# posterlistings

Each number is both the abstract number and the poster board number. Poster board numbers begin with 25. Posters are grouped in 7 categories:

Biochemical Genetics	25-33
Clinical Genetics	34-94
Cytogenetics	95-118
Education and Public Health	119-129
Genetic Counseling	130-140
Molecular Genetics	141-160
Perinatal Genetics	161-183

Within each category, posters are in alphabetical order by name of the first (presenting) author.

Exhibits and posters are located in the Oasis 3 Room of the Palm Springs Convention Center. Booths will be staffed and the poster area will be open and accessible as follows:

Friday, March 10: 11:45 am-1:00 pm

Lunch will be sold in Oasis 3.

5:00 pm-7:00 pm: Poster session with

authors

5:00 pm-6:00 pm: Authors at odd-

numbered boards

6:00 pm-7:00 pm: Authors at even-

numbered boards

Saturday, March 11: 7:30 am-8:30 am

Complimentary continental breakfast

in Oasis 3.

11:30 am-1:00 pm

Lunch will be sold in Oasis 3.

Sunday, March 12: 8:00 am-12:00 pm

(posters only, no exhibits)

#### **Biochemical Genetics**

- 25 2-Methylbutyrl-CoA dehydrogenase deficiency: a new inborn error of L-isoleucine metabolism. *Gibson KM*, *Burlingame T*, *Hogema B*, *Jakobs C*, *Schutgens RBH*, *Millington D*, *Roe C*, *Roe D*, *Sweetman L*, *Steiner RD*, *Linck L*, *Pohowalla P*, *Kiss D*, *Sacks M*, *Rinaldo P*, *Vockley J*.
- **26** Neonatal cholestasis: new presentation of X-linked adrenoleukodystrophy. *Gibson WT, Lepage G, Smith K, Moser H, Moser A, Mitchell GA*.
- **27** Riboflavin responsive ethylmalonic encephalopathy in two Korean sibs. *Hahn SH*, *Lee EH*, *Eun BL*, *Rinaldo P*.
- **28** Severe Conradi-Hunermann syndrome (CDPX2) is a phenocopy of peroxisomal rhizomelic chondrodysplasia punctata (RCDP). *Kronn D, Shapiro LR, Kelly R, Braverman N.*
- **29** Methylmalonic aciduria, hyperhomocysteinemia, hematologic and/or neurologic abnormalities in 3 infants born to mothers with asymptomatic B<sub>12</sub> deficiency. Nikkel SM, Mhanni A, Dilling L, Seargeant L, Stobart K, Rosenblatt D, Gorlin IB, Korson MS, Greenberg CR, Prasad C.
- **30** RadioHPLC profiles of acyl-carnitines improve detection of mild glutaric acidemia type II and short chain acyl-CoA dehydrogenase deficiency. *Rhead WJ, Lambert D.*
- **31** Vitamin  $B_{12}$ -responsive methylmalonic aciduria due to a new inborn error of adenosylcobalamin synthesis, *cblH*. *Rosenblatt DS*, *Watkins D*, *Matiaszuk N*.

- **32** Detection of carbohydrate deficient glycoprotein syndrome by capillary electrophoresis. *Taylor HA*.
- **33** Correction of the cystinotic phenotype in cultured cells by an aminoglycoside. *Thoene J, Lemons R.*

#### **Clinical Genetics**

- **34** Familial aortic aneurysms. *Abuelo DN*, *Guo D*, *Cantu A*, *Carmical S*, *Milewicz D*.
- **35** Clinical and molecular diagnosis of Nager syndrome in a preterm infant (27 week gestation): case report. *Ahmed MN*, *Smith W, Koeberl DD, Qumsiyeh M, Lee B, Goldstein R*.
- **36** BRCA1 and BRCA2 mutation analysis in at-risk African-American families: results and implications. *Baumbach L, Gayol L, Scholl T, Basterrechea H, Pfeifer I, Davies J, Perera E, Smith S, Fernando Arena J.*
- **37** Mitochondrial disease and disorders of energy metabolism: a recognizable pattern of systemic disease. *Bay CA*, *Matika GL*, *Del Vecchio MA*.
- **38** FGFR3 mutations K650N and K650Q cause hypochondroplasia. Bellus GA, Garber AT, Bryke CR, Weaver CA, Speiser PW, Webster MK, Donoghue DA, Francomano CA, Spector EB.
- **39** Knowledge and attitudes about carrier testing for hemophilia A among patients and their relatives. Callanan N, Jennings-Grant T, Lakon C, Spinney T, Sorenson JR.

