chromosome 15q21.1 in autosomal-recessive Weill-

- Marchesani syndrome in an inbred Lebanese family. *Clinical Genetics* **58**: 473-478.
- Megarbane, A., Noujeim, Z., Fabre, M. and Der Kaloustian, V. (1998). New form of hidrotic ectodermal dysplasia in a Lebanese family. *American Journal of Medical Genetics* **75**: 196-199.
- Megarbane, A., Ruchoux, M. M., Loeys, B., Ayoub, N. and Nuytinck, L. (2001). Short stature, abnormal face, joint laxity, dislocation, hernias, delayed bone age, and severe psychomotor retardation in two brothers: previously undescribed MCA/MR syndrome. *American Journal of Medical Genetics* **104**: 221-224.
- Meggouh, F., Benomar, A., Rouger, H., Tardieu, S., Birouk, N., Tassin, J., Barhoumi, C., Yahyaoui, M., *et al.* (1998). The first de novo mutation of the connexin 32 gene associated with X-linked Charcot-Marie-Tooth disease. *Journal of Medical Genetics* **35**: 251-252.
- Meguid, N. A., Temtamy, S. A. and Mazen, I. (2003). Transposition of external genitalia and associated malformations. *Clinical Dysmorphology* **12**: 59-62.
- Mehta, L., Trounce, J. Q., Moore, J. R. and Young, I. D. (1986). Familial calcification of the basal ganglia with cerebrospinal fluid pleocytosis. *Journal of Medical Genetics* **23**: 157-160.
- Melkonieni, M., Brunner, H. G., Manouvrier, S., Hennekam, R., Superti-Furga, A., Kaariainen, H., Pauli, R. M., van Essen, T., *et al.* (2000). Autosomal recessive disorder otospondylomegaepiphyseal dysplasia is associated with loss-of-function mutations in the COL11A2 gene. *American Journal of Human Genetics* **66**: 368-377.
- Mendonca, B. B., Leite, M. V., de Castro, M., Kino, T., Elias, L. L., Bachega, T. A., Arnhold, I. J., Chrousos, G. P., *et al.* (2002). Female pseudohermaphroditism caused by a novel homozygous missense mutation of the GR gene. *Journal of Clinical Endocrinology and Metabolism* **87**: 1805-1809.
- Merin, S., Lapithis, A. G., Horovitz, D. and Michaelson, I. C. (1972). Childhood blindness in Cyprus. *American Journal of Ophthalmology* **74**: 538-542.
- Michaelides, M., Aligianis, I. A., Ainsworth, J. R., Good, P., Mollon, J. D., Maher, E. R., Moore, A. T. and Hunt, D. M. (2004). Progressive cone dystrophy associated with mutation in CNGB3. *Investigative Ophthalmology and Visual Science* **45**: 1975-1982.
- Middleton, L., Ohno, K., Christodoulou, K., Brengman, J., Milone, M., Neocleous, V., Serdaroglu, P., Deymeer, F., *et al.* (1999). Chromosome 17p-linked myasthenias stem from defects in the acetylcholine receptor epsilon-subunit gene. *Neurology* **53**: 1076-1082.

- Mikaeloff, Y., Pinton, F., Sevin, C., Dhondt, J. L. and Ponsot, G. (1999). [Progressive convulsive encephalopathy: considering a abnormality of biopterin metabolism]. *Archives de Pediatrie* **6**: 759-761 [In French].
- Miladi, N., Larnaout, A., Dhondt, J. L., Vincent, M. F., Kaabachi, N. and Hentati, F. (1998). Dihydropteridine reductase deficiency in a large consanguineous Tunisian family: clinical, biochemical, and neuropathologic findings. *Journal of Child Neurology* **13**: 475-480.
- Milan, M., Astolfi, G., Volpato, S., Garani, G. P., Clementi, M. and Tenconi, R. (1994). 766 cases of oral cleft in Italy. *European Journal of Epidemiology* **10**: 317-324.
- Milford, D. V., Shackleton, H. L. and Stewart, P. M. (1994). Mineralocorticoid hypertension and congenital deficiency of 11 β -hydroxysteroid dehydrogenase in a family with the syndrome of 'apparent' mineralocorticoid excess. *Clinical Endocrinology* **43**: 241-246.
