REPRODUCTIVE GENETIC COUNSELLING ASSOCIATED WITH PRENATAL WES RESULTS

Krechmar, Marina[1]

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Abstract Body

OBJECTIVE: Whole exome sequencing (WES) is rapidly entering prenatal diagnostics, but understanding the benefit of this clinical results is in the early stages. The introduction of WES into prenatal practice may improve the diagnostic potential for risk determination and effective reproductive genetic care for the high risk family.

METHOD: We performed WES to evaluate the presence of genetic patogenic variants in genes after priority exclusion of aneuploidy by karyotyping and or other pathogenic cytogenetic abnormality by CMA in a cohort of fetuses with structural anomaly phenotypes with suspected a monogenic syndrome. For 12 casess, DNA tests was sent after TOP, and in 5 families during an ongoing pregnancy. All families counseled by complex genetic diagnostic and received an explanation regarding the benefits and limitations.

RESULTS: In 65% (11/17) cases, WES provided definitive fetuses diagnoses (8 cases – AR, 3 – AD, 2 - ADde novo). Parental DNA was examined to determine the origin of fetus mutations and calculate predictions, determine indications and possibilities PGD and prenatal diagnostic. In 2 prenatal cases, a single mutation in an AR type genes was identified. Complex testing of two fetuses with skeletal abnormalities ruled out a difficult prognosis. The application WES makes it possible in some cases to detect the genetic cause of fetal abnormalities, to determine the prenatal and postnatal prognosis in a current pregnancy. The combination of genetic tests allowed effective risk identification and reproductive counseling with a certain ability to identify and exclude the pathological mutations at the embryonic (PGD– M) or prenatal stage.

CONCLUSION: Future enhanced practices the integration of WES in reproductive genetic counseling associated with genetic testing of embryos and fetuses will be important for ongoing and future pregnancies. The development of clinical algorithms for high risk families is an extremely urgent task in the reproductive genetic practice.