- **40** Unilateral congenital lymphedema with intestinal lymphangiectasia, elevated liver transaminases, and hypopigmentation. *Campbell WM*, *Noel JM*, *Martin LS*.
- **41** Clinical findings in mitochondrial neurogastrointestinal encephalomyopathy syndrome. Chacin JA, Martinez-Basalo C, Pineda L, Gonzalez S, Morales de Machin A, Ramos N, Molina O, Soto I, Cardozo J, Castillo M.
- **42** Lethal Pallister-Killian syndrome and Fryns syndrome: diagnostic considerations. *Clarkson KB, Corning K, Toburen A.*
- **43** Rubinstein-Taybi syndrome with hepatoblastoma. Claus JA, Kousseff BG, Ranells JR, Jervis GA.
- 44 Scapuloiliac dystosis (Kosenow syndrome) spectrum: two additional cases. *Elliott AM, Rimoin DL, Witt DR, Lachman RS.*
- 45 Withdrawn
- **46** Say-Meyer syndrome: a new case with magnetic resonance imaging of the brain, cardiac abnormality and X-linked dominant inheritance pattern. *Gottesman GS*, *Silhavy JA*, *Singh GK*, *Martin DS*.
- **47** Minimal phenotypic findings of Down syndrome in a patient with true trisomy 21. Hajianpour MJ, Hajianpour AK, Sayar H, Manoochehri F, Mackie Ogilvie C.
- **48** Retinitis pigmentosa, growth hormone deficiency and acromelic skeletal dysplasia in two male siblings: possible familial RHYNS syndrome. *Hedera P, Gorski JL*.
- **49** Testing for the Jewish BRCA founder mutations in archived tissue. *Hixon HEC, Scheuner MT*.
- **50** Concordance among monozygotic and dizygotic twins from a population-based sample for self-reported atopic triad, syndrome x, and psychiatric conditions. *Huang WY, Maier W, Murrelle L, Corey LA, Eaves LJ, Shepherd NS.*
- **51** Oculoauriculofrontonasal syndrome in a 9-month-old male. *Ishmael HA*, *Begleiter ML*, *Butler MG*.
- **52** Cervical spine anomalies in Weaver syndrome: a diagnostic clue in adults. *Kelly TE*.
- **53** Evaluation of telemedicine use for clinical genetics services in Iowa. *Keppler-Noreuil KM*, *Welch J*, *Sebille S*, *Grigsby J*, *Zollo S*.

- **54** Identification of mitochondrial mutation (tRNA<sup>lys</sup>) and genetic counseling in a family with MERRF syndrome. *Kim HJ, Park SJ, Ha MJ, Kim CW, Kim JS.*
- **55** Clinical and molecular studies in a large unique family with limb-girdle muscular dystrophy and Paget disease of bone. Kimonis VE, Kovach MJ, Salam A, Leal S, Waggoner B, Davis K, Khardori R, Gelber D.
- **56** Microgastria in the genetic clinic. Kousseff BG.
- **57** New case of hepatic glycogen synthase deficiency: biochemical findings and comparison with reported cases. Laberge AM, Mitchell G, Van de Werve G, Lambert M.
- **58** Autosomal recessive syndrome due to amazing consanguinity. *Lacassie Y, Avegno J, Tilton AH*.
- **59** Ovarian cancer screening in women from hereditary breast/ovarian cancer families. *Laframboise S, Nedelcu R, Murphy KJ, Cole DEC, Rosen B.*
- **60** BRCA1 mutation carrier with three breast primaries and childhood ionizing radiation treatments: possible gene/environment interaction. *Levonian PJ, Williams MS*.
- **61** Ruvalcaba syndrome (a rare progeroid syndrome): new case and review of the literature. *Martinez-Basalo C, Chacin JA, Gonzalez S, Marquez M, Castillo M, Levy-Bercowski D.*
- **62** New genetic syndrome among the Old Order Amish of Smicksburg, PA. *Matika GL, Del Vecchio MA, Bay CA*.
- **63** Unique skeletal dysplasia with cataracts, ataxia, learning disability and mild facial dysmorphism. *McLeod DR*, *Boag G*, *Trussell R*, *Skov C*, *Adams C*.
- **64** Acrorenal-ectodermal dysplasia-lipoatrophic diabetes syndrome with focal segmental glomerulosclerosis. *McPherson E.*
- **65** Anonychia and absence of distal phalanges in a patient with apparently balanced t(17;21)(q24.2;q11.2). *McPherson E, Prosen T, Surti U.*
- **66** Syndrome of ocular, skeletal and abdominal abnormalities. *M'rad R*, *Mazzoul F*, *Belguith N*, *Ben Jemaa L*, *Smaoui N*, *Chaabouni H*.
- **67** Genetic study of two Tunisian Ehlers-Danlos syndrome type VI. M'rad R, Maazoul F, Belguith N, Ben Jemaa L, Smaoui N, Chaabouni H.

