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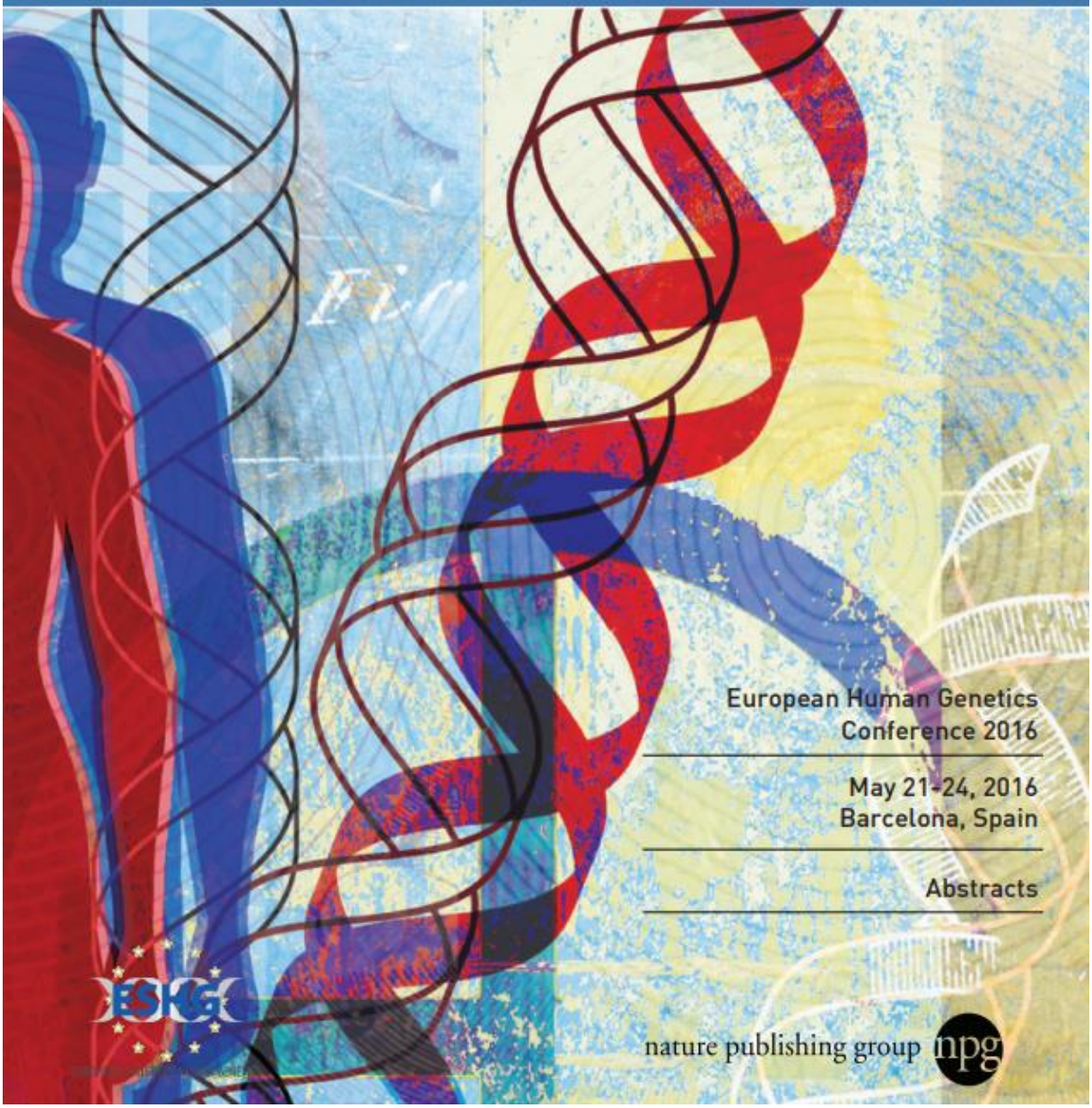

