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GENETICS OF MALE INFERTILITY IN THE ERA OF ICSI

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Male infertility is a multifactorial pathological condition affecting about 7% of the general male population. The genetic landscape of male infertility is highly complex since semen/testis histology phenotypes are extremely heterogeneous and at least 2000 genes are involved in spermatogenesis. Although the highest frequency of known genetic factors (25%) is found in azoospermia (mainly primary testicular failure), the number of identified genetic anomalies in the other semen/etiologic categories is constantly growing. Besides its diagnostic value, genetic screening is relevant for clinical decision-making (prior testicular sperm extraction) and for appropriate genetic counselling (including general health issues). While males with impaired sperm production due to a genetic factor may now benefit from the wide availability and utilization of ART, the potential risk of transmitting genetic defects to the offspring deserves thoughtful consideration. Routine genetic testing is available for hypogonadotrophic hypogondism (Kallmann sdr and the normosmic form), Congenital absence of vas deferens (CFTR gene mutation screening), monomorphic terato/asthenozoospermia, and primary hypogonadism (karyotype and Y chromosome microdeletion analysis). In about 50% of cases the etiology remain unknown and is called “idiopathic infertility”. Results from whole genome association studies suggest a marginal role for common variants as causative factors. Copy Number Variations (CNV) demonstrated a significantly higher CNV load in patients in respect to controls whereas whole exome analysis resulted a highly successful diagnostic tool in familial cases of male infertility. The discovery of novel genetic factors in idiopathic infertility represents a major challenge for the field of andro-genetics and requests large international consortiums.

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