- **68** Withdrawn
- **69** Analysis of 70 adult patients referred for genetics evaluation. *Novak S, Williams MS, Levonian PJ, Josephson KD, Williams JL.*
- **70** Utility of both bone marrow and renal transplants in the management of individuals with Schimke immunooseous dysplasia. *Petty EM, Castle VP.*
- **71** Osteopetrosis, progressive sensorineural deafness, glaucoma, alopecia and cardiomyopathy in a 13-year-old female: new syndrome or mild variant of Yunis-Varon syndrome? *Pletcher BA, Suslak L, Carruth SG, Kolor K, Boyd L.*
- **72** Close relationship to Smith-Lemli-Opitz patients positively correlates with an increased incidence of high cholesterol, late-onset diabetes and infertility, and negatively correlates with alcoholism. *Poss AF, Metherall JE, Opitz JM*.
- **73** Circadian rhythm abnormalities of melatonin and haploinsufficiency of *COPS3* in Smith-Magenis syndrome. *Potocki L, Glaze D, Park S-S, Kasork CD, Shaffer LG, Tan D-X, Reiter RJ, Lupski JR.*
- **74** Mutation in the CSB gene in a patient with cerebrooculo-facio-skeletal syndrome. *Powell CM*, *Meira LB*, *Friedberg EC*.
- **75** Craniosynostosis, ectopia lentis and congenital heart defect: further delineation of an autosomal dominant syndrome with reduced penetrance. *Quercia NL, Teebi A.*
- **76** Oral clefting, cartilaginous auricular malformations and other anomalies: a provisionally unique autosomal dominant syndrome. *Roeder ER*, *Ali Khan-Catts Z*, *Fisher JH*, *Daack-Hirsch S*, *Murray JC*, *Curry CJR*.
- **77** Incidence and severity of pain in Stickler and Ehlers-Danlos syndromes. Rose P, Davis J, Magyari T, Levy H, Francomano C.
- **78** Comparison of the Berlin and Gent nosologies in the diagnosis of Marfan syndrome: the NIH experience. Rose P, Levy H, Ahn N, Sponseller P, Magyari T, Davis J, Francomano C.
- **79** Molecular pathogenesis of Schmid metaphyseal chondrodysplasia. Savarirayan R, Freddi S, Keene DR, Rogers JG, Bateman JF.

