

PRE-IMPLANTATION GENETIC DIAGNOSIS - A CASE STUDY

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Izabella and her partner sought pre-implantation genetic diagnosis (PGD) because Izabella had retinoblastoma due to a deletion in chromosome 13 and they want to have children not at genetic risk of retinoblastoma. Fortunately, Izabella's tumour was unilateral and was treated successfully and she is well. Izabella's chromosome abnormality is mosaic with 70% of lymphocytes having the deletion. This mosaicism may not be present in Izabella's ovaries. The couple went through PGD on two occasions and 13 embryos were tested. None had the deleted chromosome 13. IVF and PGD failed to produce a pregnancy. The couple seek to know what the experience provides as to the risk to their offspring: in particular, does it indicate a risk low enough to be acceptable if they go ahead with a natural pregnancy instead of another resort to PGD? Also the couple do not want prenatal diagnosis. The situation therefore requires an estimate of the probability that an embryo will have the deletion. Counselling is problematic because there is no obvious way of selecting a prior probability from which to compute a Bayesian estimate of risk. Two solutions are offered, depending on the amount of information available about genes transmitted from the maternal grandparents.