

Clinical Outcomes, Physical Disorders (A)

- Abbas, I. S. and Hilal, M. A. (2000). Cystic fibrosis and hypertelorism: a case report. *Annals of Tropical Paediatrics* **20**: 67-69.
- Abdel-al, Y. K., Badawi, M. H., Yaeesh, S. A., Habib, Y. Q., al-Khuffash, F. A., al-Ghanim, M. M. and al-Najidi, A. K. (1999). Bartter's syndrome in Arabic children: review of 13 cases. *Pediatrics International* **41**: 299-303.
- Abdul Wahab, A., Al Thani, G., Dawod, S. T., Kambouris, M. and Al Hamed, M. (2001). Heterogeneity of the cystic fibrosis phenotype in a large kindred family in Qatar with cystic fibrosis mutation (I1234V). *Journal of Tropical Pediatrics* **47**: 110-112.
- Abdul Wahab, A., Dawod, S. T. and al Thani, G. (2000). Cystic fibrosis in a large kindred family in Qatar. *Annals of Tropical Paediatrics* **20**: 203-207.
- Abdul Wahab, A., Janahi, I. A., Eltohami, A., Zeid, A., Ul Haque, N. F. and Teebi, A. S. (2003). A new type of Ehlers-Danlos syndrome associated with tortuous systemic arteries in a large kindred from Qatar. *Acta Paediatrica* **92**: 456-462.
- Abdullah, M. A., Al-Hasnan, Z., Okamoto, E. and Abomelha, A. M. (2000). Arthrogyriposis, renal dysfunction and cholestasis syndrome. *Saudi Medical Journal* **21**: 297-299.
- Abdulrazzaq, Y. M. (1999). Consanguineous marriages and their effect on health. *Middle East Paediatrics* **4**: 24-29.
- Abdulrazzaq, Y. M., Bener, A., Al-Gazali, L. I., Al-Khayat, A. I., Micallef, R. and Gaber, T. (1997). A study of possible deleterious effects of consanguinity. *Clinical Genetics* **51**: 167-173.
- Abe, T., Yamaguchi, Y., Izumino, K., Ozaki, M., Yamakawa, K., Kondo, H., Sera, Y., Uotani, S., *et al.* (2000). Evaluation of insulin response in glucose tolerance test in a patient with Werner's syndrome: a 16-year follow-up study. *Diabetes, Nutrition & Metabolism* **13**: 113-118.
- Abe, Y., Oshima, N., Hanakita, R., Amako, T. and Hirohata, R. (1960). Thirteen cases of alkaptonuria from one family tree with special reference to osteo-arthrosis alkaptonuria. *Journal of Bone and Joint Surgery* **42-A**: 817-831.
- Abou-Dauod, K. T. (1969). Diabetes mellitus in a Lebanese population group. *American Journal of Epidemiology* **89**: 644-650.
- Abraham, L., Moses, P. D., Jacob, M., Kirubakaran, C. and George, R. (1995). Congenital erythropoietic porphyria. *Indian Journal of Pediatrics* **32**: 99-100.

- Abramowicz, M. J., Albuquerque-Silva, J. and Zanen, A. (2002). Corneal dystrophy and perceptive deafness (Harboyan syndrome): CDPD1 maps to 20p13. *Journal of Medical Genetics*: 110-112.
- Abramowicz, M. J., Duprez, L., Parma, J., Vassart, G. and Heinrichs, C. (1997). Familial congenital hypothyroidism due to inactivating mutation of the thyrotropin receptor causing profound hypoplasia of the thyroid gland. *Journal of Clinical Investigation* **99**: 3018-3024.
- Abu-Amero, K. K., Al-Hamed, M. H. and Al-Batniji, F. S. (2003). Homozygous protein C deficiency with purpura fulminans: report of a new case and a description of a novel mutation. *Blood Coagulation and Fibrinolysis* **14**: 303-306.
