Caksen, H., Odabas, D., Oner, A. F., Abuhandan, M. and Calebi, V. (2002). Ophthalmo-acromelic syndrome in a Turkish infant: case report. *East African Medical Journal* **79**: 339-340.

Callaghan, M., Hand, C. K., Kennedy, S. M., FitzSimon, J. S., Collum, L. M. and Parfrey, N. A. (1999). Homozygosity mapping and linkage analysis demonstrate that autosomal recessive congenital hereditary endothelial dystrophy (CHED) and autosomal dominant CHED are genetically distinct. *British Journal of Ophthalmology* **83**: 115-119.

Camacho Vanegas, O., Bertini, E., Zhang, R. Z., Petrini, S., Minosse, C., Sabatelli, P., Giusti, B., Chu, M. L., *et al.* (2001). Ullrich scleroatonic muscular dystrophy is caused by recessive mutations in collagen type VI. *Proceedings of the National Academy of Sciences of the United States of America* **98**: 7516-7521.

Camargo, R., Limbert, E., Gillam, M., Henriques, M. M., Fernandes, C., Catarino, A. L., Soares, J., Alves, V. A., *et al.* (2001). Aggressive metastatic follicular thyroid carcinoma with anaplastic transformation arising from a long-standing goiter in a patient with Pendred's syndrome. *Thyroid* **11**: 981-998.

Camuzat, A., Dollfus, H., Rozet, J. M., Gerber, S., Bonneau, D., Bonnemaison, M., Braird, M. L., Dufier, J. L., *et al.* (1995). A gene for Leber's congenital amaurosis maps to chromosome 17p. *Human Molecular Genetics* **4**: 1447-1452.

Can, S., Zhu, Y. S., Cai, L. Q., Ling, Q., Katz, M. D., Akgun, S., Shackleton, C. H. and Imperato-McGinley, J. (1998). The identification of 5 alpha-reductase and 17 beta-hydroxysteroid dehydrogenase-3 gene defects in male pseudohermaphrodites from a Turkish kindred. *Journal of Clinical Endocrinology and Metabolism* **83**: 560-569.

Carakushansky, M., Whatmore, A. J., Clayton, P. E., Shalet, S. M., Gleeson, H. K., Price, D. A., Levine, M. A. and Salvatori, R. (2003). A new missense mutation in the growth hormone-releasing hormone receptor gene in familial isolated GH deficiency. *European Journal of Endocrinology* **148**: 25-30.

Cardoso, E., Hawary, M. and Mahmud, S. (1998). A recessively inherited non-lethal form of popliteal pterygium syndrome. *British Journal of Maxillofacial Surgery* **36**: 138-140.

Carew, J. A., Pollak, E. S., High, K. A. and Bauer, K. A. (1998). Severe factor VII deficiency due to a mutation disrupting an Sp1 binding site in the factor VII promoter. *Blood* **92**: 1639-1645.

Carter, C. O. (1977). Monogenic disorders. *Journal of Medical Genetics* **14**: 316-320.

Carvalho, L. R., Woods, K. S., Mendonca, B. B., Marcal, N., Zamparini, A. L., Stifani, S., Brickman, J. M., Arnhold, I. J., *et al.* (2003). A homozygous mutation

in HESX1 is associated with evolving hypopituitarism due to impaired repressor-corepressor interaction. *Journal of Clinical Investigation* **112**: 1192-1201.

Casali, C., Bonifati, V., Santorelli, F. M., Casari, G., Fortini, D., Patrignani, A., Fabbrini, G., Carrozzo, R., *et al.* (2001). Mitochondrial myopathy, parkinsonism, and multiple mtDNA deletions in a Sephardic Jewish family. *Neurology* **56**: 802-805.

Casas, K. A. and Fischel-Ghodsian, N. (2004). Mitochondrial myopathy and sideroblastic anemia. *American Journal of Medical Genetics* **125A**: 201-204.