- Millar, D. S., Allgrove, J., Rodeck, C., Kakkar, V. V. and Cooper, D. N. (1994). A homozygous deletion/insertion mutation in the protein C (PROC) gene causing neonatal *Purpura fulminans*: prenatal diagnosis in an at-risk pregnancy. *Blood Coagulation and Fibrinolysis* **5**: 647-649.
- Millat, G., Marcais, C., Tomasetto, C., Chikh, K., Fensom, A. H., Harzer, K., Wenger, D. A., Ohno, K., *et al.* (2001). Niemann-Pick C1 disease: correlations between NPC1 mutations, levels of NPC1 protein, and phenotypes emphasize the functional significance of the putative sterol-sensing domain and of the cysteine-rich luminal loop. *American Journal of Human Genetics* **68**: 1373-1385.
- Mirghomizadeh, F., Pfister, M., Apaydin, F., Petit, C., Kupka, S., Pusch, C. M., Zenner, H. P. and Blin, N. (2002). Substitutions in the conserved C2C domain of otoferlin cause DFNB9, a form of nonsyndromic autosomal recessive deafness. *Neurobiology of Disease* **10**: 157-164.
- Mitchell, S. J., Cole, T. and Redford, D. H. A. (1997). Congenital diaphragmatic hernia with probable autosomal recessive inheritance in an extended consanguineous Pakistani kindred. *Journal of Medical Genetics* **34**: 601-603.
- Mitsubuchi, H., Matsuda, I., Wobukuni, Y., Heidemreich, Indo, Y., Endo, F. *et al.* (1992). Gene analysis of Mennonite maple syrup urine disease kindred using primer-specified restriction map modification. *Journal of Inherited Metabolic Disease* **15**: 181-187.
- Modilevsky, T., Manor, E. and Marmor, A. (1978). Alkaptonuria: prevalence among Bedouins related by consanguinity. *Monographs of Human Genetics* **9**: 123-125.
- Moghadaszadeh, B., Desguerre, I., Topaloglu, H., Muntoni, F., Pavek, S., Sewry, C., Mayer, M., Fardeau, M., *et al.* (1998). Identification of a new locus for a peculiar form of congenital muscular dystrophy with early rigidity of the spine, on chromosome 1p35-36. *American Journal of Human Genetics* **62**: 1439-1445.

- Moghadaszadeh, B., Topaloglu, H., Merlini, L., Muntoni, F., Estournet, B., Sewry, C., Naom, I., Barois, A., *et al.* (1999). Genetic heterogeneity of congenital muscular dystrophy with rigid spine syndrome. *Neuromuscular Disorders* **9**: 376-382.
- Mohamed, M. D., Topping, N. C., Jafri, H., Raashed, Y., McKibbin, M. A. and Inglehearn, C. F. (2003). Progression of phenotype in Leber's congenital amaurosis with a mutation at the LCA5 locus. *British Journal of Ophthalmology* **87**: 473-475.
- Mohan, N. and Sarkar, R. (1994). Hematological status of β -Thalasseemics in Madras. *Indian Journal of Pediatrics* **61**: 237-248.
- Mokhtar, M. M., Kotb, S. M. and Ismail, S. R. (1998). Autosomal recessive disorders among patients attending the genetics clinic in Alexandria. *Eastern Mediterranean Health Journal* **4**: 470-479.
- Molven, A., Matre, G. E., Duran, M., Wanders, R. J., Rishaug, U., Njolstad, P. R., Jellum, E. and Sovik, O. (2004). Familial hyperinsulinemic hypoglycemia caused by a defect in the SCHAD enzyme of mitochondrial fatty acid oxidation. *Diabetes* **53**: 221-227.
- Molven, A., Rishaug, U., Matre, G. E., Njolstad, P. R. and Sovik, O. (2002). Hunting for a hypoglycemia gene: severe neonatal hypoglycemia in a consanguineous family. *American Journal of Medical Genetics* **113**: 40-46.
- Monnier, N., Gout, J. P., Pin, I., Gauthier, G. and Lunardi, J. (2001). A novel 3600+11.5 kb C>G homozygous splicing mutation in a black African, consanguineous CF family. *Journal of Medical Genetics* **38**: E4.