is a powerful ASD research tool since it can assay many types of genetic variation in a single test. In recent years, there has been a lot of debate around return of results following genetic testing; however, there is a paucity of empirical data on research subject's experience with WGS, in particular. To elicit and analyze participants' experiences with WGS, we invited parents (and capable children) who were enrolled in an ASD genomic study to participate in semi-structured interviews at two times during the WGS testing cycle: 1) following informed consent, and 2) following return of results. To date, we have interviewed 19 parents at time 1 and 8 parents at time 2. Emerging themes from time 1 interviews include, hope for a genetic diagnosis, hope for treatment, and altruism. Parents also hoped for findings that they could use for planning and prevention purposes, although they expressed concerns over inflicted insight and insurance discrimination. Following return of results at time 2, emerging themes include, the value of learning genetic information, the resolution of blame between biological parents, and disappointment at not receiving treatment-guiding information. Our findings suggest that participants may have different motivations/concerns before and after testing; whereas many begin with altruistic motivations, their desire for actionable results weighs heavily post testing. Though our analysis is provisional, these results suggest that novel strategies for education and informed consent are necessary.

ELECTRONIC POSTERS

E-P01 Reproductive Genetics/Prenatal Genetics

E-P01.03
Outcome of aneuploidy screen positive test based on amniocentesis results in 100 Iranian pregnant women

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Background: About half of spontaneous abortions are caused by chromosomal abnormalities. While autosomal trisomy is the most common karyotypic anomaly among these, polyploidy, sex chromosome anomalies and structural rearrangement can also be found at high rates in spontaneous miscarriages.

Conclusions: Aneuploidy is the primary cause of the first trimester miscarriages. Cytogenetic studies showed that aneuploidy level can be at 50-80% in certain populations. While autosomal trisomy is the most common karyotypic anomaly among these, polyploidy, sex chromosome anomalies and structural rearrangement can also be found at high rates in spontaneous miscarriages.

E-P01.06
Usefulness of array-CGH technique for genetic counseling regarding Y;autosome translocations in prenatal diagnosis

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frequent ones (45,2%). Positive prenatal screening and advanced maternal age (AMA) were the most common referral reasons with 56.1% and 38.9%. Positive predictive value of ultrasound abnormalities was highest with 5,8% and for increased triple test screening risk and AMA it was 2,3% and 2,7%, respectively. When the karyotype analysis results of amniocentesis according to abnormal and normal karyotype results performed for AMA and increased triple test compared, the difference was statistically not significant ($p>0.05$).

E-P01.05
Aneuploidy findings on two different pregnancy loss materials from the same mother

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Introduction: Early miscarriages are the most common complications of pregnancies and frequency of such cases is about 10-15% of all pregnancies. Endocrinological and anatomical causes, infectious diseases, environmental factors, immunological defects, genetic factors including numerical and structural chromosomal aberrations are the main causes for early miscarriages. The most common karyotypic anomaly in early miscarriages is autosomal trisomy. Chromosomal aberrations play an important role in the etiology of early miscarriages.
Materials and Methods: 37 years old pregnant female patient who has a healthy daughter was admitted to Polyclinic of Gynecology and Obstetrics at Faculty of Medicine in Mersin University for the missed abortion. Abortion materials from two separate pregnancies were sent to laboratory at different times. Samples from the first fetus from the 8th week of the pregnancy and the samples from the second fetus from the 9th week of pregnancy were cultured and metaphase plates are obtained using GTG banding technique. 15 metaphase plates from the first fetus and 20 from the second fetus were cytogenetically examined.
Results: As the result of cytogenetic analysis, 15 cells obtained from the fetal tissue of the first pregnancy showed 47,XX+12 karyotype and 20 cells from the second pregnancy showed 48,XY+16,+22 karyotype.

E-P01.13
A novel TAZ gene mutation detected prenatally in a family with Barth syndrome

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Barth syndrome (OMIM 302060) is a rare X-linked disease characterized by dilated cardiomyopathy, proximal skeletal myopathy and cyclic neutropenia. Barth syndrome is caused by various mutations in the tafazzin (TAZ) gene which causes abnormalities of cardiolipin, an essential mitochondrial phospholipid.
A 22-year-old woman, G1 P0, was referred to our unit at 13 weeks of gestation for genetic counselling because of familial history of cardiomyopathy. Her first brother died at 6 months of age due to cardiac failure. The second