

181

Do X- or Y-chromosome bearing spermatozoa competes with older eggs in human? R.S. Verma, T. Shklovskaya and S.M. Baheig. Division of Molecular Medicine and Genetics, Wyckoff Heights Medical Center, Brooklyn-New York Hospital/Weill Medical College of Cornell University, New York, Institute of Molecular Biology and Genetics and SUNY Health Science Center at Brooklyn, NY

Human spermatozoa pass a torturous journey during fertilization. A slight deviation from 1:1 has been observed in the sex ratio and a number of variables ranging from seasonal, religious days and position of females during mating have been implicated in producing offsprings of a desired gender. The selective choice of sperm by a female based on the aforementioned factors [superstitions] has not been substantiated. In this investigation we collected data from 2,071 pregnant women who visited our two laboratories for genetic amniocentesis during the past two decades. The findings are classified by age group:

Age	XY	XX	XY (%)	XX (%)
20-24	70	46	60.3%	39.7%
25-29	91	112	44.8%	55.2%
30-34	192	205	48.4%	51.6%
35-39	536	540	49.8%	50.2%
>40	146	133	52.3%	47.7%
Total	1035	1036	50.0%	50.0%

The sex ratio remains the same for all age groups except the group of women in the range of 20-24 years. This finding is apparently fortuitous because of the smaller sample size. The concept of sex determination based on a non-chromosomal [X- or Y-chromosome] basis will remain an enigma in the mind of religious orthodoxies as even older eggs did not have any preferential attraction for either X- or Y-chromosome bearing sperm.

183

Prevalence of Methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism in Northwest Louisiana newborn population. K. Yanamandra, D. Napper, D.W. Jananivich, and T.F. Thurmon. Department of Pediatrics, LSU School of Medicine, Shreveport, LA.

The Pediatric Genetics laboratory at LSU School of Medicine in Shreveport, LA, began offering prenatal service in NTD and chromosomal defect screening through triple screening program since 1995. The laboratory also began epidemiological research studies on the newborn population of Northwest Louisiana for several genetic disease mutations for the past 4 years as part of March of Dimes birth defects foundation and LSU foundation grants. We have recently reported the gene frequencies for MCAD deficiency, Factor V Leiden in our population and as part of thrombophilia markers we also started investigating the frequencies of MTHFR C677T and prothrombin G20210A polymorphism. Our local population composed of 76% African-Americans, 20% Caucasians, 2% Hispanics, and the rest other ethnic groups. Following English multicenter multivitamin studies on NTD in 1970 and '72 revealing possible risk reduction in recurrences of NTD with periconceptional supplementation of folic acid, folate-activating enzyme, MTHFR, has become the subject of intense study in various diseases. MTHFR was also linked to cardiovascular diseases through hyperhomocystinemia and recently to other complex genetic traits such as Orofacial clefts.

In our polymorphism studies, the carrier and homozygous C677T frequencies were found to be 24% and 3.5%, respectively, in the total population. However, on stratification by ethnicity, we found that the heterozygote and homozygote frequencies were 18%, 46%, 45%, and 0.9%, 21%, 11%, respectively, in African-Americans, Hispanics and Caucasians. Our data found higher C677T homozygotes in our Hispanic newborn population than other ethnic groups and may explain the higher frequency of NTDs and Orofacial Clefts found in this population.

182

Fetal methotrexate syndrome. M.E. Wheeler¹, M. Stanford², P. O' Meara¹. ¹Denver Health Medical Center, Denver, and ²University of Colorado, Denver.

Fetal methotrexate syndrome was first described several decades ago when this agent was being used as abortifacient. After this first trimester drug exposure was associated with fetal anomalies, this use as abortifacient became rare. Over the last several years the use of methotrexate for termination of pregnancy and medical management of ectopic pregnancies has again become more widespread.

Case: A 23 year old G₃ P₂ female sought an abortion at 8 weeks of gestation. She was given an unknown dose of methotrexate. She had no follow-up until she presented at 10 weeks of gestation. At that time she was noted to have a viable intrauterine gestation. An ultrasound was done that showed a normal scan for 10 weeks of gestation. The patient had no prenatal care until she presented at 39 weeks of gestation. At that time she was noted to have severe preeclampsia and HELLP syndrome. The fetus on ultrasound was symmetrically growth retarded with estimated fetal weight at less than the 5th percentile. Secondary to a breech presentation the infant was delivered by cesarean section. The infant was small for gestational age at 2050grams which is at the first percentile by the Lubcenco intrauterine growth standard. On physical exam the infant was hypotonic with the following anomalies: mild dolichocephaly, high forehead, prominent, broad nose, hypertelorism., small palpebral fissures, sacral crease, small mouth, clinodactyly, and hypoplastic toe nails. The infant's hospital course was complicated by temperature instability. A head CT and ultrasound were negative for any pathology. Chromosomes were normal. After a 7 day hospital course the infant was discharged home with a cardiac and respiratory monitor.

Discussion: Methotrexate have been extensively use in autoimmune disorder and as chemotherapy. Since their use is becoming more extensive as a abortifacient and for the medical management of ectopic pregnancies, we will see many cases of first trimester exposure of fetuses to this agent. This exposure should be considered when readily offered by the mother.