- Montague, C. T., Farooqi, I. S., Whitehead, J. P., Soos, M. A., Rau, H., Wareham, N. J., Sewter, C. P., Digby, J. E., *et al.* (1997). Congenital leptin deficiency is associated with severe early-onset obesity in humans. *Nature* **387**: 903-908.
- Moog, U., Bleeker-Wagemakers, E. M., Crobach, P., Vles, J. S. and Schrandt-Stumpel, C. T. (1998). Sibs with Axenfeld-Rieger anomaly, hydrocephalus, and leptomeningeal calcifications: a new autosomal recessive syndrome? *American Journal of Medical Genetics* **78**: 263-266.
- Moreno Fuenmayor, H. (1980). The spectrum of frontonasal dysplasia in an inbred pedigree. *Clinical Genetics* **17**: 137-142.
- Morgan, N. V., Bacchelli, C., Gissen, P., Morton, J., Ferrero, G. B., Silengo, M., Labrune, P., Casteels, I., *et al.* (2003). A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. *Journal of Medical Genetics* **40**: 431-435.
- Morgan, N. V., Gissen, P., Sharif, S. M., Baumber, L., Sutherland, J., Kelly, D. A., Aminu, K., Bennett, C. P., *et al.* (2002). A novel locus for Meckel-Gruber syndrome, MKS3, maps to chromosome 8q24. *Human Genetics* **111**: 456-461.

- Morton, J., Kilby, M. and Ruston, I. (1998). A new lethal autosomal recessive skeletal dysplasia with associated dysmorphic features. *Clinical Dysmorphology* **7**: 109-114.
- Morton, N. E. (1991). Genetic epidemiology of hearing impairment. *Annals of New York Academy of Sciences* **630**: 16-31.
- Mota, C. R., Azevedo, M., Rocha, G., Manuela, F., Coelho, R. and Lima, M. R. (2000). Axial mesodermal dysplasia complex: a new case with parental consanguinity. *Clinical Dysmorphology* **9**: 73-75.
- Motohashi, N., Shinoroha, M., Shioe, K., Fukuzawa, H., Akiyama, Y. and Kariya, T. (1995). A case of membranous lipodystrophy (Nasu-Hakola disease) with unique MRI findings. *Neuroradiology* **37**: 549-550.
- Motulsky, A. G. (1995). Jewish diseases and origins. *Nature Genetics* **9**: 99-101.
- Moynihan, L., Houseman, M., Newton, V., Mueller, R. and Lench, N. (1999). DFNB20: a novel locus for autosomal recessive, non-syndromal sensorineural hearing loss maps to chromosome 11q25-qter. *European Journal of Human Genetics* **7**: 243-246.
- Moynihan, L. M., Bunday, S. E., Heath, D., Jones, E. L., McHale, D. P., Mueller, R. F., Markham, A. F. and Lench, N. J. (1998). Autozygosity mapping, to chromosome 11q25, of a rare autosomal recessive syndrome causing histiocytosis, joint contractures, and sensorineural deafness. *American Journal of Human Genetics* **62**: 1123-1128.
- Mukhtarov, Z., Alieva, K. A. and Rasulov Ie, M. (2000). [Population genetics research in the Lerik region of Azerbaijan]. *Tsitologiya I Genetika* **34**: 22-29 [In Russian].
- Muller, T., Schafer, H., Rodeck, B., Haupt, G., Koch, H., Bosse, H., Welling, P., Lange, H., *et al.* (1999). Familial clustering of infantile cirrhosis in Northern Germany: A clue to the etiology of idiopathic copper toxicosis. *Journal of Pediatrics* **135**: 189-196.
- Muller, T., Wijmenga, C., Phillips, A. D., Janecke, A., Houwen, R. H., Fischer, H., Ellemunter, H., Fruhwirth, M., *et al.* (2000). Congenital sodium diarrhea is an autosomal recessive disorder of sodium/proton exchange but unrelated to known candidate genes. *Gastroenterology* **119**: 1506-1513.
