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INFORMATION – POLAR BODY DIAGNOSIS

Laser assisted opening of the zona pellucida of the oocyte, removal of polar bodies 1 and 2, genetic analysis - FISH (chromosomes 13, 16, 18, 21, 22)

Dear patient,

our institute offers an additional diagnosis, which could increase your chances for the birth of a child as a consequence of assisted reproduction. The following information provides the scientific and medical background of this approach. It is of importance for us to offer you all the detailed information necessary to clarify each of your questions. Please do not hesitate to contact us, if there are any additional open questions.

Introduction:

The 46 human chromosomes are the carriers of the genetic information. Each individual has 22 pairs of chromosomes (autosomes) and in addition two sex-chromosomes - females have two X-chromosomes and males have one X- and one Y-chromosome. Each individual inherits 22 autosomes from the mother and 22 autosomes from the father. In the course of the development of oocytes during the first meiotic cell division one pair of chromosomes remains in the oocyte and the second one is transfered into the first polar body. Upon sperm injection the second meiotic cell division of the oocyte occurs. As a consequence of this second division one set of chromatides remains in the oocyte and the second set of chromatides is transferred into the second polar body.

Accordingly, normaly the number of chromosomes in the first polar body should reflect the number of chromosomes in the oocyte. However, it is known that numerical chromosome aberrations (aneuploidies) can occur. Such aneuploidies occur mainly (70-80%) during the first meiotic cell division (the formation of the first polar body). In case of chromosomal aneuploidies the numbers of chromosomes in the first polar body and in the

oocyte are not identical. The incidence of chromosome aneuploidies increases with maternal age. 50-70% of the oocytes of a 40 year old female are already carriers of chromosomal aneuploidies. This is one reason why the chance for becoming pregnant and for the birth of a baby decreases, whereas the risk for miscarriages increases, with maternal age.

- 95% of all embryos carrying a genetic aberration or having a developmental impairment will not lead to a pregnancy or will end in early abortions

- A 3% basic genetic risk for physical or mental retardation of the child exists for every pregnancy, occuring naturally or via assisted reproduction. Here it is important to note, that polar body diagnosis does not affect this basic genetic risk.

Removal of the polar bodies and analysis of the chromosomes via fluorescence in situ hybridization (FISH) offer the opportunity to detect chromosomal aneuploidies. It is the aim of this approach to determine the number of specific chromosomes in the polar bodies. Detecting one copy of a specific chromosome in the first polar body allows the conclusion that there is also one copy of this chromosome in the oocyte, what reflect the normal situation. If no copy of a specific chromosome is detected in the first polar body, the oocyte harbors two copies of this chromosome. Such a result would be representative for a chromosomal aneuploidy.

Polar body biopsy and FISH diagnosis is an established approach to detect chromosomal aneuploidies, which is used in international laboratories with good success. We offer this genetic diagnosis at our institute. Some hours after the intracytoplasmic sperm injection (ICSI) the zona pellucida of the oocyte will be opened and the polar bodies will be removed. The used laser as well as the technique itself do not affect the oocyte and do not interfere with the development of the embryo. From a technical point of view it is not always possible to isolate also the second polar body. We would not isolate it, if we cannot exclude to mediate negative influence on the oocyte. After isolation of the polar bodies FISH analysis will be performed to determine the numbers of the chromosomes 13, 16, 18, 21, 22. This allows indirect conclusion about putative chromosomal aneuploidies in the corresponding oocytes.

Benefits:

According to the current status of research polar body diagnosis can increase the chances for the birth of a child as a consequence of assisted reproduction. The aim of this approach is to avoid the usage of oocytes, haboring a genetic aberration, which is either incompatible with the induction of a pregnancy, or which would lead to the birth a child with Pätau-Syndrome, Edwards-Syndrome, or Down-Syndrome.

Risks and limits:

In all the currently published international studies a risk for the embryo could not be detected. Independently of the polar body diagnosis we want to recommend the usage of the different prenatal diagnosis approaches in case of pregnancy. The here offered polar body diagnosis only detects aberrations of the chromosomes 13, 16, 18, 21, and 22 with a 90-95% sensitivity. For example, the here offered polar body diagnosis can decrease the risk for having a Down-Syndrome baby, but cannot exclude that with 100% certainty. Aberrations of the chromosomes, which are not analysed via polar body diagnosis, cannot be excluded. In addition, a 20-30% risk for chromosome aberrations remains given the second polar body could not be isolated. Chromosomal aberrations transferred/inherited via the sperm cell cannot be analysed by polar body diagnosis. Prenatal diagnosis approaches, such as ultrasound investigation, peripheral maternal blood analyses or genetic analyses (e.g. amniocentesis) allow the detection/exclusion of specific fetal retardations/malformations or fetal genetic alterations.

Counseling:

Counseling will be performed by a responsible medical doctor. If polar body diagnosis should be performed the patient and the counsellor need to sign the informed consent.