- Abul-Einen, M. and Topozada, H. K. (1966). Aspects of births in the Shatby Hospital, Alexandria. *British Journal of Preventative and Social Medicine* **20**: 176-180.
- Acierno, J. S., Jr., Shagoury, J. K., Bo-Abbas, Y., Crowley, W. F., Jr. and Seminara, S. B. (2003). A locus for autosomal recessive idiopathic hypogonadotropic hypogonadism on chromosome 19p13.3. *Journal of Clinical Endocrinology and Metabolism* **88**: 2947-2950.
- Ackerman, M. J. (1998). The long QT syndrome. *Pediatric Review* **19**: 232-238.
- Acosta, A. X., Peres, L. C., Chimelli, L. C. and Pina-Neto, J. M. (2000). Raine dysplasia: a Brazilian case with a mild radiological involvement. *Clinical Dysmorphology* **9**: 99-101.
- Adato, A., Raskin, L., Petit, C. and Bonne-Tamir, B. (2000). Deafness heterogeneity in a Druze isolate from the Middle East: novel OTOF and PDS mutations, low prevalence of GJB2 35delG mutation and indication for a new DFNB locus. *European Journal of Human Genetics* **8**: 437-442.
- Afzal, A. R., Rajab, A., Fenske, C., Crosby, A., Lahiri, N., Ternes-Pereira, E., Murday, V. A., Houlston, R., *et al.* (2000). Linkage of recessive Robinow syndrome to a 4 cM interval on chromosome 9q22. *Human Genetics* **106**: 351-354.
- Afzal, A. R., Rajab, A., Fenske, C. D., Oldridge, M., Elanko, N., Ternes-Pereira, E., Tuysuz, B., Murday, V. A., *et al.* (2000). Recessive Robinow syndrome, allelic to dominant brachydactyly type B, is caused by mutation of ROR2. *Nature Genetics* **25**: 419-422.
- Agarwal, G., Bhatia, V., Cook, S. and Thomas, P. Q. (2000). Adrenocorticotropin deficiency in combined pituitary hormone deficiency patients homozygous for a novel PROP1 deletion. *Journal of Clinical Endocrinology & Metabolism* **85**: 4556-4561.
- Agarwal, S. S., Singh, V., Singh, P. S., Singh, S. S., Das, V., Sharma, A., Mehra, P., Chandravati, G. K., *et al.* (1991). Prevalence and spectrum of congenital malformations in a prospective study at a teaching hospital. *Indian Journal of Medical Research* **94**: 413-419.

- Aguila, A., Nazer, J., Cifuentes, L., Mella, P., de la Barra, P. and Gutierrez, D. (2000). [Prevalence of congenital malformations at birth and associated factors in Easter Island, Chile (1988-1998)]. *Revista Medica de Chile* **128**: 162-166 [In Spanish].
- Ahmad, M., Abbas, H. and Haque, S. (1993). Alopecia universalis as a single abnormality in an inbred Pakistani kindred. *American Journal of Medical Genetics* **46**: 369-371.
- Ahmad, M., Abbas, H., Wahab, A. and Haque, S. (1990). Fibular hypoplasia and complex brachydactyly (Du Pan syndrome) in an inbred Pakistani kindred. *American Journal of Medical Genetics* **36**: 292-296.
- Ahmad, M., Haque, M. F., Ahmad, W., Abbas, H., Haque, S., Krakow, D., Rimoin, D. L., Lachman, R. S., *et al.* (1998). Distinct, autosomal recessive form of spondyloepimetaphyseal dysplasia segregating in an inbred Pakistani kindred. *American Journal of Medical Genetics* **78**: 468-473.
- Ahmad, W., Irvine, A. D., Buckley, C., Bingham, E. A., Panteleyev, A. A., Ahmad, M., McGrath, J. A. and Christiano, A. M. (1998). A missense mutation on the Zinc-Finger domain of the human hairless gene underlies congenital atrichia in a family of Irish travellers. *American Journal of Human Genetics* **63**: 984-991.
