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Polar body diagnosis **Information/Informed consent**

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 transferred 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.

The success rate of in vitro fertilisation (assisted reproduction) clearly depends on a normal chromosomal status of oocytes and sperms. However, it is known that numerical chromosome aberrations (aneuploidies) can occur. 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 % of the oocytes of a 35 year old female and already up to 70 % of the oocytes of a 40 year old female are carries 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 and newborns with developmental disorders increases, with maternal age. Accordingly, in the course of in vitro fertilisation the selection of oocytes with normal chromomal status is of importance. An approach to achieve this aim is the so called polar body diagnosis. Removal of the polar bodies and analysis of the chromosomes offer the opportunity to detect chromosomal aberrations in oocytes.

Removal of the polar bodies: 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.

Genetic analyses of the polar bodies: After isolation of the polar bodies fluorescence in situ hybridization (FISH; analysis of the chromosome 13, 16, 18, 21, 22) or comparative genomic hybridization (CGH; analysis of the chromosomes 1-22, and X) will be performed. This allows indirect conclusion about putative chromosomal aberrations in the corresponding oocytes.

Benefits: According to the current status of research for a specific group of patients polar body diagnosis can increase the chances for the birth of a healthy 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, which could be responsible for miscarriages or which could lead to the birth of children with certain genetic developmental disorders.

Risks and limits: In all the currently published international studies a risk for the embryo could not be detected. Independently of the polar diagnosis we want to recommend the usage of the different prenatal diagnosis approaches in case of pregnancy. The here offered polar body diagnosis is not of 100% validity and does not detect all aberrations of the chromosomes. 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 or occurring during early embryonic development 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.

Patient: I have been detaily informed about polar body diagnosis and a copy of this information has been provided. I herewith agree that polar body diagnosis via FISH ☐, CGH ☐ will be performed. I know that I can ask for a stop at any time point of the procedure after having talked to the responsible medical doctor.

Name:

Signature: Date:

Medical doctor: I have explained the approach of polar body diagnosis to the patient. The patient is informed about the details and benefits of the procedure.

Name:

Signature: Date:

Univ.-Prof. Dr. Markus Hengstschläger: The patient has been informed about the genetic aspects and the laboratory methods of polar body diagnosis.

Signature Date: