

Clinical Outcomes, Genetic Counselling, Prenatal Diagnosis, Screening

- Abdel-Meguid, N., Zaki, M. S. and Hammad, S. A. (2000). Premarital genetic investigations: effect of genetic counselling. *Eastern Mediterranean Health Journal* **6**: 652-660.
- Ahmed, S., Saleem, M., Modell, B. and Petrou, M. (2002). Screening extended families for genetic hemoglobin disorders in Pakistan. *New England Journal of Medicine* **347**: 1162-1168.
- Albar, M. A. (2002). Ethical considerations in the prevention and management of genetic disorders with special emphasis on religious considerations. *Saudi Medical Journal* **23**: 627-632.
- Alkuraya, F. S. and Kilani, R. A. (2001). Attitude of Saudi families affected with hemoglobinopathies towards prenatal screening and abortion and the influence of religious ruling (Fatwa). *Prenatal Diagnosis* **21**: 448-451.
- 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.
- Aslan, H., Gul, A., Polat, I., Mutaf, C., Agar, M. and Ceylan, Y. (2002). Prenatal diagnosis of Neu-Laxova syndrome: a case report. *BMC Pregnancy and Childbirth* **2**: 1.
- Aslan, H., Ulker, V., Gulcan, E. M., Numanoglu, C., Gul, A., Agar, M. and Ark, H. C. (2002). Prenatal diagnosis of Joubert syndrome: a case report. *Prenatal Diagnosis* **22**: 13-16.
- Auslander, R., Nevo, O., Diukman, R., Morrad, E., Bardicef, M. and Abramovici, H. (1999). Johanson-Blizzard syndrome: a prenatal ultrasonographic diagnosis. *Ultrasound in Obstetrics & Gynecology* **13**: 450-452.
- Aynaci, F. M., Aynaci, O., Ahmetoglu, A. and Celep, F. (2001). Fuhrmann syndrome associated with cortical dysplasia. *Genetic Counseling* **12**: 49-54.
- Bahakim, H. M. (1987). Muslim parents' perception of and attitudes towards cancer. *Annals of Tropical Paediatrics* **7**: 22-26.
- Balci, S., Altinok, G., Ozaltin, F., Aktas, D., Niron, E. A. and Onol, B. (1999). Laryngeal atresia presenting as fetal ascites, oligohydramnios and lung appearance mimicking cystic adenomatoid malformation in a 25-week-old fetus with Fraser syndrome. *Prenatal Diagnosis* **19**: 856-858.
- Balci, S., Bostanoglu, S., Altinok, G. and Beksac, M. S. (2003). Early prenatal diagnosis of familial intestinal polyatresia (FIPA) in a 19 weeks old fetus with sonographic and postmortem findings. *Genetic Counselling* **14**: 373-377.

- Balci, S., Bostanoglu, S., Altinok, G. and Ozaltin, F. (2000). New syndrome?: Three sibs diagnosed prenatally with situs inversus totalis, renal and pancreatic dysplasia, and cysts. *American Journal of Medical Genetics* **90**: 185-187.
- Balci, S., Guler, G., Kale, G., Soylemezoglu, F. and Besim, A. (1999). Mohr syndrome in two sisters: prenatal diagnosis in a 22-week-old fetus with post-mortem findings in both. *Prenatal Diagnosis* **19**: 827-831.
- Becker, R., Kunze, J., Horn, D., Gasiorek-Wiens, A., Entezami, M., Rossi, R., Guschmann, M. and Sarioglu, N. (2002). Autosomal recessive type of Adams-Oliver syndrome: prenatal diagnosis. *Ultrasound Obstetrics and Gynecology* **20**: 506-510.
- Bennett, R. L., Hudgins, L., Smith, C. O. and Motulsky, A. G. (1999). Inconsistencies in genetic counseling and screening for consanguineous couples and their offspring: the need for practice guidelines. *Genetics in Medicine* **1**: 286-292.
- Bennett, R. L., Motulsky, A. G., Bittles, A. H., Hudgins, L., Uhrich, S., Lochner Doyle, D., Silvey, K., Scott, R., *et al.* (2002). Genetic counseling and screening of consanguineous couples and their offspring: recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling* **11**: 97-119.
- Bitoun, E., Bodemer, C., Amiel, J., de Prost, Y., Stoll, C., Calvas, P. and Hovnanian, A. (2002). Prenatal diagnosis of a lethal form of Netherton syndrome by SPINK5 mutation analysis. *Prenatal Diagnosis* **22**: 121-126.
- Bittles, A. H. (1994). The role and significance of consanguinity as a demographic variable. *Population and Development Review* **20**: 561-584.
- Bonaiti, C. (1978). Genetic counselling of consanguineous families. *Journal of Medical Genetics* **15**: 109-112.
- Bonaiti, C., Demenais, F., Briard, M.-L. and Feingold, J. (1978). Consanguinity in malfactorial inheritance. Application to data on congenital glaucoma. *Human Heredity* **28**: 361-371.
- Bonneau, D., Marechaud, M., Odent, S., Piegay, I., Godard, A. and Amati, P. (1999). Heterotaxy-neural tube defect and holoprosencephaly occurring independently in two sib fetuses. *American Journal of Medical Genetics* **84**: 373-376.
- Brady, T. B., Kramer, R. L., Qureshi, F., Feldman, B., Kupsy, W. J., Johnson, M. P. and Evans, M. I. (1999). Ontogeny of recurrent hydrocephalus: presentation in three fetuses in one consanguineous family. *Fetal Diagnosis and Therapy* **14**: 198-200.
- Braga, M. C., Otto, P. A. and Frota-Pessoa, O. (2000). Calculation of recurrence risks for heterogeneous genetic disorders. *American Journal of Medical Genetics* **95**: 36-42.