- Ahmad, W., ul Haque, M. F., Brancolini, V., Tsou, H. C., ul Haque, S., Lam, H.-N., Aita, V. M., Owen, J., *et al.* (1998). Alopecia universalis associated with a mutation in the human hairless gene. *Science* **279**: 720-724.
- Ahmad, W., Zlotogorski, A., Panteleyev, A. A., Lam, H., Ahmad, M., ul Haque, M. F., Abdallah, H. M., Dragan, L., *et al.* (1999). Genomic organization of the human hairless gene (HR) and identification of a mutation underlying congenital atrichia in an Arab Palestinian family. *Genomics* **56**: 141-148.
- Ahn, J. K., Lev, D., Leshinsky-Silver, E., Ginzberg, M. and Lerman-Sagie, T. (2003). A new autosomal recessive syndrome with Zellweger-like manifestations. *American Journal of Medical Genetics* **119A**: 352-355.
- Ainsworth, J. R., Morton, J. E., Good, P., Woods, C. G., George, N. D., Shield, J. P., Bradbury, J., Henderson, M. J., *et al.* (2001). Micro syndrome in Muslim Pakistan children. *Ophthalmology* **108**: 491-497.
- Airede, K. I. (1992). Neural tube defects in the middle belt of Nigeria. *Journal of Tropical Pediatrics* **38**: 27-30.
- Ajlouni, K., Jarrah, N., El-Khateeb, M., El-Zaheri, M., El Shanti, H. and Lidral, A. (2002). Wolfram syndrome: identification of a phenotypic and genotypic variant from Jordan. *American Journal of Medical Genetics* **115**: 61-65.

- Åkesson, H. D., Hagberg, B., Wahlström, J. and Engerström, I. W. (1992). Rett syndrome: A search for gene sources. *American Journal of Medical Genetics* **42**: 104-108.
- Åkesson, H. O., Wahlström, J., Engerström, I. W. and Hagberg, B. (1995). Rett syndrome: potential gene sources - phenotypical variability. *Clinical Genetics* **48**: 169-172.
- Akiyama, M., Christiano, A. M., Yoneda, K. and Shimizu, H. (1998). Abnormal cornified cell envelope formation in mutilating palmoplantar keratoderma unrelated to epidermal differentiation complex. *Journal of Investigative Dermatology* **111**: 133-138.
- Aksu, G., Kutukculer, N., Genel, F., Vergin, C. and Omowaire, B. (2003). Griscelli syndrome without hemophagocytosis in an eleven-year-old girl: expanding the phenotypic spectrum of Rab27A mutations in humans. *American Journal of Medical Genetics* **116A**: 329-333.
- Al Aqeel, A., Al Sewairi, W., Edress, B., Gorlin, R. J., Desnick, R. J. and Martignetti, J. A. (2000). Inherited multicentric osteolysis with arthritis: a variant resembling Torg syndrome in a Saudi family. *American Journal of Medical Genetics* **93**: 11-18.
- Al Essa, M., Rahbeeni, Z., Jumaah, S., Joshi, S., Al Jishi, E., Rashed, M. S., Al Amoudi, M. and Ozand, P. T. (1998). Infectious complications of propionic acidemia in Saudia Arabia. *Clinical Genetics* **54**: 90-94.
- Al Hosani, H. and Czeizel, A. E. (1996). Unique demographic situation in the United Arab Emirates [letter]. *American Journal of Medical Genetics* **61**: 1.
- Al Rajeh, S., Majumdar, R., Awada, A., Adeyokunnu, A., Al Jumah, M., Al Bunyan, M. and Snellen, A. (1998). Molecular analysis of the SMN and NAIP genes in Saudi spinal muscular atrophy patients. *Journal of Neurological Science* **158**: 43-46.