- **80** Partial monosomy of chromosome 5 in two male siblings: phenotypic correlates. *Schafer IA*, *Robin N*, *Clark B*, *Izumo S*, *Schwartz S*.
- **81** BRCA testing uptake and participation in ovarian cancer prevention in women at risk for an inherited ovarian cancer susceptibility. *Scheuner MT*, *Cheng LS-C*, *Hixon HEC*, *Rotter JI*.
- **82** Marden-Walker syndrome: case report and review. Schweitzer DN, Earl DL, Graham JM Jr.
- **83** Natural history of the Adams-Oliver syndrome: a report of progressive central and peripheral nervous system involvement in a mother and son. *Scribanu N, Foa R.*
- **84** Sudden death in Proteus syndrome. Slavotinek AM, Vacha S, Peters K, Biesecker LG.
- **85** Severe Saethre-Chotzen syndrome in an infant with a complex chromosome rearrangement. *Storm AL, Drumheller TC, Katz SN, Low J, Airheart C, Gripp KW, Curry C.*
- **86** Seckel syndrome phenotype in a live-born with ring 4/monosomy 4 chromosomal mosaicism. *Taylor MRG*, *Sujansky E*.
- **87** Clinical manifestations of NF1 in African-Americans and Caucasians. *Tekin M, Bodurtha J, Korf B, Riccardi V.*
- **88** Chiari I malformation associated with a P250R mutation of FGFR3. Terry L, Allen W, Schaefer F, Jewett T.
- **89** Acampomelic campomelic dysplasia with SOX9 mutation. *Thong MK, Scherer G, Kozlowski K, Haan E, Morris L.*
- **90** Early clinical features of Angelman syndrome in infants with chromosomal deletion of 15q11-q13. *Tsai CH, Taylor M, Siegel-Bartelt J.*
- **91** Spondyloepimetaphyseal dysplasia with multiple joint dislocations. *Unger S, Savarirayan R, Cormier-Daire V, Cohn D, Wilcox W, Lachman RS, Rimoin DL.*
- **92** Infant with trisomy 9 mosaicism and features of CHARGE association. Walker ME, Blough RI, Bove KE, Hopkin RJ.
- **93** Third Prader-Willi syndrome phenotype due to maternal uniparental disomy 15 with mosaic trisomy 15. Wulfsberg EA, Olander E, Stamberg J, Steinberg L.

ternal (segmental) isodisomy for chromosome 14 without a Robertsonian translocation. Yang SP, Towner DR, Sherman MP, Shaffer LG, Johnson JP.

Phenotypic recognition of maternal (mosaic) and pa-

#### Cytogenetics

- Double trisomy in spontaneous abortions: an 11-year review. *Al-Kouatly HB, Johnson C, Skupski D, Lita Alonso M.*
- Case of 46,XX African American male. *Asamoah A, Dev VG, Misra R, Onadeko O, Parsh B, Groening P.*
- **97** Family with multiple chromosome anomalies. Dawson AJ, Riordan D, Vust A, Konkin D, Wicksrom DE, Prasad C, Greenberg CR.
- Can pericentric inversion and C-heterochromatin cause interchromosomal effect leading to an increased aneuploidy in sperm nuclei? *Diukman R, Sardos Albertini F, Fejgin M, Shacham A, Amiel A.*
- Isochromosome 9p and choroid plexus papilloma: coincidence or cause? *Fischer JM*, *Toriello HV*.
- Terminal deletion of 11q in two brothers: clinical, cytogenetic, molecular genetic and counseling issues. *Haag MM*, *Phillips SM*, *Tunby ML*, *Beischel LS*, *McCann CL*, *Hansen JC*, *Johnson JP*, *Reynolds JF*.
- FISH delineation of multiple chromosome abnormalities in a mentally retarded patient with severe chronic disabilities. *Harrison KB, Eddey G, Barabas G, Mintz J.*
- Electronic karyotype transmission. *Harrison KB, Warburton D.*
- Molecular and cytogenetic analysis of Y;1 familial translocation. *Kim JW, Kang IS, Nam SA, Kim YM, Lee YS, Park SY.*
- Chromosomal nosology in referred populations. Kleyman SM, Mizhiritskaya V, Macera MJ, Verma RS.
- Smith-Magenis syndrome diagnosed at birth. *Lozzio C, Ryan T, Bamberger E, Holland E, Carter W.*
- **106** Chromosome analysis of spermatozoa extracted from testes of men with nonobstructive azoospermia. *Martin RH, Greene C, Rademaker A, Barclay L, Ko E, Chernos J.*

