posterlistings

The number at the left is both the abstract number and the poster board number. Poster board numbers begin with 25. Posters appear in 4 categories (biochemical genetics, clinical genetics, cytogenetics and molecular genetics). Within each category, they are in alphabetical order by name of first author. Asterisks indicate presenting authors.

Friday, March 19

7:15 am–8:30 am

10:30 am–2:00 pm

3:00 pm–5:30 pm

Saturday, March 20

7:30 am–8:30 am

9:45 am–2:00 pm
Poster Session I: Odd numbers
Authors present 1:00–2:00 pm

3:30 pm–6:00 pm

Sunday, March 21

9:50 am–2:00 pm
Poster Session II: Even numbers
Authors present 1:00–2:00 pm

POSTER PRESENTATIONS

Biochemical Genetics

25 Preliminary cytogenetic characterization of an AZT-resistant subclone, clone 5, of the human colon tumor cell line, HCT-15. A.N. Ahsanuddin,* J.W. Darnowski and H.F.L. Mark.

26 Epilepsy, genetics, neurotransmitter amino acids and anticonvulsants. N.A. Janjua,* T. Itano, M. Ando and S. Onishi.

27 Implications of atypical biochemical data in a case of presumed anencephaly. T. Marini,* J. Sullivan, M. Murray, C. Kanaan, T. Boyd and R. Naeem.

28 Mosaicism in Lowe syndrome. R.L. Nussbaum,* B.M. Orrison, K.E.C. Meyers, A. Caputo and R.A. Lewis.

29 Experience with mucolipidosis type IV in Newfoundland. C. Prasad,* D.J. Buckley, C. Pushpanathan, E.J. Ives, E. Goldin and R. Schiffmann.

30 Study of some morphological, physiological and serological parameters in twins. M. Singh,* S.P. Singh and H. Kaur.

31 Heterogeneity of apolipoprotein E polymorphism and its isoforms in five caste groups of Punjab, India. P. Singh,* P.P. Balgir and I.J.S. Bansal.

Clinical Genetics

32 Cole-Carpenter syndrome: a case report. D.J. Amor* and A. Bankier.

33 Prenatal diagnosis of dup(3q) syndrome. S.K. Barrett,* G.S. Kupchik, D. Rosa, S. Haberman, T. Burgess, M.J. Macera and R.S. Verma.

34 Unique ichthyosis/MR syndrome with seizures, severe developmental delay, failure to thrive, mild dysmorphology and eye findings. P.J. Benke,* M. Lallouz, B. Franco, J. Hannanian, M. Munoz and B. Lam.

35 Alpha thalassemia major: counseling for the possibility of long-term survival. J. Bojanowski,* T. Singer, L. Styles, E. Vichinsky, D. Foote and K. Quirolo.

36 A minimally invasive assay detects BRCA 1 germline mutations. T.J. Byrne,* M. Reece, M. Lane, L. Adams and G. Cohn.

37 Limb deformations in oligohydramnios sequence: effects of gestational age and duration of oligohydramnios. C. Christianson,* D. Huff and E. McPherson.

38 Mixoploidy with a Proteus syndrome phenotype. J.A. Claus,* H.A. Farag and B.G. Kousseff.

39 Caudal regression and holoprosencephaly in a fetus of a diabetic mother. R. Einy,* J.C. Ferreira, G.S. Sachs, A. Orda, N.M. Allen and S.J. Gross.

40 Fragile X mutation screening among unselected pregnant women seen for genetic counseling: findings of a pilot study. B. Finucane,* V. Speare, B. Allitto, J. Adelberg, S. Schmidt, E. Simon and N. Blagowidow.

41 Hirschsprung disease in a patient with translocation resulting in a partial trisomy of the long arm of chromosome 1. E.M. Garcia Soto,* B.K. Goodman, G. Thomas and C. Theda.