- al Rashed, A. A., al-Jarallah, A. A., Salih, M. A., Kolawole, T. and al-Jarallah, J. (1999). Sotos syndrome (cerebral gigantism): a clinical and radiological study of 14 cases from Saudi Arabia. *Annals of Tropical Paediatrics* **19**: 197-203.
- Al Tawari, A. A., Ramadan, D. G., Neubauer, D., Heberle, L. C. and Al Awadi, F. (2002). An early onset form of methylenetetrahydrofolate reductase deficiency: a report of a family from Kuwait. *Brain Dev* **24**: 304-9.
- al-Abdulkareem, A. A. and Ballal, S. G. (1998). Consanguineous marriage in an urban area of Saudi Arabia: rates and adverse health effects on the offspring. *Journal of Community Health* **23**: 75-83.
- Al-Aqeel, A. I., Rashed, M. S., Ruitter, J. P., Al-Husseini, H. F., Al-Amoudi, M. S. and Wanders, R. J. (2001). Carnitine palmityl transferase I deficiency. *Saudi Medical Journal* **22**: 1025-1029.

- Al-Attas, R. A. and Rahi, A. H. S. (1998). Primary antibody deficiency in Arabs: first report from Eastern Saudi Arabia. *Journal of Clinical Immunology* **18**: 368-371.
- Al-Awadi, S. A., Cuschieri, A., Farag, T. I., Naguib, K., Teebi, A. S., Al-Othman, S. A. and Bahig, A. H. (1983). Ullrich-Turner syndrome in monozygotic twins. *American Journal of Medical Genetics* **15**: 537-542.
- Alba, M., Hall, C. M., Whatmore, A. J., Clayton, P. E., Price, D. A. and Salvatori, R. (2004). Variability in anterior pituitary size within members of a family with GH deficiency due to a new splice mutation in the GHRH receptor gene. *Clinical Endocrinology (Oxford)* **60**: 470-475.
- Alba, M. and Salvatori, R. (2004). Familial Growth Hormone Deficiency and Mutations in the GHRH Receptor Gene. *Vitamins and Hormones* **69**: 209-220.
- Al-Batniji, F. S., Mahmoud, M. A., Van Dijken, P. J., Al-Asiri, R. H., Al-Swaid, A. F. and Al-Marshedy, A. M. (2001). A new autosomal recessive syndrome. Early onset of pancytopenia, distinct facial features, growth retardation and developmental delay. *Saudi Medical Journal* **22**: 1122-1126.
- Al-Eissa, Y. A. (1995). Febrile seizures: rate and risk factors of recurrence. *Journal of Child Neurology* **10**: 315-319.
- Alembik, Y., Abdulrazzaq, Y. M., Bener, A., Al-Gazali, L. I., Al-Jhayat, A. I., Micallef, R. and Gaber, T. (1997). A study of possible deleterious effects of consanguinity. *Clinical Genetics* **51**: 167-713.
- al-Essa, M. A., Rashed, M. S., Bakheet, S. M., Patay, Z. J. and Ozand, P. T. (2000). Glutaric aciduria type II: observations in seven patients with neonatal- and late-onset disease. *Journal of Perinatology* **20**: 120-128.
- Alfadley, A. and Parkes, R. M. (2002). Two siblings born preterm with large ears and hypopigmented hair who developed palmoplantar keratoderma and frontal skull bossing: a new syndrome? *Pediatric Dermatology* **19**: 224-228.
- Alfi, O. S., Chang, R. and Azen, S. P. (1980). Evidence for genetic control of nondisjunction in man. *American Journal of Human Genetics* **32**: 477-483.
- al-Fuzae, L., Aboolbacker, K. C. and al-Saleh, Q. (1998). Beta-Thalassaemia major in Kuwait. *Journal of Tropical Pediatrics* **44**: 311-312.
