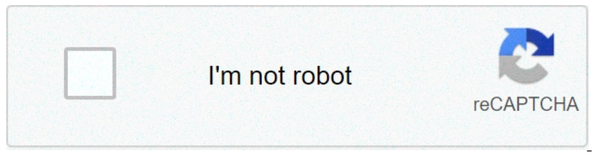

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Abnormalities of the sex chromosomes

Article Title: The epidemiology of sex chromosome abnormalities Authors: Berglund, Stochholm, and Gravholt Date of Publication: May 11, 2020 “Sex chromosome abnormalities (SCAs) are characterized by gain or loss of entire sex chromosomes or parts of sex chromosomes with the best-known syndromes being Turner syndrome, Klinefelter syndrome, 47,XXX syndrome, and 47,XYY syndrome. Since these syndromes were first described more than 60 years ago, several papers have reported on diseases and health related problems, neurocognitive deficits, and social challenges among affected persons. However, the generally increased comorbidity burden with specific comorbidity patterns within and across syndromes as well as early death of affected persons was not recognized until the last couple of decades, where population-based epidemiological studies were undertaken. Moreover, these epidemiological studies provided knowledge of an association between SCAs and a negatively reduced socioeconomic status in terms of education, income, retirement, cohabitation with a partner and parenthood. This review is on the aspects of epidemiology in Turner, Klinefelter, 47,XXX and 47,XYY syndrome.” Read more Open Access Peer-reviewed X&Y chromosomal aneuploidies are among the most common human whole-chromosomal copy number changes, but the population-based incidence and prevalence in the child-bearing population is unclear. This retrospective analysis of prospectively collected data leveraged a routine non-invasive prenatal test (NIPT) using parental genotyping to estimate the population-based incidence of X&Y chromosome variations in this population referred for NIPT (generally due to advanced maternal age). From 141,916 women and 29,336 men, 119 X&Y chromosomal abnormalities (prevalence: 1 in 1,439) were identified. Maternal findings include: 43 cases of 45,X (40 mosaic); 30 cases of 47,XXX (12 mosaic); 3 cases of 46,XX uniparental disomy; 2 cases of 46,XY/46,XX; 23 cases of mosaicism of unknown type; 2 cases of 47,XX,i(X)(q10). Paternal findings include: 2 cases of 47,XXY (1 mosaic); 10 cases of 47,XYY (1 mosaic); 4 partial Y deletions. Single chromosome aneuploidy was present in one of every 1,439 individuals considered in this study, showing 47,XXX; 47,XX,i(X)(q10); 47,XYY; 47,XXY, partial Y deletions, and a high level of mosaicism for 45,X. This expands significantly our understanding of X&Y chromosomal variations and fertility issues, and is critical for families and adults affected by these disorders. This current and extensive information on fertility will be beneficial for genetic counseling on prenatal diagnoses as well as for newly diagnosed postnatal cases. Citation: Samango-Sprouse C, Kırkızlar E, Hall MP, Lawson P, Demko Z, Zneimer SM, et al. (2016) Incidence of X and Y Chromosomal Aneuploidy in a Large Child Bearing Population. PLoS ONE 11(8): e0161045. Osman El-Maarri, University of Bonn, Institute of Experimental Hematology and Transfusion Medicine, GERMANYReceived: March 16, 2016; Accepted: June 23, 2016; Published: August 11, 2016Copyright: © 2016 Samango-Sprouse et al. This is an open access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.Data Availability: The data for this paper were generate from a commercial cohort of patients who ordered the Panorama non-invasive prenatal test to screen for fetal aneuploidy. As such, before analysis for this manuscript, we anonymized the data so as to avoid any inadvertent release of patient health identifiers. The data contained in this anonymized data set is disclosed fully in the paper, and there is no additional information to be disclosed.Funding: Natera, Inc. provided support in the form of salaries for authors EK, MPH, ZD, SMZ, KJC, and SG, but did not have any additional role in the study design, data collection and analysis, decision to publish, or preparation of the manuscript. The specific roles of these authors are articulated in the “author contributions” section.Competing interests: EK, ZD, and SZ are employees of Natera with stock or options to hold stock in the company. MPH, KJC, and SG are former employees of Natera. There are no other conflicts of interest to report. Commercial affiliation with Natera, Inc. does not alter authors' adherence to PLOS ONE policies on sharing data and materials. X and Y chromosomal aneuploidies (the presence of an abnormal number of sex chromosome) are among the most common human whole-chromosomal copy number variations, with an estimated incidence in the general population between 1 in 400 to 1 in 1,000 [1,2–4] for each of the sex chromosome syndromes, with complex aneuploidies occurring far less frequently [5]. However, differences exist between newborn incidences and prenatal and adult incidences [6–8]. The population-based incidence and prevalence of the sex chromosomal aneuploidies (SCA) in the general adult population continue to be unclear. For example, prenatal incidence estimates vary for 47,XXY from between 1 in 500 [9] to 1 in 1,500 [2]. Women of advanced maternal age are more likely to receive prenatal testing, and since the probability of having an XXY child increases with maternal age, there may be an over representation of 47,XXY in prenatally diagnosed populations [2]. In postnatally diagnosed men, the prevalence of 47,XXY is reported to be 1 in 2,500 [2]. It is estimated that