

### Clinical Outcomes, Physical Disorders (D-F)

- da Silva, E. O., Janovitz, D. and Cavalcanti de Albuquerque (1980). Ellis-Van Creveld syndrome: report of 15 cases in an inbred kindred. *Journal of Medical Genetics* **17**: 349-356.
- Dagher, H., Yan Wang, Y., Fassett, R. and Savige, J. (2002). Three novel COL4A4 mutations resulting in stop codons and their clinical effects in autosomal recessive Alport syndrome. *Human Mutation* **20**: 321-322.
- Damji, K. F., Sohocki, M. M., Khan, R., Gupta, S. K., Rahim, M., Loyer, M., Hussein, N., Karim, N., *et al.* (2001). Leber's congenital amaurosis with anterior keratoconus in Pakistani families is caused by the Trp278X mutation in the AIPL1 gene on 17p. *Canadian Journal of Ophthalmology* **36**: 252-259.
- Danciger, M., Hendrickson, J., Lyon, J., Toomes, C., McHale, J. C., Fishman, G. A., Inglehearn, C. F., Jacobson, S. G., *et al.* (2001). CORD9 a new locus for arCRD: mapping to 8p11, estimation of frequency, evaluation of a candidate gene. *Investigative Ophthalmology & Visual Science* **42**: 2458-265.
- Dandona, L., Dandona, R., Naduvilath, T. J., McCarty, C. A., Nanda, A., Srinivas, M., Mandal, P. and Rao, G. N. (1998). Is current eye-care-policy focus almost exclusively on cataract adequate to deal with blindness in India. *Lancet* **351**: 1312-1316.
- Dave, S., Thappa, D. M. and Karthikeyan, K. (2003). Disseminated and disfiguring molluscum contagiosum in a child. *Pediatric Dermatology* **20**: 436-439.
- Davies, D. L., Dutton, G. N., Farquharson, J., Logan, R. W. and Tolmie, J. L. (1989). Hurler-Scheie phenotype associated with consanguinity. *Journal of Inherited Metabolic Disease* **12**: 365-368.
- Davis, C. J., Davison, R. M., Payne, N. N., Rodeck, C. H. and Conway, G. S. (2000). Female sex preponderance for idiopathic familial premature ovarian failure suggests an X chromosome defect: opinion. *Human Reproduction* **15**: 2418-2422.
- De Braekeleer, M. (1995). Geographical distribution of 18 autosomal recessive disorders in the French Canadian population of Saguenay-Lac-Saint-Jean, Québec. *Annals of Human Biology* **22**: 111-122.
- De Braekeleer, M. (1996). Autosomal recessive disorders in Saguenay-Lac-Saint-Jean (Québec, Canada): estimation of inbreeding from isonomy. *Annals of Human Biology* **23**: 95-99.
- De Braekeleer, M., Daigneault, J., Allard, C., Simard, F. and Aubin, G. (1996). Genealogy and geographical distribution of CFTR mutations in Saguenay-Lac-Saint-Jean (Québec, Canada). *Annals of Human Biology* **23**: 345-352.

- De Braekeleer, M. and Gauthier, S. (1996). Autosomal recessive disorders in Saguenay-Lac-St-Jean, Québec: study of kinship. *Human Biology* **68**: 371-381.
- De Braekeleer, M., Giasson, F., Mathieu, J., Roy, M., Bouchard, J. P. and Morgan, K. (1993). Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Sagueney in North Eastern Quebec. *Genetic Epidemiology* **10**: 17-25.
- De Braekeleer, M., Pérusse, L., Cantin, L., Bouchard, J. and Mathieu, J. (1996). A study of inbreeding and kinship in intracranial aneurysms in the Saguenay Lac-Saint-Jean region (Québec, Canada). *Annals of Human Genetics* **60**: 99-104.
- de Costa, C. (1986). Congenital abnormalities and consanguinity. *Medical Journal of Australia* **144**: 721-722.
- de Delbue, S. M. (1950-1951). Family size in some hereditary disorders. *Annals of Eugenics* **15**: 184-185.
- de Haar, S. F., Jansen, D. C., Schoenmaker, T., De Vree, H., Everts, V. and Beertsen, W. (2004). Loss-of-function mutations in cathepsin C in two families with Papillon-Lefevre syndrome are associated with deficiency of serine proteinases in PMNs. *Human Mutation* **23**: 524.
- de la Calle-Martin, O., Hernandez, M., Ordi, J., Casamitjana, N., Arostegui, J. I., Caragol, I., Ferrando, M., Labrador, M., *et al.* (2001). Familial CD8 deficiency due to a mutation in the CD8 alpha gene. *Journal of Clinical Investigation* **108**: 117-123.