- Al-Gazali, L. I. (1998). A genetic aetiological survey of severe childhood deafness in the United Arab Emirates. *Journal of Tropical Pediatrics* **44**: 157-160.
- Al-Gazali, L. I. (1998). Mental retardation, iris coloboma, optic atrophy and distinctive facial appearance in two sibs. *Clinical Dysmorphology* **7**: 201-203.

- Al-Gazali, L. I. and Abou Al-Asaad, F. (1995). Autosomal recessive omodysplasia. *Clinical Dysmorphology* **4**: 52-56.
- Al-Gazali, L. I., Al Talabani, J., Mosawi, A. and Lytle, W. (1994). Anterior segment anomalies of the eye, clefting and skeletal abnormalities in two sibs of consanguineous parents: Michels syndrome or new syndrome? *Clinical Dymorphology* **3**: 238-244.
- Al-Gazali, L. I. and Bakalinova, D. (1998). Autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia and distinctive facial appearance. *Clinical Dysmorphology* **7**: 177-184.
- Al-Gazali, L. I., Bakalinova, D., Aziz, S., Anwer, O., Shather, W. and Sztriha, L. (1999). Hypocalvaria associated with intrauterine growth retardation, facial dysmorphism, congenital heart disease and camptomelia. *Clinical Dysmorphology* **8**: 129-134.
- Al-Gazali, L. I., Bakalinova, D. and Bakir, M. (1998). Central nervous system malformations, dense bones and facial dysmorphism: a new autosomal recessive syndrome. *Clinical Dysmorphology* **7**: 123-126.
- Al-Gazali, L. I., Bakir, M., Dawodu, A., Nath, R., al-Tatari, H. M. and Gerami, M. (1999). Recurrence of the severe form of microgastria-limb reduction defect in a consanguineous family. *Clinical Dysmorphology* **8**: 253-258.
- Al-Gazali, L. I., Bakir, M., Hamud, O. A. and Gerami, S. (2002). An autosomal recessive syndrome of nasal anomalies associated with renal and anorectal malformations. *Clinical Dysmorphology* **11**: 33-38.
- Al-Gazali, L. I., Bakir, M., Sadaghatian, M. R., Nath, R. and Haas, D. (1999). Anterior segment anomalies of the eye associated with multiple skeletal abnormalities and early lethality: confirmation of an autosomal recessive syndrome. *Clinical Dysmorphology* **8**: 87-92.
- Al-Gazali, L. I., Dawodu, A. H., Sabarinathan, K. and Varghese, M. (1995). The profile of major congenital abnormalities in the United Arab Emirates (UAE) population. *Journal of Medical Genetics* **32**: 7-13.
- Al-Gazali, L. I., Khalil, M. and Devadas, K. (1993). A syndrome of insulin resistance resembling leprechaunism in five sibs of consanguineous parents. *Journal of Medical Genetics* **30**: 470-475.
- Al-Gazali, L. I. and Lytle, W. (1994). Otospondylomegaepiphyseal dysplasia: report of three sibs and review of the literature. *Clinical Dysmorphology* **3**: 46-54.
- Al-Gazali, L. I., Sabarinathan, D. K. and Khidir, A. (1994). Microphthalmia and distal limb abnormalities in a child of consanguineous parents. *Clinical Dysmorphology* **3**: 258-262.

- Al-Gazali, L. I., Sabrinathan, K. and Nair, K. G. R. (1994). A syndrome of osteogenesis imperfecta, optic atrophy, retinopathy and severe developmental delay in two sibs of consanguineous parents. *Clinical Dysmorphology* **3**: 55-62.
- Al-Gazali, L. I., Sztriha, L., Dawodu, A., Bakir, M., Varghese, M., Varady, E., Scorer, J., Abdulrazzaq, Y. M., *et al.* (1999). Pattern of central nervous system anomalies in a population with a high rate of consanguineous marriages. *Clinical Genetics* **55**: 95-102.