- Zeroing in on breakpoint susceptibility regions on chromosomes in breast carcinoma. *Mattoo A, Verma RS.*
- Interphase spectral FISH: tailoring a diagnostic and minimal residual disease assay for oncology. *Murata-Collins JL, Zhang F, Tcheurekdjian L, Slovak ML*.
- Ring chromosome 14 syndrome: prenatal diagnosis of two cases with 45,XY,-14/46,XY,r(14)(p11.2q32). *Schmidt RT*, *Ravnan JB*, *Lamb AN*, *Weinstein ME*.
- **110** Tandem duplication of bands q13.13q13.33 resulting in partial trisomy of long arm of chromosome 19. *Sekhon GS*, *Johnson EB*.
- Maternal age-specific chromosomal abnormalities at amniocentesis. *Shklovskaya T, Verma RS.*
- **112** Severe growth retardation and limb anomalies in a boy with 47,XY,+r(7) and maternal uniparental disomy for chromosome 7. Stadter CS, Stamberg J, Das S, Wulfsberg EA.
- **113** Double and triple trisomy in spontaneous abortions: an older maternal age and earlier gestational age than seen in single trisomies. *Sullivan J, Yusef R, Marini T, Naeem R*.
- Further clinical and cytogenetic delineation in 1p36 deletion syndrome. Vargas FR, Ramos M, Goncalves-Neto JB, Martins RR, Ramos H, Llerena JC Jr, Cabral de Almeida JC.
- Familial report of duplication 9p syndrome. Wenger SL, Borsa V, Holt CD, Hummel M, Mullet M.
- Proximal 6q deletion phenotype: findings in de novo interstitial deletion 6q14.1q15. White BJ, Schwartz AT, Levin SW, Coll EJ, Anguiano A, Wang S, Yang X-J.
- Tetraploidy in prenatal diagnosis: "cultural artifact" or clinical diagnosis? *Winsor EJT, Chitayat D, Skidmore MB*.
- **118** Partial monosomy 12p and ring chromosome 12 mosaicism in a male with developmental delay and mild dysmorphism. *Yee HA*, *Chernos JE*, *Veale PM*, *Clarke ME*, *Graham GE*.

#### **Education and Public Health Genetics**

The genetic counselor as a resource for families with a medical indication for cord blood banking. *Epstein JM*, *Meyers R*, *Reed WF*, *Smith RS*, *Taylor K*, *Haaz S*, *Lubin BH*.

### Poster Listings

- **120** Public acceptance of an interactive kiosk to educate about folic acid to prevent neural tube defects. Jackson DN, Brown L.
- **121** Public's quest for genetic information: the role of a telephone helpline. Lander LE, Kramer EA, Davidson ME, Collins DL.
- **122** Factors affecting the incidence of Down syndrome live births in Illinois from 1989 to 1997. Leonard DR, Pergament E, Schmidt H, Egler T, Shen T.
- 123 The state of public health genetics in Rhode Island. Mark HFL, Caldarone R, Zimmerman A, Viner-Brown S, Simon P, Colt A, Hollinshead W, Nolan P.
- **124** Assessment of genetic knowledge and utilization among mental health care providers and consumers. Petty EM, Madeo AC, Smith LB, Milner KK.
- **125** MCHB SPRANS projects: collaborative efforts to guide primary care providers to access and use medical genetic information electronically. Proud V, Silvey K, Barash C, Pletcher B, Hanson N.
- **126** Ethical issues encountered in establishment of the Texas Birth Defect Research Center. Scheuerle A, Wright D.
- **127** BRCA genetic testing: where are physicians in the decision-making process? Velicer CM, Taplin S.
- **128** Evaluating data from newborn screening programs: Georgia, 1998. Wang SS, Fernhoff PM, Grinzaid K, Ramachandran M, Franko EA, Henson M, Buehler J, Khoury MJ.
- 129 Visual presentations at 1998 meeting suggest insensitivity of geneticists to a common genetic disorder. Williams MS, Williams JL.

## **Genetic Counseling**

- 130 Attitudes regarding genetic counseling issues of the Pakistani population at Maimonides Medical Center in Brooklyn. Barrett SK, Rosa DA, Begum S, Hafeez A, Kupchik GS.
- **131** Patient perspectives on the process of informed consent for DNA testing. Cytrynbaum C, Babul-Hirji R, Rowell M, Henderson K, Australie K, Druker H, Dupuis L, Quercia N, Shuman C, Kennedy S.