- Briard, M. L., Frezal, J., Feingold, J. and Kaplan, J. (1979). Genetic counselling in consanguineous marriages. *Journal de Génétique Humaine* **27**: 175-188.
- Briard, M. L., Kaplan, J., Aymé, S., Dodinval, P., Hayez, F., Lamarec, B., Nivelonchevallier, A., Plauchu, H., *et al.* (1981). Consanguine marriages in genetic consultation. *Journal de Génétique Humaine* **29**: 337-348.
- Cao, A. and Galanello, R. (2002). Effect of consanguinity on screening for thalassemia. *New England Journal of Medicine* **347**: 1200-1202.
- Cherif, A., Oueslati, B., Marrakchi, Z., Chaouachi, S., Chaabouni, M., Abassi, M., Dammak, M., Chaabouni, H., *et al.* (2003). [Diastematomyelia: antenatal diagnosis with successful outcome, two cases] *Journal de Gynecologie, Obstetrique et Biologie de la Reproduction (Paris)* **32**: 476-480 [In French].
- Cruz-Coke, R. (1982). Monogram for estimating specific consanguinity risks. *Journal of Medical Genetics* **19**: 216-217.
- den Hollander, N. S., Kleijer, W. J., Schoonderwaldt, E. M., Los, F. J., Wladimiroff, J. W. and Niermeijer, M. F. (2000). In-utero diagnosis of mucopolysaccharidosis type VII in a fetus with an enlarged nuchal translucency. *Ultrasound in Obstetrics & Gynecology* **16**: 87-90.
- 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.
- Durr-e-Sabih, Khan, A. N. and Sabih, Z. (2001). Prenatal sonographic diagnosis of Neu-Laxova syndrome. *Journal of Clinical Ultrasound* **29**: 531-534.
- Edwards, J. H. (1989). Familiarity, receptivity and genuine mosaicism. *Annals of Human Genetics* **53**: 33-47.
- El-Badramany, M. H., Farag, T. I., Al-Awadi, S. A. and Teebi, A. S. (1997). Psychosocial and medical aspects of genetic counseling among Arabs: the example of Kuwait, In *Genetic Disorders among Arab Populations* (eds. Teebi, A. S. and Farag, T. I.) 474-486 (Oxford University Press, New York).
- Eshra, D. J. and Dorgham, L. S. (1989). Knowledge and attitudes towards premarital counselling and examination. *Journal of the Egyptian Public Health Association* **64**: 1-15.
- Felderhoff-Muser, U., Brauer, M., Buhner, C., Wagner, M., Hierholzer, J. and Obladen, M. (2001). Familial recurrence of spontaneous fetal intracranial hemorrhage: ultrasonographic diagnosis and postnatal magnetic resonance imaging (MRI). *Ultrasound in Obstetrics & Gynecology* **17**: 248-251.
- Finckh, U., Kohlschutter, A., Schafer, H., Spermhake, 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.