Castanet, M., Park, S. M., Smith, A., Bost, M., Leger, J., Lyonnet, S., Pelet, A., Czernichow, P., *et al.* (2002). A novel loss-of-function mutation in TTF-2 is associated with congenital hypothyroidism, thyroid agenesis and cleft palate. *Human Molecular Genetics* **11**: 2051-2059.

Castriota-Scanderbeg, A., Zelante, L., Masala, S., Gasparini, P. and Lachman, R. S. (1999). Acrodysplasia, severe ossification abnormalities with short stature, and fibular hypoplasia. *American Journal Medical Genetics* **84**: 68-73.

Castro-Gago, M., Gonzalez-Conde, V., Fernandez-Seara, M. J., Rodrigo-Saez, E., Fernandez-Cebrian, S., Alonso-Martin, A., Campos, Y., Arenas, J., *et al.* (1999). Early mitochondrial encephalomyopathy due to complex IV deficiency consistent with Alpers-Huttenlocher syndrome: report of two cases. *Revista de Neurologia* **29**: 912-917.

Cataltepe, S. and Tunçbilek, E. (1992). A family with one child with acrocallosal syndrome, one child with anencephaly-polydactyly, and parental consanguinity. *European Journal of Pediatrics* **151**: 288-290.

Cavalcanti, D. P. and Salomao, M. A. (1999). Dandy-Walker malformation with postaxial polydactyly: further evidence for autosomal recessive inheritance. *American Journal of Medical Genetics* **85**: 183-184.

Cavalier, L., BenHamida, C., Amouri, R., Belal, S., Bomont, P., Lagarde, N., Gressin, L., Callen, D., *et al.* (2000). Giant axonal neuropathy locus refinement to a < 590 kb critical interval. *European Journal of Human Genetics* **8**: 527-534.

Ceballos-Quintal, J. M., Pinto-Escalante, D. and Gongora-Biachi, R. A. (1992). TAR-like syndrome in a consanguineous Mayan girl. *American Journal of Medical Genetics* **43**: 805-807.

Cendrowski, W. S. (1965). Parental consanguinity in multiple sclerosis. *Journal de Génétique Humaine* **14**: 313-317.

Centerwall, W. R. and Centerwall, S. A. (1966). Consanguinity and congenital anomalies in South India: a pilot study. *Indian Journal of Medical Research* **54**: 1160-1167.

Centerwall, W. R. and Ittyerah, T. R. (1966). Phenylketonuria among the mentally retarded in India. *Lancet* **2**: 193-4.

Chaabouni, M., Ben Slimen, M., Boudawara, M., Ben Amar, H., Mahfoudh, A., Ayadi, F., Ben Halima, N., Hachicha, M., *et al.* (2001). [Mucopolysaccharidoses in children. Experience of a general pediatric service. 11 cases]. *La Tunisie Medicale* **79**: 222-230 [In French].

Chalkley, G. and Harris, A. (1991). A cystic fibrosis patient who is homozygous for the G8SE mutation has very mild disease. *Journal of Medical Genetics* **28**: 875-877.

Chan, I., El-Zurghany, A., Zendah, B., Benghazil, M., Oyama, N., Hamada, T. and McGrath, J. A. (2003). Molecular basis of lipoid proteinosis in a Libyan family. *Clinical Experimental Dermatology* **28**: 545-548.

Chantret, I., Dupre, T., Delenda, C., Bucher, S., Dancourt, J., Barnier, A., Charollais, A., Heron, D., *et al.* (2002). Congenital disorders of glycosylation type Ig is defined by a deficiency in dolichyl-P-mannose: Man7GlcNAc2-PP-dolichyl mannosyltransferase. *Journal of Biological Chemistry* **12:** 25815-25822.

Chapman, S., Gray, R. G. F., Constable, T. J. and Bundey, S. (1989). Atypical radiological features of beta-glucuronidase deficiency (mucopolysaccharidosis VII) occurring in an elderly patient from an inbred kindred. *British Journal of Radiology* **62**: 491-494.

Charlesworth, A., Gagnoux-Palacios, L., Bonduelle, M., Ortonne, J. P., De Raeve, L. and Meneguzzi, G. (2003). Identification of a lethal form of epidermolysis bullosa simplex associated with a homozygous genetic mutation in plectin. *Journal of Investigative Dermatology* **121**: 1344-1348.