- De Los Rios, P. and Pla, O. (2000). Statistical analysis of genealogical trees for polygamic species. *Physical Review E (Statistical Physics, Plasmas, Fluids, and Related Interdisciplinary Topics)* **61**: 5620-5623.
- De Michele, G., De Fusco, M., Cavalcanti, F., Filla, A., Marconi, R., Volpe, G., Monticelli, A., Ballabio, A., *et al.* (1998). A new locus for autosomal recessive hereditary spastic paraplegia maps to chromosome 16q24.3. *American Journal of Human Genetics* **63**: 135-139.
- De Sandre-Giovannoli, A., Chaouch, M., Kozlov, S., Vallat, J. M., Tazir, M., Kassouri, N., Szepetowski, P., Hammadouche, T., *et al.* (2002). Homozygous defects in LMNA, encoding lamin A/C nuclear-envelope proteins, cause autosomal recessive axonal neuropathy in human (Charcot-Marie-Tooth disorder type 2) and mouse. *American Journal of Human Genetics* **70**: 726-736.
- de Schepper, S., Loeys, B., de Paepe, A., Lambert, J. and Naeyaert, J. M. (2003). Cutis laxa of the autosomal recessive type in a consanguineous family. *European Journal of Dermatology* **13**: 529-533.
- de Silva, D., Suriyawansa, D., Mangalika, M. and Samarasinghe, D. (2001). Meckel Gruber syndrome--a single gene cause of recurrent neural tube defects. *Ceylon Medical Journal* **46**: 30.

- De Vaal, O. M. (1955). Genetic intersexuality in three brothers, connected with consanguineous marriages in the previous generations. *Acta Paediatrica* **44**: 35-39.
- De Vitto, L. P., Costa, O. A., Bevilaqua, M. C., Passerotti, S. and Richieri-Costa, A. (1997). New autosomal recessive syndrome of progressive sensorineural hearing loss and cataracts: Report on two Brazilian patients. *American Journal of Medical Genetics* **70**: 247-249.
- Delague, V., Bareil, C., Bouvagnet, P., Salem, N., Chouery, E., Loiselet, J., Megarbane, A. and Claustres, M. (2002). A new autosomal recessive non-progressive congenital cerebellar ataxia associated with mental retardation, optic atrophy, and skin abnormalities (CAMOS) maps to chromosome 15q24-q26 in a large consanguineous Lebanese Druze Family. *Neurogenetics* **4**: 23-27.
- Delague, V., Bareil, C., Tuffery, S., Bouvagnet, P., Chouery, E., Koussa, S., Maisonobe, T., Loiselet, J., *et al.* (2000). Mapping of a new locus for autosomal recessive demyelinating Charcot-Marie-Tooth disease to 19q13.1-13.3 in a large consanguineous Lebanese family: exclusion of MAG as a candidate gene. *American Journal of Human Genetics* **67**: 236-243.
- Delague, V., Souraty, N., Khallouf, E., Tardy, V., Chouery, E., Halaby, G., Loiselet, J., Morel, Y., *et al.* (2000). Mutational analysis in Lebanese patients with congenital adrenal hyperplasia due to a deficit in 21-hydroxylase. *Hormone Research* **53**: 77-782.
- Delanghe, J., Vlamincq, H., Bernard, D., Robberecht, E., Praet, M., De Buyzere, M., Quatacker, L., Van Roy, N., *et al.* (1997). Absence of serum alanine aminotransferase activity in a neonate. *Clinical Chemistry* **43**: 1665-1667.
- Delepine, M., Nicolino, M., Barrett, T., Golamaully, M., Lathrop, G. M. and Julier, C. (2000). EIF2AK3, encoding translation initiation factor 2-alpha kinase 3, is mutated in patients with Wolcott-Rallison syndrome. *Nature Genetics* **25**: 406-409.
- Dellow, E. L., Harley, K. E., Unwin, R. J., Wrong, O., Winter, G. B. and Parkins, B. J. (1998). Amelogenesis imperfecta, nephrocalcinosis, and hypocalciuria syndrome in two siblings from a large family with consanguineous parents. *Nephrology, Dialysis, Transplantation* **13**: 3193-3196.
- Demir, E., Irobi, J., Erdem, S., Demirci, M., Tan, E., Timmerman, V., De Jonghe, P. and Topaloglu, H. (2003). Andermann syndrome in a Turkish patient. *Journal of Child Neurology* **18**: 76-79.
- Demir, E., Sabatelli, P., Allamand, V., Ferreira, A., Moghadaszadeh, B., Makrelouf, M., Topaloglu, H., Echenne, B., *et al.* (2002). Mutations in COL6A3 cause severe and mild phenotypes of Ullrich congenital muscular dystrophy. *American Journal of Human Genetics* **70**: 1446-1458.