- Freire-Maia, N. (1984). Effects of consanguineous marriages on mortality and precocious mortality; genetic counselling. *American Journal of Medical Genetics* **18**: 401-406.
- Gasser, B., Lindner, V., Dreyfus, M., Feidt, X., Leissner, P., Treisser, A. and Stoll, C. (1998). Prenatal diagnosis of Walker-Warburg syndrome in three sibs. *American Journal of Medical Genetics* **76**: 107-110.
- Ged, C., Ozalla, D., Herrero, C., Lecha, M., Mendez, M., de Verneuil, H. and Mascaro, J. M. (2002). Description of a new mutation in hepatoerythropoietic porphyria and prenatal exclusion of a homozygous fetus. *Archives of Dermatology* **138**: 957-960.
- Golbahar, J., Karamizadeh, Z. and Honardar, Z. (2002). Selective screening of amino acid disorders in the south-west of Iran, Shiraz. *Journal of Inherited Metabolic Disease* **25**: 519-521.
- Gortzak-Uzan, L., Sheiner, E. and Gohar, J. (2000). Prenatal diagnosis of congenital hypophosphatasia in a consanguineous Bedouin couple. A case report. *Journal of Reproductive Medicine* **45**: 588-590.
- Has, R., Ermis, H., Yuksel, A., Ibrahimoglu, L., Yildirim, A., Sezer, H. D. and Basaran, S. (2004). Dandy-walker malformation: a review of 78 cases diagnosed by prenatal sonography. *Fetal Diagnosis and Therapy* **19**: 342-347.
- Hussain, R. (1999). Community perceptions of reasons for preference for consanguineous marriages in Pakistan. *Journal of Biosocial Science* **31**: 449-461.
- Hussain, R. (2002). Lay perceptions of genetic risks attributable to inbreeding in Pakistan. *American Journal of Human Biology* **14**: 264-274.
- Jaber, L., Dolfin, T., Shohat, T., Halpern, G. J., Reish, O. and Fejgin, M. (2000). Prenatal diagnosis for detecting congenital malformations: acceptance among Israeli Arab women. *Israel Medical Association Journal* **2**: 346-350.
- Khlat, M. (1997). Endogamy in the Arab world, In *Genetic Disorders among Arab Populations* (eds. Teebi, A. S. and Farag, T. I.) 63-80 (Oxford University Press, New York).
- Killick, C. J., Barton, C. J., Aslam, S. and Standen, G. (1999). Prenatal diagnosis in factor XIII-A deficiency. *Archives of Disease in Childhood. Fetal and Neonatal Edition* **80**: F238-F239.
- Lewando-Hundt, G., Shoham-Vardi, I., Beckerleg, S., Belmaker, I., Kassem, F. and Jaafar, A. A. (2001). Knowledge, action and resistance: the selective use of prenatal screening among Bedouin women of the Negev, Israel. *Social Science and Medicine* **52**: 561-569.

- 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.
- 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.
- Modell, B. and Darr, A. (2002). Science and society: genetic counselling and customary consanguineous marriage. *Nature Reviews Genetics* **3**: 225-229.
- Murphy, E. A., Rosell, M. I. and Rosell, E. M. (1982). Theory of genetic counselling for rare recessive traits with consanguinity. *American Journal of Medical Genetics* **13**: 163-178.
- Neerman-Arbez, M., Vu, D., Abu-Libdeh, B., Bouchardy, I. and Morris, M. A. (2003). Prenatal diagnosis for congenital afibrinogenemia caused by a novel nonsense mutation in the FGB gene in a Palestinian family. *Blood* **101**: 3492-3494.
- Niemann, S., Zhao, C., Pascu, F., Stahl, U., Aulepp, U., Niswander, L., Weber, J. L. and Muller, U. (2004). Homozygous WNT3 mutation causes tetra-amelia in a large consanguineous family. *American Journal of Human Genetics* **74**: 558-563.
- Panter-Brick, C. (1991). Parental responses to consanguinity and genetic disease in Saudi Arabia. *Social Science and Medicine* **33**: 1295-1302.
- Petrikovsky, B. M., Gross, B., Bialer, M., Solamanzadeh, K. and Simhaee, E. (1997). Prenatal diagnosis of pseudothalidomide syndrome on consecutive pregnancies of a consanguineous couple. *Ultrasound and Obstetric Gynecology* **10**: 425-428.
- Planas, S., Peiro, R., Rubio, R., Villanueva, R., Seres, A. and Carreras, R. (2003). A new report of mesomelic camptomelia, polydactyly and Dandy-Walker complex in siblings. *Prenatal Diagnosis* **23**: 372-374.
- Poulain, P., Odent, S., Maire, I., Milon, J., Proudhon, J. F., Jouan, H. and Le-Marec, B. (1995). Fetal ascites and oligohydramnios: prenatal diagnosis of a sialic acid storage disease (index case). *Prenatal Diagnosis* **15**: 864-867.
- Reardon, W., Lewis, N. and Hughes, H. E. (1993). Consanguinity, cardiac arrest, hearing impairment, and ECG abnormalities: counselling pitfalls in the Romano-Ward syndrome. *Journal of Medical Genetics* **30**: 325-327.
- Roberts, A., Cullen, R. and Bunday, S. (1996). The representation of ethnic minorities at genetics clinics in Birmingham. *Journal of Medical Genetics* **33**: 56-58.
- Sack, J., Feldman, I. and Kaiserman, I. (1998). Congenital hypothyroidism screening in the West Bank: a test case for screening in developing regions. *Hormone Research* **50**: 151-154.