- Mullis, P. E., Deladoey, J. and Dannies, P. S. (2002). Molecular and cellular basis of isolated dominant-negative growth hormone deficiency, IGHD type II: insights on the secretory pathway of peptide hormones. *Hormone Research* **58**: 53-66.
- Mullis, P. E., Deladoey, J. and Dannies, P. S. (2002). New GH-1 gene mutations: expanding the spectrum of causes of isolated growth hormone deficiency. *Journal of Pediatric Endocrinology and Metabism* **15**: 1301-1310.

- Mungan, Z., Akyuz, F., Bugra, Z., Yonall, O., Ozturk, S., Acar, A. and Cevikbas, U. (2003). Familial visceral myopathy with pseudo-obstruction, megaduodenum, Barrett's esophagus, and cardiac abnormalities. *American Journal of Gastroenterology* **98**: 2556-2560.
- Munhoz, R. P., Sa, D. S., Rogaeva, E., Salehi-Rad, S., Sato, C., Medeiros, H., Farrer, M. and Lang, A. E. (2004). Clinical findings in a large family with a parkin ex3delta40 mutation. *Archives of Neurology* **61**: 701-704.
- Muramatsu, T., Sakai, N., Yanagihara, L., Yamada, M., Nishigaki, T., Kokubu, C., Tsukamoto, H., Ito, M., *et al.* (2002). Mutation analysis of the acid ceramidase gene in Japanese patients with Farber disease. *Journal of Inherited Metabolic Disease* **25**: 585-592.
- Murphy, M., McHugh, B., Tighe, D., Mayne, P., O'Neill, C., Naughten, E. and Croke, D. T. (1999). Genetic basis of transferase-deficient galactosaemia in Ireland and the population history of the Irish Travellers. *European Journal of Human Genetics* **7**: 549-554.
- Murshid, W. R. (2000). Spina bifida in Saudi Arabia: is consanguinity among the parents a risk factor? *Pediatric Neurosurgery* **32**: 10-12.
- Murshid, W. R., Jarallah, J. S. and Dad, M. I. (2000). Epidemiology of infantile hydrocephalus in Saudi Arabia: Birth prevalence and associated factors. *Pediatric Neurosurgery* **32**: 119-123.
- Mustapha, M., Azar, S. T., Moglabey, Y. B., Saouda, M., Zeitoun, G., Loiselet, J. and Slim, R. (1998). Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425. *Journal of Medical Genetics* **35**: 202-204.
- Mustapha, M., Chardenoux, S., Nieder, A., Salem, N., Weissenbach, J., el-Zir, E., Loiselet, J. and Petit, C. (1998). A sensorineural progressive autosomal recessive form of isolated deafness, DFNB13, maps to chromosome 7q34-q36. *European Journal of Human Genetics* **6**: 245-250.
- Mustapha, M., Chouery, E., Chardenoux, S., Naboulsi, M., Paronnaud, J., Lemainque, A., Megarbane, A., Loiselet, J., *et al.* (2002). DFNB31, a recessive form of sensorineural hearing loss, maps to chromosome 9q32-34. *European Journal of Human Genetics* **10**: 210-212.
- Mustapha, M., Chouery, E., Torchard-Pagnez, D., Nouaille, S., Khrais, A., Sayegh, F. N., Megarbane, A., Loiselet, J., *et al.* (2002). A novel locus for Usher syndrome type I, USH1G, maps to chromosome 17q24-25. *Human Genetics* **110**: 348-350.
- Mustapha, M., Chouery, E., Torchard-Pagnez, D., Nouaille, S., Khrais, A., Sayegh, F. N., Megarbane, A., Loiselet, J., *et al.* (2002). A novel locus for Usher syndrome type I, USH1G, maps to chromosome 17q24-25. *Human Genetics* **110**: 348-350.

- Mustapha, M., Salem, N., Weil, D., el-Zir, E., Loiselet, J. and Petit, C. (1998). Identification of a locus on chromosome 7q31, DFNB14, responsible for prelingual sensorineural non-syndromic deafness. *European Journal of Human Genetics* **6**: 548-551.