- Demirel, S., Kaplanoglu, N., Acar, A., Bodur, S. and Paydak, F. (1997). The frequency of consanguinity in Konya, Turkey, and its medical effects. *Genetic Counselling* **8**: 295-301.
- den Hollander, N. S., Van der Harten, H. J., Laudy, J. A., van de Weg, P. and Wladimiroff, J. W. (1998). Early transvaginal ultrasonographic diagnosis of Breemer-Langer dysplasia: a report of two cases. *Ultrasound and Obstetric Gynecology* **11**: 298-302.
- Denecke, J., Zimmer, K. P., Kleta, R., Koch, H. G., Rabe, H., August, C. and Harms, E. (2000). Arthrogryposis, renal tubular dysfunction, cholestasis (ARC) syndrome: case report and review of the literature. *Klinische Pädiatrie* **212**: 77-780.
- Denic, S. (2003). Consanguinity as risk factor for cervical carcinoma. *Medical Hypotheses* **60**: 321-324.
- Denic, S. and Bener, A. (2001). Consanguinity decreases risk of breast cancer--cervical cancer unaffected. *British Journal of Cancer* **85**: 1675-1679.
- Der Kaloustian, V. M., Naffah, J. and Loiselet, J. (1980). Genetic diseases in Lebanon. *American Journal of Medical Genetics* **7**: 187-203.
- Derekoy, F. S. (2000). Etiology of deafness in Afyon school for the deaf in Turkey. *International Journal of Pediatric Otorhinolaryngology* **55**: 125-131.
- Desai, M. P. and Khatri, J. V. (1998). Persistent hyperinsulinemic hypoglycemia of infancy. *Indian Pediatrics* **35**: 317-328.
- Dessa Sadovnick, A., Yee, I. M. and Ebers, G. C. (2001). Recurrence risks to sibs of MS index cases: impact of consanguineous matings. *Neurology* **56**: 784-785.
- Desviat, L. R., Perez, B., Gutierrez, E., Sanchez, A., Barrios, B. and Ugarte, M. (2001). Molecular basis of phenylketonuria in Cuba. *Human Mutation* **18**: 252.
- Devos, E. A., Leroy, J. G., Frijns, J. P. and Vanderberghe, H. (1981). The Wiedemann-Rautenstrauch or neonatal progeroid syndrome - report of a patient with consanguineous parents. *European Journal of Pediatrics* **136**: 245-248.
- Devriendt, K., Keymolen, K., Roelen, L., Van Goethem, G., Meireleire, J. and Fryns, J. P. (2000). Severe short stature, hyperphalangy of the index fingers, mental retardation and facial dysmorphism. *Clinical Dysmorphology* **9**: 111-114.
- Di Blasi, C., He, Y., Morandi, L., Cornelio, F., Guicheney, P. and Mora, M. (2001). Mild muscular dystrophy due to a nonsense mutation in the LAMA2 gene resulting in exon skipping. *Brain* **124**: 698-704.
- Di Cerbo, A., Biason-Lauber, A., Savino, M., Piemontese, M. R., Di Giorgio, A., Perona, M. and Savoia, A. (2002). Combined 17alpha-Hydroxylase/17,20-lyase

- deficiency caused by Phe93Cys mutation in the CYP17 gene. *Journal of Clinical Endocrinology and Metabolism* **87**: 898-905.
- Di Rocco, M. (2002). Distal aphyalangia, an extra metatarsal, short stature and microcephaly: a second case. *Clinical Dysmorphology* **11**: 295-296.
- Diaz, G. A., Khan, K. T. and Gelb, B. D. (1998). The autosomal recessive Kenny-Caffey syndrome locus maps to chromosome 1q42-q43. *Genomics* **54**: 13-18.
- Diego Nunez, M. A. and Cortijo Gonzalez, C. (2002). [Familial hypobetalipoproteinemia: Report of a family and review of Spanish contributions]. *Anales Espanoles de Pediatria* **56**: 64-67 [In Spanish].
- Digilio, M. C., Marino, B., Banaudi, E., Marasini, M. and Dallapiccola, B. (1998). Familial recurrence of transposition of the great arteries. *Lancet* **351**: 1661.
- Dincer, P., Balci, B., Yuva, Y., Talim, B., Brockington, M., Dincel, D., Torelli, S., Brown, S., *et al.* (2003). A novel form of recessive limb girdle muscular dystrophy with mental retardation and abnormal expression of alpha-dystroglycan. *Neuromuscular Disorders* **13**: 771-778.