42 Cerebrovascular accident leading to cortical blindness in a patient with bird-headed dwarfism. M.J. Hajianpour,* A. Jamali, S.H. Sayar, A. Talebian and A.K. Hajianpour.

43 Immune deficiency (IgG2 deficiency), minimal centromeric association and facial dysmorphism. M.J. Hajianpour,* H. Golkari, S.H. Sayar, A. Farhoodi and A.K. Hajianpour.

44 Breast C.A.R.E. Breaks: an innovative cancer genetics education model for health care professionals. H. Hampel,* P. Gregory and J. Graham.

45 Oro-facial-digital syndrome or Joubert syndrome? K. Haug,* S. Khan and R. Konig.

46 Familial posterior urethral valves as a cause for prune-belly. R.J. Hopkin,* M. Walker and J. Stanek.

47 Prenatal diagnosis of a skeletal deformation resulting from leiomyomata uteri. S. Hudson,* J.L. Roberts, E. Clarin and V.M. Anderson.

48 Carrier screening in Gaucher disease: reproductive decision-making in couples with mixed heritage. V. Jansen,* A. Starkman, L. Zajac and R. Wallerstein.

49 Kabuki makeup syndrome in a Caribbean black child from Trinidad. M. Jaquez,* S. Wu and E. Shirazi.

50 Teaching clinical genetics to pediatric residents in an outpatient setting: a proposed model. M. Jaquez,* R. Haun, R. Frye and T. Frazer.

51 Retrospective analysis of patients with overlapping features of Townes-Brocks syndrome and Goldenhar syndrome. C.E. Keegan and B.R. Korf.*

52 Fetal valproate syndrome through the USF Teratogen Information Service. B.G. Kousseff,* S. Sage and G. Jervis.

53 45,X/46,XY mosaicism: clinical management and counseling issues. M.K. Kukolich* and S.L. Martin.

54 Genetics education of health professionals. E.V. Lapham,* J.O. Weiss, C. Kozma, J.L. Benkendorf and M.A. Wilson.

55 Pregnancy termination policies in community hospitals. R.R. Lebel* and G.A. Lofthouse.

56 Ritscher-Schinzel cardio-cranio-cerebellar (3C) syndrome: report of three new cases. M. Leonardi,* B. Wilkes and G.S. Pai.

57 VATER association with female hypospadia. J.L. Lin,* C.J. Chen, J.Y. Lai and M.S. Hwang.

58 Fryns-Soekerman syndrome: case report and review. D.R. McLeod* and C. Farr.

59 Natural history of trisomy 13 in unselected liveborn patients: comparison with the S.O.F.T. data. E. McPherson* and M. Clemens.

60 Long deletions of the Williams syndrome region on chromosome 7 result in more severe mental retardation. C.A. Morris,* C.B. Mervis, B.F. Robinson, M.T. Keating, X. Lu, X. Meng, P. Spallone, T.R. Dennis and A.D. Stock.

61 Parental mosaicism for a point mutation in a type III collagen (COL3A1) allele produces Ehlers-Danlos type IV (EDS4) in heterozygous offspring. C. Nardi,* U. Schwarze, J.A. Bufill, B. Sinha, M. Pepin and P.H. Byers.

62 Clinical, cytogenetic and epidemiological approaches to the genetic heterogeneity of holoprosencephaly: Buenos Aires, 1988-1997. C. Perandones,* C. Haefliger, L. Campora, J.D. Scheifer, M. Torrado, M. Gallego and C.Z. Barreiro.

63 A unique X-linked syndrome characterized by congenital deafness, mild dysmorphic features, mid-phalangeal hypoplasia, hernias and late-onset pancytopenia. E.M. Petty.*

64 Duplication 17p11.2 in two patients: clinical features and molecular cytogenetic findings. C.M. Powell,* A.S. Aylsworth, K.A. Kaiser-Rogers and K.W. Rao.