- Al-Gazali, L. I., Sztriha, L., Dawodu, A., Varady, E., Bakir, M., Khdir, A. and Johansen, J. (1999). Complex consanguinity associated with short rib-polydactyly syndrome III and congenital infection-like syndrome: a diagnostic problem in dysmorphic syndromes. *Journal of Medical Genetics* **36**: 461-466.
- Al-Gazali, L. I., Sztriha, L., Skaff, F. and Haas, D. (2001). Geroderma osteodysplastica and wrinkly skin syndrome: are they the same? *American Journal of Medical Genetics* **101**: 213-220.
- Al-Hawsawi, Z. M. and Ismail, G. A. (2001). Acute splenic sequestration crisis in children with sickle cell disease. *Saudi Medical Journal* **22**: 1076-1079.
- Al-Idrissi, I., Al-Kaff, A. S. and Senft, S. H. (1992). Cumulative incidence of retinoblastoma in Riyadh, Saudi Arabia. *Ophthalmic Paediatrics and Genetics* **13**: 9-12.
- Alif, N., Hess, K., Straczek, J., Sebbar, S., Belahsen, Y., Mouane, N., Abkari, A., Nabet, P., *et al.* (2000). Mucopolysaccharidosis type I in Morocco: clinical features and genetic profile. *Archives of Pediatrics* **7**: 597-604.
- Aligianis, I. A., Forshew, T., Johnson, S., Michaelides, M., Johnson, C. A., Trembath, R. C., Hunt, D. M., Moore, A. T., *et al.* (2002). Mapping of a novel locus for achromatopsia (ACHM4) to 1p and identification of a germline mutation in the alpha subunit of cone transducin (GNAT2). *Journal of Medical Genetics* **39**: 656-660.
- Alikasifoglu, M., Topaloglu, H., Tuncbilek, E., Ceviz, N., Anar, B., Demir, D. and Ozme, S. (1999). Clinical and genetic correlates in childhood onset Friedreich ataxia. *Neuropediatrics* **30**: 72-76.
- Al-Khenaizan, S. (2003). Hyperpigmentation in Chediak-Higashi syndrome. *Journal of the American Academy of Dermatology* **49**: S244-246.
- Allikmets, R., Singh, N., Sun, H., Shroyer, N. F., Hutchinson, A., Chidambaram, A., Gerrard, B., Baird, L., *et al.* (1997). A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Stargardt macular dystrophy. *Nature Genetics* **15**: 236-246.
- Al-Mahroos, F. (1998). Cystic fibrosis in Bahrain : incidence, phenotype, and outcome. *Journal of Tropical Pediatrics* **44**: 35-39.

- Al-Mahroos, F. and McKeigue, P. M. (1998). High prevalence of diabetes in Bahrainis. Associations with ethnicity and raised plasma cholesterol. *Diabetes Care* **21**: 936-942.
- Al-Mayouf, S. M., Majeed, M., Hugosson, C. and Bahabri, S. (2000). New form of idiopathic osteolysis: nodulosis, arthropathy and osteolysis (NAO) syndrome. *American Journal of Medical Genetics* **93**: 5-10.
- Al-Najjadah, I., Bang, R. L., Ghoneim, I. E. and Kanjoor, J. R. (2003). Infantile systemic hyalinosis. *Journal of Craniofacial Surgery* **14**: 719-723.
- Al-Qattan, M. M. (2003). Congenital duplication of the palm in a patient with multiple anomalies. *Journal of Hand Surgery [Br]* **28**: 276-279.
- Al-Quadah, A. A. (1998). Clinical patterns of neuronal migrational disorders and parental consanguinity. *Journal of Tropical Pediatrics* **44**: 351-354.
- Al-Quadah, A. A. and Tarawneh, M. (1998). Congenital muscular dystrophy in Jordanian children. *Journal of Child Neurology* **13**: 383-386.