- Dinçer, P., Piccolo, F., Leturcq, F., Kaplan, J. C., Jeanpierre, M. and Topaloglu, H. (1998). Prenatal diagnosis of limb-girdle muscular dystrophy type 2C. *Prenatal Diagnosis* **18**: 1300-1303.
- Ding, H. X., Yang, X. S., Xiao, B., Wu, Z. G. and Zhang, L. F. (2004). [Quantitative analysis of SMN gene copies in spinal muscular atrophy] *Zhonghua Yi Xue Yi Chuan Xue Za Zhi* **21**: 153-155 [In Chinese].
- Dogan, Y., Aygun, D., Yilmaz, Y., Kanra, G., Secmeer, G., Besbas, N. and Gurgey, A. (2003). Severe protein S deficiency associated with heterozygous factor V Leiden mutation in a child with purpura fulminans. *Pediatric Hematology and Oncology* **20**: 1-5.
- dos Santos, R., Castro, N. H., Ferraz, O. P., Walter-Moura, J., Mustachi, Z., Pagnan, N. A. and Gollop, T. R. (1992). Ophthalmological, skeletal, and cardiac abnormalities in sibs born to consanguineous parents: a new syndrome. *American Journal of Medical Genetics* **43**: 946-948.
- Dow, E., Cross, S., Wolgemuth, D. J., Lyonnet, S., Mulligan, L. M., Mascari, M., Ladda, R. and Williamson, R. (1994). Second locus for Hirschsprung disease/Waardenburg syndrome in a large Mennonite kindred. *American Journal of Medical Genetics* **53**: 75-80.
- Driss, A., Amouri, R., Ben Hamida, C., Souilem, S., Gouider-Khouja, N., Ben Hamida, M. and Hentati, F. (2000). A new locus for autosomal recessive limb-girdle muscular dystrophy in a large consanguineous Tunisian family maps to chromosome 19q13.3. *Neuromuscular Disorders* **10**: 240-246.

- Drouet, A., Swalduz, B., Guilloton, L., Faivre, A. and Felten, D. (2003). [Usher syndrome: a case report] [In French]. *Revue Neurologique (Paris)* **159**: 323-325.
- Duarte, R. and Leal, M. J. (1999). [The range of congenital malformations associated with cleft lip and palate]. *Acta Medica Portuguesa* **12**: 147-154 [In Portuguese].
- Dudin, A. A. 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.
- Dudley, J., Allen, J., Tizard, J. and McGraw, M. (1998). Benign methylmalonic acidemia in a sibship with distal renal tubular acidosis. *Pediatric Nephrology* **12**: 564-566.
- Dundar, M., Demiryilmaz, F., Demiryilmaz, I., Kumandas, S., Erkilic, K., Kendirci, M., Tuncel, M., Ozyazgan, I., *et al.* (1997). An autosomal recessive adducted thumb-club foot syndrome observed in Turkish cousins. *Clinical Genetics* **51**: 61-64.
- Eden, E. R., Naoumova, R. P., Burden, J. J., McCarthy, M. I. and Soutar, A. K. (2001). Use of homozygosity mapping to identify a region on chromosome 1 bearing a defective gene that causes autosomal recessive homozygous hypercholesterolemia in two unrelated families. *American Journal of Human Genetics* **68**: 653-660.
- Editorial (1991). Consanguinity and health. *Lancet* **338**: 85-86.
- Eggers, S. and Zatz, M. (1998). Social adjustment in adult males affected with progressive muscular dystrophy. *American Journal of Medical Genetics* **81**: 4-12.
- Ehlers, K. H. and Engle, M. A. (1966). Familial congenital heart disease. I. Genetic and environmental factors. *Circulation* **34**: 503-516.
- Eiberg, H. and Nielsen, I. M. (2000). Linkage of Cholestasis Familiaris Groenlandica/Byler-like disease to chromosome 18. *International Journal of Circumpolar Health* **59**: 57-62.
- Eikenboom, J. C. (2001). Congenital von Willebrand disease type 3: clinical manifestations, pathophysiology and molecular biology. *Best Practice & Research in Clinical Haematology* **14**: 365-379.
- Elbedour, K., Zucker, N., Barki, Y. and Carmi, R. (1994). Cardiac abnormalities in the Bardet-Biedl syndrome: Echocardiographic studies of 22 patients. *American Journal of Medical Genetics* **52**: 164-169.
- Elder, M. J. and De Cock, R. (1993). Childhood blindness in the West Bank and Gaza Strip: Prevalence, aetiology and hereditary factors. *Eye* **7**: 580-583.

- Eldon, B. J., Axelsson, J., Sigurdsson, S. B. and Arnason, E. (2001). Cardiovascular risk factors and relatedness in an Icelandic subpopulation. *International Journal of Circumpolar Health* **60**: 499-502.