65 22q13 Deletion syndrome: a genetic basis for neurobehavioral disorders? C. Prasad,* B.N. Chodirker, C. Lee, A.K. Dawson, L.J. Jocelyn and A.E. Chudley.

66 Multifaceted genetics educational program for nursing faculty. C.A. Prows,* K.K. Latta and C. Hetteberg.

67 Heart-hand syndrome II (Tabatznik syndrome): a new case with mild phenotype. F.J. Ramos,* I. Bueno, J.L. Olivares and M. Bueno.

68 Infantile Huntington disease in Mexico's National Institute of Neurology and Neurosurgery. A. Rasmussen,* M.E. Alonso, R. Macias, P. Yescas, A. Ochoa and G. Davila.

69 Value of using gestational age by ultrasonography in Down syndrome and neural tube defects screening. B.S. Reddy* and S. S. Reddy.

70 Carpal tunnel syndrome: familial autosomal inheritance. B.S. Reddy,* S.S. Reddy and E.H. Ryu.

71 Severe amniotic band syndrome with limb body wall complex: ultrasonographic-clinicopathologic correlations. J.L. Roberts,* L. Csury, F. Schneider, H. Cohen and V.M. Anderson.

72 Blunt trauma to the gravid abdomen as a cause of cerebral palsy and epilepsy. R.M. Roberts.*

73 On hiccuping and yawning: why we do it. R.M. Roberts.*

74 An unusual case of Smith-Lemli-Opitz syndrome and first trimester ultrasonographic exclusion in a subsequent pregnancy. R.M. Roberts* and R.I. Kelley.

75 Brain and cranial disruption sequence from amniotic bands: a result of traction from fetal and embryonic swallowing of attached bands. R.M. Roberts* and W. Blackburn.

76 Serum leptin concentration and lipid profiles in Puerto Rican Bardet-Biedl syndrome. A. Santiago-Cornier,* W. Arias, R. Soto, J. Acevedo, S. Carlo, D. Valencia, L. Caban and T. Frazer.

77 Genetic studies in the focal dermal hypoplasia of Goltz syndrome. R.E. Schnur,* L.A. Reed, K.A. Mockridge and M. Gao.

78 Typical "soup kid" facies of Albright hereditary osteodystrophy in early infancy and natural history of the phenotype through old age. L.R. Shapiro* and H. Taska.

79 Prenatal diagnosis of Smith-Lemli-Opitz syndrome via an abnormal maternal serum screen. M.B. Swing,* G. Hirata, K.M. Gibson, R. Steiner and T. Burlingame.

80 De novo unbalanced translocation of chromosomes 3 and 10 and associated phenotype in a twin gestation. M.S. Verp,* C.J. De Ruiter and R. Spiro.

81 New syndromic association of male pseudohermaphroditism with female external genitalia and adrenal hypoplasia congenita suggests additional sex determining gene(s). E. Vilain,* C. Quigley, J. Aisenberg, Y.-H. Zhang, G. Freidenberg, B.-L. Huang and E.R.B. McCabe.

82 Prenatal diagnosis of asymmetric fetal overgrowth: a diagnostic dilemma. V. Vincent,* L. Seaver, A.M. Sanders, B. Allen and J.V. Dacus.

83 Atypical Down syndrome phenotype with translocation trisomy 21. J. Welch,* K. Keppler-Noreuil, H. Major, Q. Qiau, C. Epstein and S. Patil.

84 New variation on an old theme: subtleties of the Smith-Lemli-Opitz syndrome (RSH/SLOS). G. Yoon,* M.M. Nezarati, E. Fung, F.F. Snyder and G.E. Graham.

85 Assessment of service load in clinical genetics practice: report of a survey conducted in the GENES region. R.E. Zinberg,* K. Greendale, E.S. Salsberg, R. Rosenthal, K.A. Pass and the GENES Clinical Services Committee.