Chen, A., Wayne, S., Bell, A., Ramesh, A., Srisailapathy, C. R. S., Scott, D. A., Sheffield, V. C., Van Hauwe, P., *et al.* (1997). New gene for autosomal recessive non-syndromic hearing loss maps to either chromosome 3q or 19p. *American Journal of Medical Genetics* **71**: 467-471.

Chen, K. W., Juang, J. H. and Lin, J. D. (2001). Extreme insulin resistance syndrome. *Chang Gung Medical Journal* **24**: 640-645.

Cheng, C. J. and Stenson, S. (2003). Bilateral corneal anesthesia associated with diaphragmatic paralysis, ovarian failure, and developmental delay. *Eye Contact Lens* **29**: 262-265.

Chessa, L., Lisa, A., Fiorani, O. and Zei, G. (1994). Ataxia-telangiectasia in Italy: genetic analysis. *International Journal of Radiation Biology* **66**: S31-S33.

Chikatsu, N., Fukumoto, S., Suzawa, M., Tanaka, Y., Takeuchi, Y., Takeda, S., Tamura, Y., Matsumoto, T., *et al.* (1999). An adult patient with severe hypercalcaemia and hypocalciuria due to a novel homozygous inactivating mutation of calcium-sensing receptor. *Clinical Endocrinology* **50**: 537-543.

Chinen, Y., Tohma, T., Izumikawa, Y., Taketomi, H., Iha, T., Ohta, T. and Naritomi, K. (1999). Two sisters with Toriello-Carey syndrome. *American Journal of Medical Genetics* **87**: 262-264.

Chirambo, M. C. and Benezra, D. (1976). Causes of blindness among students in blind school institutions in a developing country. *British Journal of Ophthalmology* **60**: 655-668.

Chitayat, D., Balbul, A., Hani, V., Mamer, O. A., Clow, C. and Scriver, C. R. (1992). Hereditary tyrosinaemia type II in a consanguineous Ashkenazi Jewish family: intrafamilial variation in phenotype; absence of parental phenotype effects on the fetus. *Journal of Inherited Metabolic Disease* **15**: 198-203.

Christianson, A. L., Venter, P. A., Du Toit, J. L., Shipalana, N. and Gericke, G. S. (1994). Acrocallosal syndrome in two African brothers born to consanguineous parents. *American Journal of Medical Genetics* **51**: 98-101.

Christodoulou, K., Zamba, E., Tsingis, M., Mubaidin, A., Horani, K., Abu-Sheik, S., El-Khateeb, M., Kyriacou, K., *et al.* (2000). A novel form of distal hereditary motor neuronopathy maps to chromosome 9p21.1-p12. *Annals of Neurology* **48**: 877-884.

Chun, K., Siegel-Bartelt, J., Chitayat, D., Phillips, J. and Ray, P. N. (1998). FGFR2 mutation associated with clinical manifestations consistent with Antley-Bixler syndrome. *American Journal of Medical Genetics* **77**: 219-224.

Cochat, P., Koch Nogueira, P. C., Mahmoud, M. A., Jamieson, N. V., Scheinman, J. I. and Rolland, M. (1999). Primary hyperoxaluria in infants: medical, ethical, and economic issues. *Journal of Pediatrics* **135**: 746-750.

Coelho, K. E., Ramos, E. S., Felix, T. M., Martelli, L., de Pina-Neto, J. M. and Niikawa, N. (1998). Three new cases of spondylocarpotarsal synostosis syndrome: clinical and radiographic studies. *American Journal of Medical Genetics* **77**: 12-15.

Corden, L. D., Mellerio, J. E., Gratian, M. J., Eady, R. A., Harper, J. I., Lacour, M., Magee, G., Lane, E. B., *et al.* (1998). Homozygous nonsense mutation in helix 2 of K14 causes severe recessive epidermolysis bullosa simplex. *Human Mutation* **11**: 279-285.

