CHROMOSOMES IN HUMAN EMBRYOS a review

The paper by Ivanova et al. (1) discusses why human embryos frequently carry chromosome abnormalities. Whole-chromosome aneuploidies usually originate during maternal meiosis, with risk increasing markedly with age, whereas mosaicism arises from errors in the first embryonic divisions. In humans, the oocyte meiotic spindle forms without centrosomes. This combined with less stringent checkpoint control, increases the chance of incorrect kinetochore-microtubule attachments. especially merotelic ones, where a single kinetochore attaches to microtubules from both spindle poles, leading to chromosome missegregation. Early embryonic mitoses rely on centrosomes of paternal origin; defects in sperm centrosomes increase the likelihood of mosaicism. Mutations in maternal genes such as TUBB8, MEI1, and PLK4 compromise spindle stability and cohesion, whereas aging reduces cohesion levels and the protection provided by protein shugoshin, resulting to errors at meiosis I. Estimates of mosaicism from preimplantation genetic testing for an uploidy remain uncertain, underlining the need for closer integration between basic research and clinical practice.

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