- Elfagm, A. A., Gul, S. and Bughaigis, Y. (1980). First cousin marriage and sickle-cell anaemia. *Cell and Molecular Biology* **26**: 109-110.
- El-Garf, A., Mahmoud, G., Gheith, R., Abd El-Aaty, G. and Abd El-Aaty, H. (2003). Camptodactyly, arthropathy, coxa vara, and pericarditis syndrome among egyptians. *Journal of Rheumatology* **30**: 1081-1086.
- Elisbão, T. and Friere-Maia, N. (1984). Inbreeding effect on Mortality: II. Analysis of a third survey including and excluding infant-juvenile mortality among Brazilian Whites and Negroes. *American Journal of Medical Genetics* **18**: 387-391.
- Elliott, A. M., Graham, G. E., Bernstein, M., Mazer, B. and Teebi, A. S. (1999). Dyskeratosis congenita: an autosomal recessive variant. *American Journal of Medical Genetics* **83**: 178-182.
- Ellis, N. A., Ciocci, S., Proytcheva, M., Lennon, D., Groden, J. and German, J. (1998). The Ashkenazic Jewish Bloom syndrome mutation blmAsh is present in non-Jewish Americans of Spanish ancestry. *American Journal of Human Genetics* **63**: 1685-1693.
- El-Mugamer, I. T., Ali-Zayat, A. S., Hossain, M. M. and Pugh, R. N. (1995). Diabetes, obesity and hypertension in urban and rural people of Bedouin origin in the United Arab Emirates. *Journal of Tropical Medicine and Hygiene* **98**: 407-415.
- El-Shanti, H. E., Daoud, A. S. and Batieha, A. (1999). A clinical study of a large inbred kindred with pure familial spastic paraplegia. *Brain Development* **21**: 478-482.
- Engel-Yeger, B., Zaaroura, S., Zlotogora, J., Shalev, S., Hujeirat, Y., Carrasquillo, M., Barges, S. and Pratt, H. (2002). The effects of a connexin 26 mutation--35delG--on oto-acoustic emissions and brainstem evoked potentials: homozygotes and carriers. *Hearing Research* **163**: 93-100.
- Engel-Yeger, B., Zaaroura, S., Zlotogora, J., Shalev, S., Hujeirat, Y., Carrasquillo, M., Saleh, B. and Pratt, H. (2003). Otoacoustic emissions and brainstem evoked potentials in compound carriers of connexin 26 mutations. *Hearing Research* **175**: 140-151.
- Ercal, D. and Say, B. (1998). Cerebro-oculo-nasal syndrome: another case and review of the literature. *Clinical Dysmorphology* **7**: 139-141.
- Ergin, S., Erdogan, B. S. and Aktan, S. (2003). Mal de Meleda: a new geographical localization in Anatolia. *Dermatology* **206**: 124-130.

- Ermisch, B., Gross, O., Netzer, K. O., Weber, M., Brandis, M. and Zimmerhackl, L. B. (2000). Sporadic case of X-chromosomal Alport syndrome in a consanguineous family. *Pediatric Nephrology* **14**: 758-761.
- Eroglu, G. E. and Kohler, P. F. (2002). Familial systemic lupus erythematosus: the role of genetic and environmental factors. *Annals of Rheumatic Disease* **61**: 29-31.
- Eskandarani, H. A. (2002). Cystic fibrosis transmembrane regulator gene mutations in Bahrain. *Journal of Tropical Pediatrics* **48**: 348-350.
- Eswar, N., Begum, S. R., Sivanandhy, G. and Eswar, P. (2003). Association of parental consanguinity prevalent among those patients having dental anomalies. A retrospective study of 42 cases. *Indian Journal of Dental Research* **14**: 33-38.
- Eymard, B., Laforet, P., Tome, F. M., Collin, H., Leroy, J. P., Hauw, J. J., Richard, I., Beckmann, J., *et al.* (2000). [Miyoshi distal myopathy: specific signs and incidence]. *Revue Neurologique (Paris)* **156**: 161-168 [In French].
- Fageeh, N. A. (2003). Prospective study of hearing loss in schools for deaf children in Assir region, Saudi Arabia. *West African Journal of Medicine* **22**: 321-323.
- Fagerheim, T., Raeymaekers, P., Merren, J., Mani, K., Jha, G. K., Baumbach, L., Brox, V., Breines, E., *et al.* (1999). Homozygosity mapping to the USH2A locus in two isolated populations. *Journal of Medical Genetics* **36**: 144-147.
- Faivre, L., Le Merrer, M., Lyonnet, S., Plauchu, H., Dagonneau, N., Campos-Xavier, A. B., Attia-Sobol, J., Verloes, A., *et al.* (2002). Clinical and genetic heterogeneity of Seckel syndrome. *American Journal of Medical Genetics* **112**: 379-383.