- Mustapha, M., Weil, D., Chardenoux, S., Elias, S., El-Zir, E., Beckmann, J. S., Loiselet, J. and Petit, C. (1999). An alpha-tectorin gene defect causes a newly identified autosomal recessive form of sensorineural pre-lingual non-syndromic deafness, DFNB21. *Human Molecular Genetics* **8**: 409-412.
- Nachlieli, T. and Gershoni-Baruch, R. (1992). Dextrocardia, microphthalmia, cleft palate, chorelathetosis, and mental retardation in an infant born to consanguineous parents. *American Journal of Medical Genetics* **42**: 458-460.
- Naderi, S. (1979). Congenital abnormalities in newborns of consanguineous and nonconsanguineous parents. *Obstetrics and Gynecology* **53**: 195-199.
- Nagae-Poetscher, L. M., Bibat, G., Philippart, M., Rosemberg, S., Fatemi, A., Lacerda, M. T., Costa, M. O., Kok, F., *et al.* (2004). Leukoencephalopathy, cerebral calcifications, and cysts: new observations. *Neurology* **62**: 1206-1209.
- Nagai, K., Nagao, M., Yanai, S., Minagawa, K., Takahashi, Y., Takekoshi, Y., Ishizaka, A., Matsuzono, Y., *et al.* (1998). Oral-facial-digital syndrome type IX in a patient with Dandy-Walker malformation. *Journal of Medical Genetics* **35**: 342-344.
- Nagayama, Y., Kobayashi, N. and Yamabe, H. (1992). Obstructive jaundice caused by polypoid inflammatory glaucoma in a consanguineous case. *Digestive Diseases and Science* **37**: 613-617.
- Nair, S., Ghosh, K., Kulkarni, B., Shetty, S. and Mohanty, D. (2002). Glanzmann's thrombasthenia: updated. *Platelets* **13**: 387-393.
- Najmabadi, H., Neishabury, M., Sahebjam, F., Kahrizi, K., Shafaghati, Y., Nikzat, N., Jalalvand, M., Aminy, F., *et al.* (2003). The Iranian Human Mutation Gene Bank: a data and sample resource for worldwide collaborative genetics research. *Human Mutation* **21**: 146-150.
- Nakagawa, M., Matsuzaki, T., Suehara, M., Kanzato, N., Takashima, H., Higuchi, I., Matsumura, T., Goto, K., *et al.* (2001). Phenotypic variation in a large Japanese family with Miyoshi myopathy with nonsense mutation in exon 19 of dysferlin gene. *Journal of the Neurological Sciences* **184**: 15-19.
- Nakagawa, T., Hashi, K., Kurokawa, Y. and Yamamura, A. (1999). Family history of subarachnoid hemorrhage and the incidence of asymptomatic, unruptured cerebral aneurysms. *Journal of Neurosurgery* **91**: 391-395.
- Nakano, A., Lestringant, G. G., Paperna, T., Bergman, R., Gershoni, R., Frossard, P., Kanaan, M., Meneguzzi, G., *et al.* (2002). Junctional epidermolysis bullosa in the

- Middle East: clinical and genetic studies in a series of consanguineous families. *Journal of the American Academy of Dermatology* **46**: 510-516.
- Nakura, J., Miki, T., Ye, L., Mitsuda, N., Zhao, Y., Kihara, K., Yu, C. E., Oshima, J., *et al.* (1996). Narrowing the position of the Werner syndrome locus by homozygosity analysis-extension of homozygosity analysis. *Genomics* **36**: 130-141.
- Naom, I. S., D'Alessandro, M., Topaloglu, H., Sewry, C., Ferlini, A., Helbling-Leclerc, A., Guicheney, P., Weissenbach, J., *et al.* (1997). Refinement of the laminin a2 chain locus to human chromosome 6q2 in severe and mild merosin deficient congenital muscular dystrophy. *Journal of Medical Genetics* **34**: 99-104.
- Naveh, Y., Bassan, L., Rosenthal, E., Berkowitz, D., Jaffe, M., Mandel, H. and Berant, M. (1997). Progressive familial intrahepatic cholestasis among the Arab population in Israel. *Journal of Pediatric Gastroenterology and Nutrition* **24**: 548-554.