Cytogenetics

86 Distal 5q trisomy resulting from an X;5 translocation detected by chromosome painting. D.N. Abuelo,* A.N. Ahsanuddin and H.F.L. Mark.

87 Trisomy 8 in papillary serous carcinoma of the ovary studied by FISH. A. Afify,* M. Samy, C.-L. Sun and H.F.L. Mark.

88 Chromosome painting for diagnosing a 10;11 translocation in a patient with infantile acute lymphoblastic leukemia. D. Alter,* L. Glasser and H.F.L. Mark.

89 Hypotonia and Prader-Willi syndrome in the neonatal period. D. Berube* and R. Gagne.

90 Williams syndrome: analysis by R-bands, G-bands and FISH techniques. D. Berube* and R. Gagne.

91 Prenatal phenotype of 48,XXYY with elevated MSAFP. S.M. Carter,* P.A. Levy, V. Pulijaal and S.J. Gross.

92 A possible centromeric 21/22 translocation as an alternative cause of nondisjunction in trisomy 21. V. Del Castillo,* S. Ramos, B. Molina and S. Frias.

93 Human 27-kDa heat shock protein (hsp27) gene family: chromosomal band assignments and possible involvement in Williams syndrome deletion. T.R. Dennis,* P.A. Spallone, E. Hickey, L.A. Weber, C.A. Morris and A.D. Stock.

94 Mosaicism for duplication of 17q21→qter with lymphedema and normal phenotype. M. Descartes,* L. Baldwin, P. Cosper and A. Carroll.

95 Trisomy 8 in cervical cancer. D. Feldman,* S. Das, H. Kye, C.-L. Sun, M. Samy and H.F.L. Mark.

96 A dermatofibrosarcoma protuberans with complex clonal chromosomal findings and absence of ring chromosomes. G. Hostetter,* J. Freeman and R. Naeem.

97 Molecular cytogenetic characterization of chromosome markers. C.B. Lozzio,* L. Lyall and E. Bamberger.

98 Congenital cardiac myopathy in a baby with an apparently balanced translocation t(7;8)(p21.2;q24.1). C.J. Madahar,* E. Jenkins, R. Coomaralingam, J. Roy and A. Yanza.

99 A FISH study of trisomies 7 and 8 in prostate cancer. H.F.L. Mark,* S. Das, H. Kye, C.-L. Sun, M. Samy and D. Feldman.

100 Identification of 46,XX/46,XY chimerism in an infant with ambiguous genitalia. M.W. McClellan,* J.M. McClellan, K.D. Ball, T.C. Williams, M.L. Johnson and D.T. Rigdon.

101 A small paracentric inversion of chromosome 18, inv(18)(q22.1q23), in a woman with multiple congenital anomalies and mental retardation. G.S. Sekhon,* S. Scheib-Wixted, M.S. Williams, X.T. Reveles and R.J. Leach.

102 Influence of heterochromatin on fetal loss: clinical and counseling aspects. T.V. Shklovskaya,* S.M. Kleyman, M.J. Macera and R.S. Verma.

103 Prenatal diagnosis of a nonfluorescent Y chromosome as characterized by FISH technique. M.N. Silverman,* T. Shklovskaya, M.J. Macera and R.S. Verma.

104 Discordant detection of monosomy 7 by GTG banding and FISH in a patient with Schwachman-Diamond syndrome. R.A. Sokolic,* W. Ferguson and H.F.L. Mark.

105 An unusual case of pseudodicentric Xq and one possible mechanism. J. Sullivan,* M. Murray, A. Ratti, T. Marini and R. Naeem.

106 Mosaicism for a small supernumerary chromosome 22 associated with dysmorphic features and early onset dementia. U. Tantravahi,* D. Abuelo and S.J. Patrick-MacKinnon.