- Faivre, L., Le Merrer, M., Megarbane, A., Gilbert, B., Mortier, G., Cusin, V., Munnich, A., Maroteaux, P., *et al.* (2000). Exclusion of chromosome 9 helps to identify mild variants of acromesomelic dysplasia Maroteaux type. *Journal of Medical Genetics* **37**: 52-54.
- Faivre, L., Megarbane, A., Alswaid, A., Zylberberg, L., Aldohayan, N., Campos-Xavier, B., Bacq, D., Legeai-Mallet, L., *et al.* (2002). Homozygosity mapping of a Weill-Marchesani syndrome locus to chromosome 19p13.3-p13.2. *Human Genetics* **110**: 366-370.
- Faivre, L., Megarbane, A., Alswaid, A., Zylberberg, L., Aldohayan, N., Campos-Xavier, B., Bacq, D., Legeai-Mallet, L., *et al.* (2002). Homozygosity mapping of a Weill-Marchesani syndrome locus to chromosome 19p13.3-p13.2. *Human Genetics* **110**: 366-370.
- Faiyaz-Ul-Haque, M., Ahmad, W., Zaidi, S. H., Haque, S., Teebi, A. S., Ahmad, M., Cohn, D. H. and Tsui, L. C. (2002). Mutation in the cartilage-derived morphogenetic protein-1 (CDMP1) gene in a kindred affected with fibular



- hypoplasia and complex brachydactyly (DuPan syndrome). *Clinical Genetics* **61**: 454-458.
- Faiyaz-Ul-Haque, M., Zaidi, S. H., Al-Ali, M., Al-Mureikhi, M. S., Kennedy, S., Al-Thani, G., Tsui, L. C. and Teebi, A. S. (2004). A novel missense mutation in the galactosyltransferase-I (B4GALT7) gene in a family exhibiting facioskeletal anomalies and Ehlers-Danlos syndrome resembling the progeroid type. *American Journal of Medical Genetics* **128A**: 39-45.
- Farag, T. I. and Teebi, A. S. (1988). Bardet-Biedl and Laurence-Moon syndrome in a mixed Arab population. *Clinical Genetics* **33**: 78-82.
- Farag, T. I. and Teebi, A. S. (1989). High incidence of Bardet Biedl syndrome among the Bedouin. *Clinical Genetics* **36**: 463-465.
- Farag, T. I. and Teebi, A. S. (1997). Genetic disorders among the Bedouins, In *Genetic Disorders among Arab Populations* (eds. Teebi, A. S. and Farag, T. I.) 375-410 (Oxford University Press, New York).
- Fardeau, M., Hillaire, D., Mignard, C., Feingold, N., Feingold, J., Mignard, D., de Ubeda, B., Collin, H., *et al.* (1996). Juvenile limb-girdle muscular dystrophy. Clinical, histopathological and genetic data from a small community living in the Réunion Island. *Brain* **119**: 295-308.
- Farooqi, I. S., Yeo, G. S., Keogh, J. M., Aminian, S., Jebb, S. A., Butler, G., Cheetham, T. and O'Rahilly, S. (2000). Dominant and recessive inheritance of morbid obesity associated with melanocortin 4 receptor deficiency. *Journal of Clinical Investigation* **106**: 271-279.
- Fatinni, Y., Asindi, A., Al Falki, Y., Al Harthi, A., Al Fifi, S. and Al-Daama, S. (2001). Possible new autosomal recessive syndrome of congenital lymphoedema, nail dystrophy and esotropia in a Saudi family. *Acta Paediatrica* **90**: 151-153.
- Faure, S., Bordelais, I., Marquette, C., Rittey, C., Campos-Castello, J., Goutieres, F., Ponsot, G., Weissenbach, J., *et al.* (1999). Aicardi-Goutieres syndrome: monogenic recessive disease, genetically heterogeneous disease, or multifactorial disease? *Clinical Genetics* **56**: 149-153.
- Favero, R., Rizzo, F., Baccetti, B. and Piomboni, P. (1999). Embryo development, pregnancy and twin delivery after microinjection of 'stump' spermatozoa. *Andrologia* **31**: 335-338.
- Feinmesser, M. and Bauberger, T. L. (1966). Consanguinity among parents of deaf children in the Jewish population in Israel. *Journal of Laryngology and Otology* **80**: 1253-1256.
- Feinmesser, M., Tell, L. and Levi, H. (1989). Consanguinity among parents of hearing-impaired children in relation to ethnic groups in the Jewish population of Jerusalem. *Audiology* **28**: 268-271.