- 132 Genetic counseling for mitochondrial disorders. Del Vecchio MA, Matika GL, Bay CA.
- 133 Constructing rapport in televideo genetic counseling. Flore LA, Risinger ST, Britt DW, Zador IE, Gilbert AD, Evans MI, Johnson A.
- 134 Prenatal genetic counseling by telemedicine: a feasibility study. Flore LA, Risinger ST, Britt DW, Zador IE, Gilbert AD, Evans MI, Johnson A.
- **135** Practical use of three-dimensional imaging in genetic counseling: patient perception of usefulness as a counseling tool. Jackson DN, Brown L, Keel-Thompson K.
- **136** Prenatal diagnosis of fragile X syndrome: identification of a male fetus mosaic for a premutation on chorionic villus sampling-management and follow-up. Kennedy SI, Wei C, Steele L, Teebi AS.
- **137** Interactive web-based genetic screening questionnaires in a primary care and obstetrics practice: pilot study. Neidich IA, Taswell C, Daniels K.
- **138** Genetic risk assessment in women over 35: natural pregnancies compared to assisted reproductive technology pregnancies. Pearson M, Neidich J.
- **139** Preconceived ideas about second trimester screening: a guide for counseling. Snow S, Souter V, Luthy D, Nyberg D.
- 140 Low maternal serum estriol as a marker for steroid sulfatase deficiency. Stanislaw CL, Rogers RC, Stewart K, Phelan MC.

#### **Molecular Genetics**

- **141** Sequence characterization of the  $-^{THAI}$  allele of  $\alpha$ thalassemia and rapid detection using a single-tube multiplex PCR assay. Chong SS, Boehm CD, Cutting GR, Higgs DR.
- 142 BRCA buccal immunoassay predicts BRCA1 and BRCA2 mutations. Cohn GM, Byrne TJ, Hoffman DE, Adams LA, Lane MA, Reece MT.
- 143 Variant chromosome 1 reveals centromeric DNA sequences within the 1qh region. Conte RA, Verma RS.

- **144** X-linked corneal dermoid maps to Xq24-Xter. Dar P, Javed AA, Pandita RK, Ben-Yishay M, Spiteri E, Ferreira JC, Gross SJ, Chitayat D, Edelman L, Morrow BE, Nitowsky HM.
- **145** Unexpected female patient within a classical Lesch-Nyhan family. De Gregorio L, Nyhan WL, Serafin E, Chamoles NA.
- **146** Primary ciliary dyskinesia: search for the responsible genes through linkage and candidate gene approaches. DeLozier-Blanchet CD, Bartoloni L, Gehrig C, Radhakrishna U, Meeks M, Duriaux-Sail G, Maiti A, Guerne PA, Walt H, Gardiner RM, Antonarakis SE, Blouin JL.
- **147** Mitochondrial DNA mutations in Chilean patients with Leber hereditary optic neuropathy. Fadic R, Lobos C, Schweitzer M, Luco C.
- **148** Frequency and clinical significance of the S1235R mutation in the cystic fibrosis transmembrane conductance regulator gene: results from a collaborative study. Feldman GL, Monaghan KG, Barbaratto GM, Snow K.
- **149** Genome-wide linkage study for ossification of the posterior longitudinal ligament of the spine reveals a major susceptibility locus on chromosome 21q. Furushima K, Ikari K, Maeda S, Koga H, Takeda J, Harata S, Inoue I.
- **150** Novel nonsense mutation of the GTP cyclohydrolase I gene in a family with dopa-responsive dystonia. Hong KM, Kim YS, Paik MK.
- **151** Molecular analysis of chromosome 6p rearrangement in retinoblastoma. Imbert I, Coignet LJA, Pellestor F.
- **152** Williams syndrome: on the genetic basis of human cognition. Korenberg JR, Chen X-N, Hirota H, Lai Z, Bellugi U, Burian D, Roe B, Matsuoka R.
- 153 Locus for autosomal dominant renal Fanconi syndrome maps to the long arm of chromosome 15. Lichter-Konecki U, Broman KW, Dart R, Blau E, Konecki DS.
- **154** Detection of a de novo mutation in a family with SMA type I: the importance of dosage testing. McGowan-Jordan J, Zeesman S, Whelan DT, Ray PN, Stockely TL, Prior T, Carson NL.
- **155** Association of G-protein  $\beta$ 3 subunit C825T variant  $(G\beta 3S)$  and heart valve abnormalities in obese patients treated with fenfluramine-phentermine. Ning L, Eichelberger JP, Berk BC, Qi M.

