HIGH IMPACT OF CARRIER SCREENING IN THE REFERRALS FOR PREIMPLANTATION GENETIC DIAGNOSIS FOR SINGLE GENE DISORDERS

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Abstract Body

Introduction: The referrals in Preimplantation Genetic Diagnosis for Single Gene Disorders (PGD-SGD) vary depending of the type of inheritance. In autosomal dominant conditions, the presence of the disease in one member of the couple is the main reason for performing PGD. For X-linked (XL) diseases, a previous affected family member or the presence of mild symptoms in the female partner has been the cause of the referral. Classically, a previous affected child has been the main cause for those autosomal recessive (AR) disorders. However, the carrier screening tests (CST) are increasingly a source of PGD cases for AR and XL disorders.

Design: Retrospective study where the reason of the referral has been analyzed in more than 2100 couples requesting PGD-SGD from 2003 to 2016.

Results: In AD diseases, the percentage of referrals due to the presence of symptoms in one member of the couple has been stable along the years (93.5%). In AR conditions, during 2003 to 2005, the principal PGD indication in our PGD program was the existence of a previous affected child (91.0% of AR couples) versus 3.0% of couples due to a CST results. In the last 2 years, couples with and affected child were 61.3% versus 33.1% of couples with a positive CST. Regarding the XL disorders, the number of PGD cases due to a positive CST has been increased from 0% in 2003-2005 to 28.9% in the last years. By contrast, the percentage of referrals due to an affected son has decreased from 28.4% to 21.2%.

Discussion: CST are displacing the classical causes of referrals for PGD-SGD indicating the higher awareness about the impact and incidence of carriers of mutations among population and allowing couples to increase their options for conceiving a healthy child.