- Feldmann, D., Alessandri, J. L. and Deschenes, G. (1998). Large deletion of the 5' end of the ROMK1 gene causes antenatal Bartter syndrome. *Journal of the American Society of Nephrology* **9**: 2357-2359.
- Fellin, R., Zuliani, G., Arca, M., Pintus, P., Pacifico, A., Montali, A., Corsini, A. and Maioli, M. (2003). Clinical and biochemical characterisation of patients with autosomal recessive hypercholesterolemia (ARH). *Nutrition, Metabolism and Cardiovascular Disease* **13**: 278-286.
- Fellowes, A. P., Brennan, S. O., Holme, R., Stormorken, H., Brosstad, F. R. and George, P. M. (2000). Homozygous truncation of the fibrinogen A alpha chain within the coiled coil causes congenital afibrinogenemia. *Blood* **96**: 773-775.
- Fenske, C. D., Jeffery, S., Weber, J. L., Houlston, R. S., Leonard, J. V. and Lee, P. J. (1998). Localisation of the gene for glycogen storage disease type 1C by homozygosity mapping to 11q. *Journal of Medical Genetics* **35**: 269-272.
- Ferák, V., Gécik, A. and Gécikova, A. (1982). Population genetic aspects of primary congenital glaucoma II. Fitness, parental consanguinity, founder effect. *Human Genetics* **61**: 198-200.
- Ferreiro, A., Monnier, N., Romero, N. B., Leroy, J. P., Bonnemann, C., Haenggeli, C. A., Straub, V., Voss, W. D., *et al.* (2002). A recessive form of central core disease, transiently presenting as multi-minicore disease, is associated with a homozygous mutation in the ryanodine receptor type 1 gene. *Annals of Neurology* **51**: 750-759.
- Figuera, L. E., Pandolfo, M., Dunne, P. W., Cantu, J. M. and Patel, P. I. (1995). Mapping of the congenital generalized hypertrichosis locus to chromosome Xq24-q27.1. *Nature Genetics* **10**: 203-207.
- Finckh, U., Kohlschutter, A., Schafer, H., Sperhake, K., Colombo, J. P. and Gal, A. (1998). Prenatal diagnosis of carbamoyl phosphate synthetase I deficiency by identification of a missense mutation in CPS1. *Human Mutation* **12**: 206-211.
- Finckh, U., Xu, S., Kumaramanickavel, G., Schurmann, M., Mukkadan, J. K., Fernandez, S. T., John, S., Weber, J. L., *et al.* (1998). Homozygosity mapping of autosomal recessive retinitis pigmentosa locus (RP22) on chromosome 16p12.1-p12.3. *Genomics* **48**: 341-345.
- Fischer, J., Bouadjar, B., Heilig, R., Fizames, C., Prud'homme, J. F. and Weissenbach, J. (1998). Genetic linkage of Meleda disease to chromosome 8qter. *European Journal of Human Genetics* **6**: 542-547.
- Fischer, J., Faure, A., Bouadjar, B., Blanchet-Bardon, C., Karaduman, A., Thomas, I., Emre, S., Cure, S., *et al.* (2000). Two new loci for autosomal recessive ichthyosis on chromosomes 3p21 and 19p12-q12 and evidence for further genetic heterogeneity. *American Journal of Human Genetics* **66**: 904-913.

- Fischer, R. R., Lerner, C., Bandinelli, E., Fonseca, A. S. K. and Roisenberg, I. (1989). Inheritance and prevalence of von Willebrand's disease severe form in a Brazilian population. *Journal of Inherited Metabolic Disease* **12**: 293-301.
- Fitzgerald, L. F. (2000). Familial brainstem glioma. *Clinical Neurology and Neurosurgery* **102**: 106-108.
- FitzPatrick, D. R., Hill, A., Tolmie, J. L., Thorburn, D. R. and Christodoulou, J. (1999). The molecular basis of malonyl-CoA decarboxylase deficiency. *American Journal of Human Genetics* **65**: 318-326.
- FitzSimmons, J., Tunis, S., Jackson, D., Wapner, R. J. and Jackson, L. (1984). Factors related to subsequent reproductive outcome in couples with repeated pregnancy loss. *American Journal of Medical Genetics* **18**: 407-411.
- Flanigan, K. M., Kerr, L., Bromberg, M. B., Leonard, C., Tsuruda, J., Zhang, P., Gonzalez-Gomez, I., Cohn, R., *et al.* (2000). Congenital muscular dystrophy with rigid spine syndrome: a clinical, pathological, radiological, and genetic study. *Annals of Neurology* **47**: 152-161.