- Naz, S., Giguere, C. M., Kohrman, D. C., Mitchem, K. L., Riazuddin, S., Morell, R. J., Ramesh, A., Srisailpathy, S., *et al.* (2002). Mutations in a novel gene, TMIE, are associated with hearing loss linked to the DFNB6 locus. *American Journal of Human Genetics* **71**: 632-636.
- Nazer, H., Al-Mehaidib, A., Shabib, S. and Ali, M. A. (1998). Crigler-Najjar syndrome in Saudi Arabia. *American Journal of Medical Genetics* **79**: 12-15.
- Nazer, H. and Rahbeeni, Z. (1994). Cystic fibrosis and the liver - a Saudi experience. *Annals of Tropical Paediatrics* **14**: 189-194.
- Ndiaye, O., Sall, G., Sylla, A., Diouf, S., Diagne, I. and Kuakuvi, N. (2002). [Progressive spinal amyotrophy type I or Werdnig-Hoffman disease. Apropos of 5 cases in Dakar (Senegal)] *Bulletin de la Societe de Pathologie Exotique* **95**: 81-82 [In French].
- Neel, J. V. (1958). A study of major congenital defects in Japanese infants. *American Journal of Human Genetics* **10**: 398-445.
- Neel, J. V. and Schull, W. J. (1962). The effect of inbreeding on mortality and morbidity in two Japanese cities. *Proceedings of the National Academy of Sciences, USA* **48**: 573-582.
- Nelis, E., Erdem, S., Tan, E., Lofgren, A., Ceuterick, C., De Jonghe, P., Van Broeckhoven, C., Timmerman, V., *et al.* (2002). A novel homozygous missense mutation in the myotubularin-related protein 2 gene associated with recessive Charcot-Marie-Tooth disease with irregularly folded myelin sheaths. *Neuromuscular Disorders* **12**: 869-873.
- Nelson, J., Smith, M. and Bittles, A. H. (1997). Consanguineous marriage and its clinical consequences in migrants to Australia. *Clinical Genetics* **52**: 142-146.

- Netchine, I., Sobrier, M. L., Krude, H., Schnabel, D., Maghnie, M., Marcos, E., Duriez, B., Cacheux, V., *et al.* (2000). Mutations in LHX3 result in a new syndrome revealed by combined pituitary hormone deficiency. *Nature Genetics* **25**: 182-186.
- Neto, R. M., Castilla, E. E. and Paz, J. E. (1981). Hypospadias: an epidemiological study in Latin America. *American Journal of Medical Genetics* **10**: 5-19.
- Neumann, L. M., von Moers, A., Kunze, J., Blankenstein, O. and Marquardt, T. (2003). Congenital disorder of glycosylation type 1a in a macrosomic 16-month-old boy with an atypical phenotype and homozygosity of the N216I mutation. *European Journal of Pediatrics* **162**: 710-713.
- Nevo, Y., Kutai, M., Jossiphov, J., Livne, A., Neeman, Z., Arad, T., Popovitz-Biro, R., Atsmon, J., *et al.* (2004). Childhood macrophagic myofasciitis-consanguinity and clinicopathological features. *Neuromuscular Disorders* **14**: 246-252.
- Newman, P. K. and Saunders, M. (1985). Observations on Indian neurology. *Journal of the Royal College of Physicians of London* **19**: 13-16.
- Neyroud, N., Denjoy, I., Donger, C., Gary, F., Villain, E., Leenhardt, A., Benali, K., Schwartz, K., *et al.* (1998). Heterozygous mutation in the pore of potassium channel gene KvLQT1 causes an apparently normal phenotype in long QT syndrome. *European Journal of Human Genetics* **6**: 129-133.
- Nezarati, M. M., Loeffler, J., Yoon, G., MacLaren, L., Fung, E., Snyder, F., Utermann, G. and Graham, G. E. (2002). Novel mutation in the Delta-sterol reductase gene in three Lebanese sibs with Smith-Lemli-Opitz (RSH) syndrome. *American Journal of Medical Genetics* **110**: 103-108.
- Nichols, P., Croxson, R., Vincent, A., Rutter, R., Hutchinson, M., Newsom-Davis, J. and Beeson, D. (1999). Mutation of the acetylcholine receptor epsilon-subunit promoter in congenital myasthenic syndrome. *Annals of Neurology* **45**: 439-443.