Cormier-Daire, V., Munnich, A., Lyonnet, S., Rustin, P., Delezoide, A. L., Maroteaux, P. and Le Merrer, M. (1998). Presentation of six cases of Stuve-Wiedemann syndrome. *Pediatric Radiology* **28**: 776-780.

Cormier-Daire, V., Superti-Furga, A., Munnich, A., Lyonnet, S., Rustin, P., Delezoide, A. L., De Lonlay, P., Giedion, A., *et al.* (1998). Clinical homogeneity of the Stuve-Wiedemann syndrome and overlap with the Schwartz-Jample syndrome type 2. *American Journal of Medical Genetics* **78**: 146-149.

Coskun, T., Özalp, I., Koçak, N., Yüce, A., Çaglar, M. and Berger, R. (1991). Type I hereditary tyrosinaemia: presentation of 11 cases. *Journal of Inherited Metabolic Disease* **14**: 765-770.

Costeff, H. and Dar, H. (1980). Consanguinity analysis of congenital deafness in Northern Israel. *American Journal of Human Genetics* **32**: 64-68.

Coucke, P. J., Van Hauwe, P., Everett, L. A., Demirhan, O., Kabakkaya, Y., Dietrich, N. L., Smith, R. J., Coyle, E., *et al.* (1999). Identification of two different mutations in the PDS gene in an inbred family with Pendred syndrome. *Journal of Medical Genetics* **36**: 475-477.

Coucke, P. J., Wessels, M. W., Van Acker, P., Gardella, R., Barlati, S., Willems, P. J., Colombi, M. and De Paepe, A. (2003). Homozygosity mapping of a gene for arterial tortuosity syndrome to chromosome 20q13. *Journal of Medical Genetics* **40**: 747-751.

Coyle, B., Reardon, W., Herbrick, J. A., Tsui, L. C., Gausden, E., Lee, J., Coffey, R., Grueters, A., *et al.* (1998). Molecular analysis of the PDS gene in Pendred syndrome. *Human Molecular Genetics* **7**: 1105-1112.

Cremers, F. P., van de Pol, D. J., van Driel, M., den Hollander, A. I., van Haren, F. J., Knoers, N. V., Tijmes, N., Bergen, A. A., *et al.* (1998). Autosomal recessive retinitis pigmentosa and cone-rod dystrophy caused by splice site mutations in the Stargardt's disease gene ABCR. *Human Molecular Genetics* **7**: 355-362.

Criado, G. R. and Aytes, A. P. (1999). Mobius sequence, hypogenitalism, cerebral, and skeletal malformations in two brothers. *American Journal of Medical Genetics* **86**: 492-496.

Cristina De Frutos Martinez, M., Elorza Martinez, M., Salas Hernandez, S., Garcia Sanchez, P., Jaso Cortes, E., Perez-Rodriguez, J. and Quero Jimenez, J. (2000). [Multiple type I pseudohypoaldosteronism: neonatal management and outcome]. *Anales Espanoles de Pediatria* **52**: 47-51 [In Spanish].

Croxen, R., Hatton, C., Shelley, C., Brydson, M., Chauplannaz, G., Oosterhuis, H., Vincent, A., Newsom-Davis, J., *et al.* (2002). Recessive inheritance and variable penetrance of slow-channel congenital myasthenic syndromes. *Neurology* **59**: 162-168.

Cury, V. F., Costa, J. E., Gomez, R. S., Boson, W. L., Loures, C. G. and De, M. L. (2002). A novel mutation of the cathepsin C gene in Papillon-Lefevre syndrome. *Journal of Periodontology* **73**: 307-312.

Cusco, I., Lopez, E., Soler-Botija, C., Jesus Barcelo, M., Baiget, M. and Tizzano, E. F. (2003). A genetic and phenotypic analysis in Spanish spinal muscular atrophy patients with c.399_402del AGAG, the most frequently found subtle mutation in the SMN1 gene. *Human Mutatation* **22**: 136-143.

Czeizel, A. and Révész, C. (1970). Major malformations of the central nervous system in Hungary. *British Journal of Social and Preventive Medicine* **24**: 205-222.