- Al-Rabeeah, A., Al-Ashwal, A., Al-Herbish, A., Al-Jurayyan, N., Sakati, N. and Abobakr, A. (1995). Persistent hyperinsulinemic hypoglycemia of infancy: experience with 28 cases. *Journal of Pediatric Surgery* **30**: 1119-1121.
- al-Rasheed, S. A., al-Mohrij, O., al-Jurayyan, N., al-Herbish, M., al-Salloum, A., al-Hussain, M. and el-Desouki, M. (1998). Osteoporosis in children. *International Journal of Clinical Practice* **52**: 15-18.
- Al-Salem, M. and Rawashdeh, N. (1992). Patterns of childhood blindness and partial sight among Jordanians in two generations. *Journal of Pediatric Ophthalmology and Strabismus* **29**: 361-365.
- Alsheikheh, A., Lieb, W. and Grehn, F. (2004). [Criswick-Schepens syndrome -- familial exudative vitreoretinopathy. Report of six cases in two consanguineous families]. *Ophthalmologie* **101**: 914-918.
- Altay, C., Alikasifoglu, M., Kara, A., Tunçbilek, E., Özbek, N. and Schroeder-Kurth, T. M. (1997). Analysis of 65 Turkish patients with congenital aplastic anaemia (Fanconi anaemia and non-Fanconi anaemia): Hacettepe experience. *Clinical Genetics* **51**: 296-302.
- Al-Till, M., Jarrah, N. S. and Ajlouni, K. M. (2002). Ophthalmologic findings in fifteen patients with Wolfram syndrome. *European Journal of Ophthalmology* **12**: 84-88.
- Al-Twaijri, W. A. and Shevell, M. I. (2002). A novel chronic childhood sensory predominant neuropathy. *Pediatric Neurology* **27**: 49-52.

- Al-Yahyaee, S. A., Al-Kindi, M. N., Habbal, O. and Kumar, D. S. (2003). Clinical and molecular analysis of Grebe acromesomelic dysplasia in an Omani family. *American Journal of Medical Genetics* **121A**: 9-14.
- Amadei, N., Baracho, G. V., Nudelman, V., Bastos, W., Florido, M. P. and Isaac, L. (2001). Inherited complete factor I deficiency associated with systemic lupus erythematosus, higher susceptibility to infection and low levels of factor H. *Scandinavian Journal of Immunology* **53**: 615-621.
- Ambrugger, P., Stoeva, I., Biebermann, H., Torresani, T., Leitner, C. and Gruters, A. (2001). Novel mutations of the thyroid peroxidase gene in patients with permanent congenital hypothyroidism. *European Journal of Endocrinology* **145**: 19-24.
- Amor, D. J., Leventer, R. J., Hayllar, S. and Bankier, A. (2000). Polymicrogyria associated with scalp and limb defects: variant of Adams-Oliver syndrome. *American Journal of Medical Genetics* **93**: 328-334.
- Angel, T. A., Hsu, S., Kornbleuth, S. I., Kornbleuth, J. and Kramer, E. M. (2002). Papillon-Lefevre syndrome: a case report of four affected siblings. *Journal of the American Academy of Dermatology* **46**: S8-S10.
- Anwar, A., Rambaud-Cousson, D. and Rambaud-Cousson, A. (1993). Syndrome of infantile osteopetrosis and Hirschspung disease in seven children born to four consanguineous unions in two families. *American Journal of Medical Genetics* **47**: 1083-1085.
- Appaji Rao, N., Radha Rama Devi, A., Savithri, H. S., Venkat Rao, S. and Bittles, A. H. (1988). Neonatal screening for amino acidurias in Karnataka, South India. *Clinical Genetics* **34**: 60-63.
- Applegarth, D. A., Toone, J. R., Rolland, M. O., Black, S. H., Yim, D. K. and Bemis, G. (2000). Non-concordance of CVS and liver glycine cleavage enzyme in three families with non-ketotic hyperglycinaemia (NKH) leading to false negative prenatal diagnoses. *Prenatal Diagnosis* **20**: 367-370.
