

## IMPORTANCE OF GENETIC MATCHING THROUGH EXPANDED CARRIER SCREENING IN ONE OF THE LARGEST EGG DONATION PROGRAM IN SOUTH AMERICA

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### Abstract Body

#### Objective

To establish the prevalence of mutations of autosomal recessive diseases in a population of donors as in the couple of recipients.

Design: Retrospective and descriptive study

#### Material and Methods

A retrospective review was performed for oocytes donors and spouse couples in our Egg Donation Program in CEGyR Reproductive Center, in Buenos Aires, Argentina between January 2016 and December 2016. Expanded Carrier Screening is mandatory for our donors and spouse of the recipients. We evaluated prevalence of diseases in 457 donors and 355 husbands that were treated in that period. Diseases that were informed in the Expanded Carrier Screening (Recombine 287 diseases and 2398 mutations) were divided in 4 Groups: High Impact Diseases, Moderate Impact diseases, Treatment available diseases, and X linked Genetic Disorder

#### Results

Our Program is a Shared Egg Donation one. We found that 57% of our donors have at least one of the 287 recessive autosome diseases included in the test, 74.3% corresponded to high impact diseases on the expectation and quality of life, and 25.7% had a moderate impact. In the partners we found that 69% of them has at least one of the recessive autosome diseases evaluated, 75% high impact disease and 25% moderate impact. We found different prevalence and frequency of the diseases between our donors and the partner recipients. We also found differences with published prevalence in other populations of other sites of the world. After more than one year of follow up of born children we didn't find any autosomal recessive disorder

#### Conclusions

The use of donated oocytes is increasing. It is evident that the study of gamete donors for autosomal recessive genetic diseases is necessary to reduce the GENETIC RISK of children born from this treatment. We believe that the SYSTEMATIC incorporation of these genomic platforms to oocyte donors, as well as couples in search of pregnancy, will improve the chances of achieving a healthy child at home. It improves the selection of donors for recipient couples, given that in addition to the phenotypic characteristics it is possible