- Nicholson, J. C., Jones, C. L., Powell, H. R., Walker, R. G. and McCredie, D. A. (1995). Familial hypomagnesaemia - hypercalcuria leading to end-stage renal failure. *Pediatric Nephrology* **9**: 74-76.
- Niepomniszcze, H., Sala, M., Danilowicz, K., Pitoia, F. and Bruno, O. (2004). Epidemiology of palpable goiter in greater Buenos Aires in an iodine-sufficient area. *Medicina (B Aires)* **64**: 7-12.
- Nikkels, P. G., Stigter, R. H., Knol, I. E. and van der Harten, H. J. (2001). Schneckenbecken dysplasia, radiology, and histology. *Pediatric Radiology* **31**: 27-30.
- Nirmala, A., Rice, T., Chengal Reddy, P., Sreerama Krishna, K., Venkata Ramana, P. and Rao, D. C. (1992). Comingling and segregation analysis of blood pressure in

- consanguineous and nonconsanguineous families from Andhra Pradesh, India. *American Journal of Human Biology* **4**: 703-716.
- Nishimura, G., Nakayama, M., Fuke, Y. and Suehara, N. (1998). A lethal osteochondroplasia with mesomelic brachymelia, round pelvis, and congenital hepatic fibrosis: two siblings born to consanguineous parents. *Pediatric Radiology* **28**: 43-47.
- Nishio, T., Sunohara, N., Nonaka, I., Tsujino, S. and Sugie, H. (1998). Myophosphorylase deficiency and limb-girdle muscular dystrophy in the same pedigree. *Acta Neurologica Scandinavica* **98**: 364-367.
- Nisipeanu, P., Inzelberg, R., Blumen, S. C., Carasso, R. L., Hattori, N., Matsumine, H. and Mizuno, Y. (1999). Autosomal-recessive juvenile parkinsonism in a Jewish Yemenite kindred: mutation of Parkin gene. *Neurology* **53**: 1602-1604.
- Nogueira, C. R., Leite, C. C., Chedid, E. P., Liberman, B., Pimentel-Filho, F. R., Kopp, P. and Medeiros-Neto, G. A. (1998). Autosomal recessive deficiency of combined pituitary hormones (except ACTH) in a consanguineous Brazilian kindred. *Journal of Endocrinological Investigation* **21**: 386-391.
- Nogueira, C. R., Sabacan, L., Jameson, J. L., Medeiros-Neto, G. and Kopp, P. (1999). Combined pituitary hormone deficiency in an inbred Brazilian kindred associated with a mutation in the PROP-1 gene. *Molecular Genetics and Metabolism* **67**: 58-61.
- Norio, R., Raitta, C. and Lindahl, E. (1984). Further delineation of the Cohen syndrome: report on chorioretinal dystrophy, leucopenia and consanguinity. *Clinical Genetics* **25**: 1-14.
- Novelli, G., Muchir, A., Sangiuolo, F., Helbling-Leclerc, A., D'Apice, M. R., Massart, C., Capon, F., Sbraccia, P., *et al.* (2002). Mandibuloacral dysplasia is caused by a mutation in LMNA-Encoding Lamin A/C. *American Journal of Human Genetics* **71**.
- Nozaki, J., Dakeishi, M., Ohura, T., Inoue, K., Manabe, M., Wada, Y. and Koizumi, A. (2001). Homozygosity mapping to chromosome 5p15 of a gene responsible for Hartnup disorder. *Biochemical and Biophysical Research Communications* **284**: 255-260.
- Nunes, M. L., Mugnol, F., Bica, I. and Fiori, R. M. (2002). Pyridoxine-dependent seizures associated with hypophosphatasia in a newborn. *Journal of Child Neurology* **17**: 222-224.
- Nuzzo, F., Zei, G., Stefanini, M., Colognola, R., Santachiara, A. S., Lagomarsini, P., Marinoni, S. and Salvaneschi, L. (1990). Search for consanguinity within and among families of patients with trichothiodystrophy associated with xeroderma pigmentosum. *Journal of Medical Genetics* **27**: 21-25.