- Fluck, C., Deladoey, J., Rutishauser, K., Eble, A., Marti, U., Wu, W. and Mullis, P. E. (1998). Phenotypic variability in familial combined pituitary hormone deficiency caused by a PROP1 gene mutation resulting in the substitution of Arg-->Cys at codon 120 (R120C). *Journal of Clinical Endocrinology and Metabolism* **83**: 3727-3734.
- Flynn, M. P., Martin, M. C., Moore, P. T., Stafford, J. A., Fleming, G. A. and Phang, J. M. (1989). Type II hyperprolinaemia in a pedigree of Irish travellers (nomads). *Archives of Disease in Childhood* **64**: 1699-1707.
- Foncin, J. F., Bruni, A. C. and Montesi, M. P. (1995). Detection of homozygosity by descent [letter]. *European Journal of Human Genetics* **3**: 203-205.
- Foulds, N., Fairhurst, J., Temple, I. K., Cade, S., Groves, C. and Lancaster, T. (2003). A female case of Sedaghatian type spondylometaphyseal dysplasia. *American Journal of Medical Genetics* **118A**: 377-381.
- Francannet, C., Lefrancois, P., Dechelotte, R., Robert, E., Malpeuch, G. and Robert, J. M. (1990). Fraser syndrome with renal agenesis in two consanguineous Turkish families. *American Journal of Medical Genetics* **36**: 477-479.
- Franceschini, P., Licata, D., Di Cara, G., Guala, A., Bianchi, M., Ingrosso, G. and Franceschini, D. (1999). Bladder carcinoma in Costello syndrome: report on a patient born to consanguineous parents and review. *American Journal of Medical Genetics* **86**: 174-179.
- Franceschini, P., Testa, A., Bogetti, G., Girardo, E., Guala, A., Lopez-Bell, G., Buzio, G., Ferrario, E., *et al.* (1992). Kenn-Caffey syndrome in 2 sibs born to consanguineous parents - evidence for an autosomal recessive variant. *American Journal of Medical Genetics* **42**: 112-116.

- Fraser, F. C. and Biddle, C. J. (1976). Estimating the risks for offspring of first cousin matings. *American Journal of Human Genetics* **28**: 522-526.
- Freire-Maia, N. and Elisbão, T. (1984). Inbreeding effect on morbidity III A review of the world literature. *American Journal of Medical Genetics* **18**: 391-400.
- Frey, J. B. (1995). What dwarfed Toulouse-Lautrec? *Nature Genetics* **10**: 128-30.
- Fried, K. and Kaufman, S. (1980). Congenital afibrinogenemia in 10 offspring of uncle-niece marriages. *Clinical Genetics* **17**: 223-227.
- Frydman, M., Bonn -Tamir, B., Braude, E., Zamir, R. and Creter, D. (1986). Male fertility in factor XIII deficiency. *Fertility and Sterility* **45**: 729-731.
- Frydman, M., Etzioni, A., Eidlitz-Markus, T., Avidor, I., Varsano, I., Shechter, Y., Orlin, J. B. and Gershoni-Baruch, R. (1992). Ramban-Hasharon syndrome of psychomotor retardation, short stature, defective neutrophil motility, and Bombay phenotype. *American Journal of Medical Genetics* **44**: 297-302.
- Fryns, J. P., Dereymaeker, A. M., Heremans, G., Marien, J., van Hauwaert, J., Turner, G., Hockey, A. and van den Berghe, H. (1988). Oculocerebral syndrome with hypopigmentation (Cross syndrome). Report of two siblings born to consanguineous parents. *Clinical Genetics* **34**: 81-84.
- Fryns, J. P., Dumoulin, M. and Hens, G. (1999). Progeroid syndrome with facial teleangiectatic erythema, posterior subcapsular cataracts, calcification of basal ganglia and atrium septum defect type 2. *Genetic Counseling* **10**: 395-398.
- Fujiki, K., Nakayasu, K. and Kanai, A. (2001). Corneal dystrophies in Japan. *Journal of Human Genetics* **46**: 431-435.
- Fukushima, K., Ramesh, A., Srisailapathy, C. R., Ni, L., Chen, A., O'Neill, M. and et al (1995). Consanguineous nuclear families used to identify a new locus for recessive non-syndromic hearing loss on 14q. *Human Molecular Genetics* **4**: 1643-1648.
- Fukushima, K., Ramesh, A., Srisailapathy, C. R., Ni, L., Wayne, S., O'Neill, M. E., Van-Camp, G., Coucke, P., et al. (1995). An autosomal recessive nonsyndromic form of sensorineural hearing loss maps to 3p-DFNB6. *Genome Research* **5**: 305-308.
- Fukuyama, Y. and Ohsawa, M. (1984). A genetic study of the Fukuyama type congenital muscular dystrophy. *Brain Development* **6**: 373-390.