- Saleem, R., Gofin, R., Ben-Neriah, Z. and Boneh, A. (1998). Variables influencing parental perception of inherited metabolic diseases before and after genetic counselling. *Journal of Inherited Metabolic Disease* **21**: 769-780.
- Schuster, V., Seidenspinner, S., Muller, C. and Rempen, A. (1999). Prenatal diagnosis in a family with severe type I plasminogen deficiency, ligneous conjunctivitis and congenital hydrocephalus. *Prenatal Diagnosis* **19**: 483-487.
- Shalata, A., Mandel, H., Dorche, C., Zabet, M. T., Shalev, S., Hageirat, Y., Arieh, D., Ronit, Z., *et al.* (2000). Prenatal diagnosis and carrier detection for molybdenum cofactor deficiency type A in northern Israel using polymorphic DNA markers. *Prenatal Diagnosis* **20**: 7-11.
- Shawky, S., Milaat, W. M., Abalkhail, B. A. and Soliman, N. K. (2001). Effect of maternal education on the rate of childhood handicap. *Saudi Medical Journal* **22**: 39-43.
- Shiloh, S., Reznik, H., Bat-Miriam-Katznelson, M. and Goldman, B. (1995). Pre-marital genetic counselling to consanguineous couples: attitudes, beliefs and decisions among counselled, noncounselled and unrelated couples in Israel. *Social Science and Medicine* **41**: 1301-1310.
- Shivarajan, M. A., Suresh, S., Jagadeesh, S., Lata, S. and Bhat, L. (2003). Second trimester diagnosis of Neu Laxova syndrome. *Prenatal Diagnosis* **23**: 21-24.
- Spence, M. A. and Hodge, S. E. (2000). The "circular" problems of calculating risk: dealing with consanguinity. *Journal of Genetic Counseling* **9**: 179-201.
- Stoll, C., Alembik, Y., Tchomakov, D., Messer, J., Heid, E., Boehm, N., Calvas, P. and Hovnanian, A. (2001). Severe hypernatremic dehydration in an infant with Netherton syndrome. *Genetic Counseling* **12**: 237-243.
- Suri, M., Garrett, C., Winter, R. M., Hall, C. M. and Griffiths, M. (2002). Dysplastic cortical hyperostosis (Kozlowski-Tsuruta syndrome): report of a second case. *Clinical Dysmorphology* **11**: 267-270.
- Thauvin-Robinet, C., Rousseau, T., Laurent, N., Durand, C., Maingueneau, C., Cormier-Daire, V., Sagot, P., Faivre, L., *et al.* (2002). Hypomandibular faciocranial dysostosis in consanguineous parents revealed by ultrasound prenatal diagnosis. *Prenatal Diagnosis* **22**: 710-714.
- Vanlieferinghen, P. H., Borderon, C., Francannet, C. H., Gembara, P. and Dechelotte, P. (2001). Johanson-Blizzard syndrome. a new case with autopsy findings. *Genetic Counseling* **12**: 245-250.
- Verma, I. C. and Bijarnia, S. (2002). The burden of genetic disorders in India and a framework for community control. *Community Genetics* **5**: 192-196.

- Wessels, M. W., den Hollander, N. S. and Willems, P. J. (2003). Mild fetal cerebral ventriculomegaly as a prenatal sonographic marker for Kartagener syndrome. *Prenatal Diagnosis* **23**: 239-242.
- Whitlock, N. V., Turnpenny, P. D., Tuerlings, J. and Ellard, S. (2003). Molecular genetic prenatal diagnosis for a case of autosomal recessive spondylocostal dysostosis. *Prenatal Diagnosis* **23**: 575-579.
- Wolff, G., Müller, C. R. and Grimm, T. (1989). Benign muscular dystrophy: risk calculation in families with consanguinity. *Journal of Medical Genetics* **26**: 299-304.
- Yang, B. Z., Mallory, J. M., Roe, D. S., Brivet, M., Strobel, G. D., Jones, K. M., Ding, J. H. and Roe, C. R. (2001). Carnitine/acylcarnitine translocase deficiency (neonatal phenotype): successful prenatal and postmortem diagnosis associated with a novel mutation in a single family. *Molecular Genetics and Metabolism* **73**: 64-70.
- Zahed, L. and Bou-Dames, J. (1997). Acceptance of first-trimester prenatal diagnosis for the haemoglobinopathies in Lebanon. *Prenatal Diagnosis* **17**: 423-428.
- Zahed, L., Nabulsi, M., Bou-Ghanim, M. and Usta, I. (1999). Acceptance of prenatal diagnosis for genetic disorders in Lebanon. *Prenatal Diagnosis* **19**: 1109-1112.