Introduction

As per regulatory requirements in Ukraine, test for karyotyping should be routinely performed for all oocyte donors. DNA-based carrier screening for CF, Frag X, SMA, and neurosensory deafness is implemented as good clinical practice approach of donor eligibility protocols practiced in our group. Applicants who test positive for carrying a recessive disease mutation are typically disqualified.

The aim of our study was to examine the utility of a range of expanded screening panels and the effectiveness of the screening paradigm in reducing a future child’s risk of inheriting disease.

RESULTS

Genotyping results for all donors were analyzed; 38% (35/92) of donors were identified as carriers for one condition, 34% (31/92) for two conditions, 7% (6/92) for three conditions and 7% (6/92) for four conditions, including cystic fibrosis. Among the most prevalent conditions in our study were: Hemochromatosis: Type 1: HFE Related – 22%, Cystic Fibrosis: CFTR-related conditions 11%, Biotinidase deficiency – 7%, 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia – 6%, Krabbe disease – 6.5%. Usher syndrome:USH2A-related conditions – 6.5%, Nonsyndromic deafness: GJB2-related conditions – 5.4% and Smith-Lemli-Opitz syndrome (5.4%).

Frequency and spectrum gene mutations among oocyte donors

Quantity of donors with a gene mutations

Distribution of gene mutations among oocyte donors

CONCLUSIONS

1. While performing expanded carrier testing panel using NGS data the rate of gamete donors identified as carriers of at least one condition was 86%, which supports the offering of expanded carrier screening to oocyte donors population.

2. Although the research sample size did not include the general population, the results show that genotyping is associated with a high reproductive risk since the standard PCR panel did not detect 11% of the CFTR mutation carriers. We recommend a complete sequencing of the CFTR gene by next-generation sequencing as a screening method for all donors.

3. Effective approach to reduce recessive disease risk would imply a comprehensive analysis of both donor and recipient disease mutations.