107 Vanishing twin due to an apparent genomic imbalance. R.S. Verma,* M.J. Macera and E.S. Bronstein.

108 A rare interstitial deletion (2)(p11.2p13) in a child with pericentric inversion (2)(p11.2q13) of paternal origin. B.J. White,* F.L. Lacbawan, A. Anguiano, D. Rigdon, K. Ball, G. Bromage, X.J. Yang, M. DiFazio and S.W. Levin.

109 Two patients with mosaic trisomy ring 20. Doing the right test for the wrong reasons. M.S. Williams,* K.D. Josephson, F.S. Edelman, G.S. Sekhon and S. Scheib-Wixted.

Molecular Genetics

110 Molecular analysis of SRY gene in patients with mixed gonadal dysgenesis. F. Alvarez-Nava,* R. Ortiz, A. Rojas, A. Revol, I. Martinez, S. Martinez, M. Soto, L. Borjas and H. Barrera.

111 Molecular analysis in true hermaphroditism. F. Alvarez-Nava,* R. Ortiz, A. Rojas, M. Soto, L. Borjas and H. Barrera.

112 Prevalence of common mutations of the MTHFR gene in a Puerto Rican population. M.A. Ayala-Rivera,* J. Renta, I. Garcia, L. Garcia, A. de La Vega, P.J. Santiago-Borrero and C.L. Cadilla.

113 Incomplete X-linked congenital stationary night blindness: characterization of mutations in the *CACNA1F* gene and an assessment of clinical variability. K.M. Boycott,* W.G. Pearce and N.T. Bech-Hansen.

114 New point mutation in the *RET* oncogene among an African American kindred with MEN II-A in Puerto Rico. C.L. Cadilla,* G. Vazquez, A. Alcantara, J.Y. Renta and F. Aguilo.

115 Fragile X syndrome in patients with mental retardation of unknown cause in Mexico. A. Gonzalez-del Angel,* S. Vidal, Y. Saldana, V. Del Castillo and L. Orozco.

116 Mutation analysis of the fragile histidine triad gene transcripts of primary tumors and unaffected tissue using restriction nuclease fingerprinting and sequencing. M. Kaelbling.*

117 Molecular analysis of human *Jagged1* gene in an Indian family with Alagille syndrome. P.S. Lai,* F.S.H. Cheah, M.H. Liew, M.M. Aw and S.H. Quak.

118 Hemoglobin S haplotypes among sickle cell patients in the Puerto Rican population. C.R. Lopez,* A. Rodriguez, E. Rivera-Caragol, P.J. Santiago-Borrero and C.L. Cadilla.

119 New gene for dyslexia (DYX3) is located on chromosome 2. H.A. Lubs,* R. Raeymaekers, F.E. Toennessen, M. Pedersen, L. Tranebjaerg and T. Fagerheim.

120 Characterization of the Hermansky-Pudlak syndrome in the Puerto Rican population. A.E. Maldonado-Valentin,* P.J. Santiago-Borrero, A. Gonzalez, R.A. Spritz, J. Oh and C. Cadilla.

121 Reason for nonspecific background of methodologies for random mutation identification: χ-structure formation in purified PCR products solutions. A.A. Neschastnova,* M.G. Yakubovskaya, Z. Lipatova, V.I. Popenko and G.A. Belitsky.

122 Towards the implementation of population-based genetic hemochromatosis screening in Germany. O. Schoffski, J. Schmidtke and M. Stuhrmann.*

123 Prenatal diagnosis of FGFR3 mutations in thanatophoric dysplasia types I and II. E. Spector,* A. Hansen-Higa and G. Bellus.

124 PCR-based molecular diagnosis of Prader-Willi and Angelman syndromes using restriction analysis after bisulfite treatment: potential for quantitative estimation. M. Velinov,* N. Zhong, W.T. Brown and E. Jenkins.

125 Epidermal growth factor's role on human hepatocellular carcinoma transplanted into nude mice. X. Zhao,* S.L. Zhu and T.X. Wang.