- Argent, A. C., Chetty, M. C., Fricke, B., Bertrand, Y., Philippe, N., Khogali, S., von During, M., Delaunay, J., *et al.* (2004). A family showing recessively inherited multisystem pathology with aberrant splicing of the erythrocyte Band 7.2b ('stomatin') gene. *Journal of Inherited Metabolic Disease* **27**: 29-46.
- Arico, M., Dellavecchia, C., Piantanida, M., Clementi, R., Hasle, H., Conter, V., D'Angelo, P., Varotto, S., *et al.* (1999). The breakpoints of a constitutional inversion of chromosome 9 associated with haemophagocytic lymphohistiocytosis are not linked to the disease gene. *British Journal of Haematology* **104**: 108-110.

- Arico, M., Nichols, K., Whitlock, J. A., Arceci, R., Haupt, R., Mittler, U., Kuhne, T., Lombardi, A., *et al.* (1999). Familial clustering of Langerhans cell histiocytosis. *British Journal of Haematology* **107**: 883-888.
- Arikawa-Hirasawa, E., Wilcox, W. R., Le, A. H., Silverman, N., Govindraj, P., Hassell, J. R. and Yamada, Y. (2001). Dyssegmental dysplasia, Silverman-Handmaker type, is caused by functional null mutations of the perlecan gene. *Nature Genetics* **27**: 431-434.
- Arnell, H., Mantyjarvi, M., Tuppurainen, K., Andreasson, S. and Dahl, N. (1998). Stargardt disease: linkage to the ABCR gene region on 1p21-p22 in Scandinavian families. *Acta Ophthalmologica Scandinavica* **76**: 649-652.
- Asaka, T., Ikeuchi, K., Okino, S., Takizawa, Y., Satake, R., Nitta, E., Komai, K., Endo, K., *et al.* (2001). Homozygosity and linkage disequilibrium mapping of autosomal recessive distal myopathy (Nonaka distal myopathy). *Journal of Human Genetics* **46**: 649-655.
- Asteria, C., Oliveira, J. H., Abucham, J. and Beck-Peccoz, P. (2000). Central hypocortisolism as part of combined pituitary hormone deficiency due to mutations of PROP-1 gene. *European Journal of Endocrinology* **143**: 347-352.
- Atmaca, L. S., Sayli, B. S., Akarsu, N. and Gündüz, K. (1995). Genetic features of retinitis pigmentosa in Turkey. *Documenta Ophthalmologia* **89**: 387-392.
- Aughton, D. J. (1990). New syndrome-clinical anophthalmia, dextrocardia, and skeletal anomalies in an infant born to consanguineous parents. *American Journal of Medical Genetics* **37**: 178-181.
- Ayman, T., Yerebakan, O. and Yilmaz, E. (2000). Mal de Meleda: a review of Turkish reports. *Journal of Dermatology* **27**: 664-668.
- Aymé, S., Pelissier, J. F., Garnier, J. M., Matthei, J. F. and Giraud, F. (1979). Duchenne type muscular dystrophy and consanguinity: difficulties in pedigree analysis. *Journal of Medical Genetics* **16**: 393-395.
- Aymé, S. and Philip, N. (1995). Possible homozygous Waardenburg syndrome in a fetus with exencephaly [letter]. *American Journal of Medical Genetics* **59**: 263-265.
- Azzedine, H., Bolino, A., Taieb, T., Birouk, N., Di Duca, M., Bouhouche, A., Benamou, S., Mrabet, A., *et al.* (2003). Mutations in MTMR13, a new pseudophosphatase homologue of MTMR2 and Sbf1, in two families with an autosomal recessive demyelinating form of Charcot-Marie-Tooth disease associated with early-onset glaucoma. *American Journal of Human Genetics* **72**: 1141-1153.