- **156** CFTR mutations in Chilean patients with cystic fibrosis. Repetto GM, Flores I, Lobo C, Boza ML, Perez MA, Guiraldes E, Harris P, Sanchez I.
- **157** Withdrawn
- **158** Genetic testing for Niemann-Pick type C disease. *Snow* K, Park WD, Lundquist PA, Walsh Vockley C, Patterson MC, Karnes PS, O'Brien JF.
- **159** HFE 5569A allele defines a low-risk haplotype for hereditary hemochromatosis. Somerville MJ, Sprysak KA, Hicks M, Elyas BG, Vicen-Wyhony L.
- **160** Mohr-Tranebjaerg syndrome is an X-linked recessive disorder characterized by mitochondrial dysfunction associated with neuronal cell death. Tranebjaerg L, Lindal S, Merchant S, Ingebretsen OC, Hamel B, Fung V, Hayes M, Koehler C, Nilssen O. van Ghelue M.

#### **Perinatal Genetics**

- **161** Clinical significance of isolated fetal echogenic bowel. Al-Kouatly HB, Chasen ST, Steltzoff J, Chervenak FA.
- **162** Is an elevated maternal serum  $\mu$ E3 MoM value in the second trimester associated with a poor pregnancy outcome? Aufox SA, Berry MN, Conrad D, Stamper TH, Hart PS.
- **163** Prenatal diagnosis of otocephaly. Brewer R, Dykhuizen R, Sumners J.
- **164** Prune belly syndrome in a patient with only a mildly distended bladder. Brewer R, Menzel M, Sumners J.
- **165** Pattern analysis for ultrasound anomalies in fetuses with normal karyotype. Farina A, Rizzo N, Pilu G, Caramelli E, Carinci P. Bovicelli L.
- **166** Extreme spectrum of Pallister-Hall syndrome. *Greenly* KH, Tyson RW, Tsai CH.
- **167** Evaluation of prenatal screening program for Down syndrome: analysis of 96 cases of Down syndrome for the last 10 years. Han JY, Kim MY, Ahn HK, Yang JH, Ryu HM, Kim JM, Kim YM, Park SY, Han HK, Lee YH, Kim ES.
- **168** Antenatal diagnosis of mixoploidy: a case report. *Jack*son DN, Brown L.

- **169** Cystic lymphangioma of the axilla and chest wall. *Jackson DN*, *Brown L*.
- **170** Congenital mesoblastic nephroma: perinatal, surgical and genetic features and associated 2D and 3D imaging. Jackson DN, Haag M, Brown L, Keel-Thompson K, Phillips SM, Tunby M, Beischel LS, Bennett TL.
- **171** Sonogram findings with Brachmann-de Lange syndrome. *Jervis GA, Kalter CS, Kousseff BG.*
- **172** Recurrence of triploidy in a woman with low level 45,X mosaicism. *Johnson LR*, *Blough RI*, *Miller ME*.
- **173** Characteristics of fetuses with polyhydramnios and abnormal chromosome study. *Lazebnik N*.
- **174** Long-term follow-up of amniocentesis. *Lebel RR*, *Manno MM*.
- **175** Prenatal diagnosis of MIDAS/MLS syndrome associated with a deletion at Xp22.1. Marvin ML, Duquette DA, Hogge WA, Hunter Y, Toriello HV.
- **176** Prenatal and postmortem features of a case of Ritscher-Schinzel syndrome. *Nikkel SM*, *Levi CS*, *Menticoglou S*, *Phillips S*, *Safneck J*, *Chodirker BN*.

- **177** Relationship between the Glu298Asp and intron 4 polymorphisms of endothelial nitric oxide synthase in an Hispanic population with preeclampsia. *Pellicena A, Riskin-Mashiah S, Hefler LA, Gregg AR*.
- **178** Outcomes of a prenatal cytogenetic screening program in an urban state university medical center. *Powell EM, Santolaya-Forgas J, Matheson JKB, Shulman LP.*
- **179** Prominent amnion-chorion separation between 13 and 15 weeks' gestation is associated with increased risk for fetal chromosome abnormalities. *Shulman LP, Patel S, Phillips OP.*
- 180 Withdrawn
- **181** Do X- or Y-chromosome bearing spermatozoa compete with older eggs in humans? *Verma RS*, *Shklovskaya T*, *Baheig SM*.
- **182** Fetal methotrexate syndrome. Wheeler ME, Stanford M, O'Meara P.
- **183** Prevalence of methylenetetrahydrofolate reductase C677T polymorphisms in Northwest Louisiana newborn population. *Yanamandra K, Napper D, Jalanivich DW, Thurmon TF.*