



Dr Dean Morbeck

Fertility Associates

Saturday, June 10, 2017

12:00 - 12:10 Fertile but Rogue Genes



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Fertile – but Rogue Genes

Dean Morbeck, Ph.D., MBA
Scientific Director

Leaders in Fertility



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Preimplantation Genetic Testing

Preimplantation Genetic Diagnosis

- Test for single gene diseases or chromosome rearrangement (translocation)

Preimplantation Genetic Screening (also called PGD-A)

- Test karyotype of embryo (euploid vs aneuploid)

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- Blood disorders (11%)
 - Haemophilia
 - Sickle Cell
- Brain/cognitive (23%)
 - Huntington
 - Fragile X
- Cancer (5%)
 - BRCA
 - Lynch Syndrome
- Metabolic (3%)
 - Galactosaemia
 - VCLAD
- Misc (11%)
 - Long QT Syndrome
 - PKA
 - Fabry Disease
 - Colleganopathy
- Muscular (8%)
 - DMD
 - SMA
- Nervous System (5%)
 - Charcot-marie-tooth
 - NF-1
- Respiratory (11%)
 - CF
- Skeletal (3%)
 - Otopalatodigital syndrome
 - Crouzon syndrome
 - Cleidocranial dysplasia
- Translocations/inversions (20%)

Hatch and Biopsy

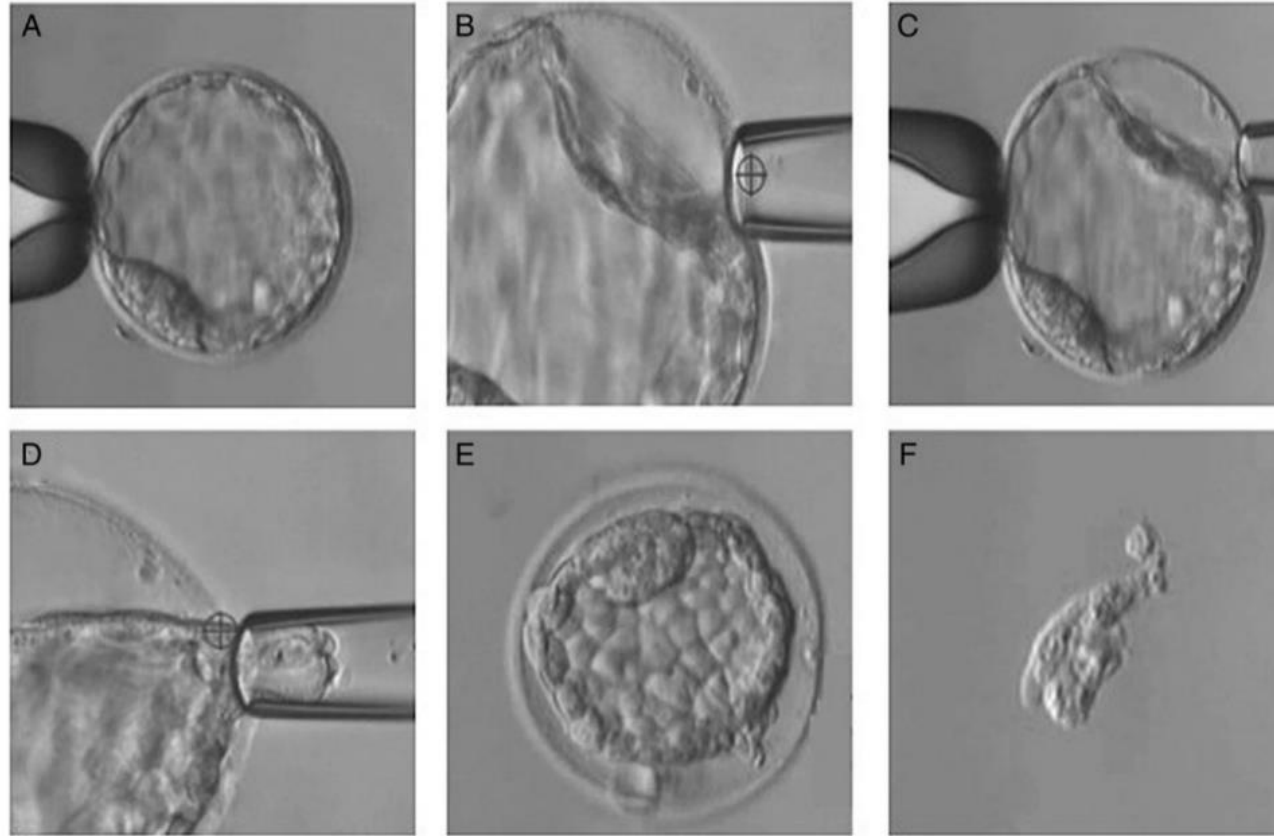


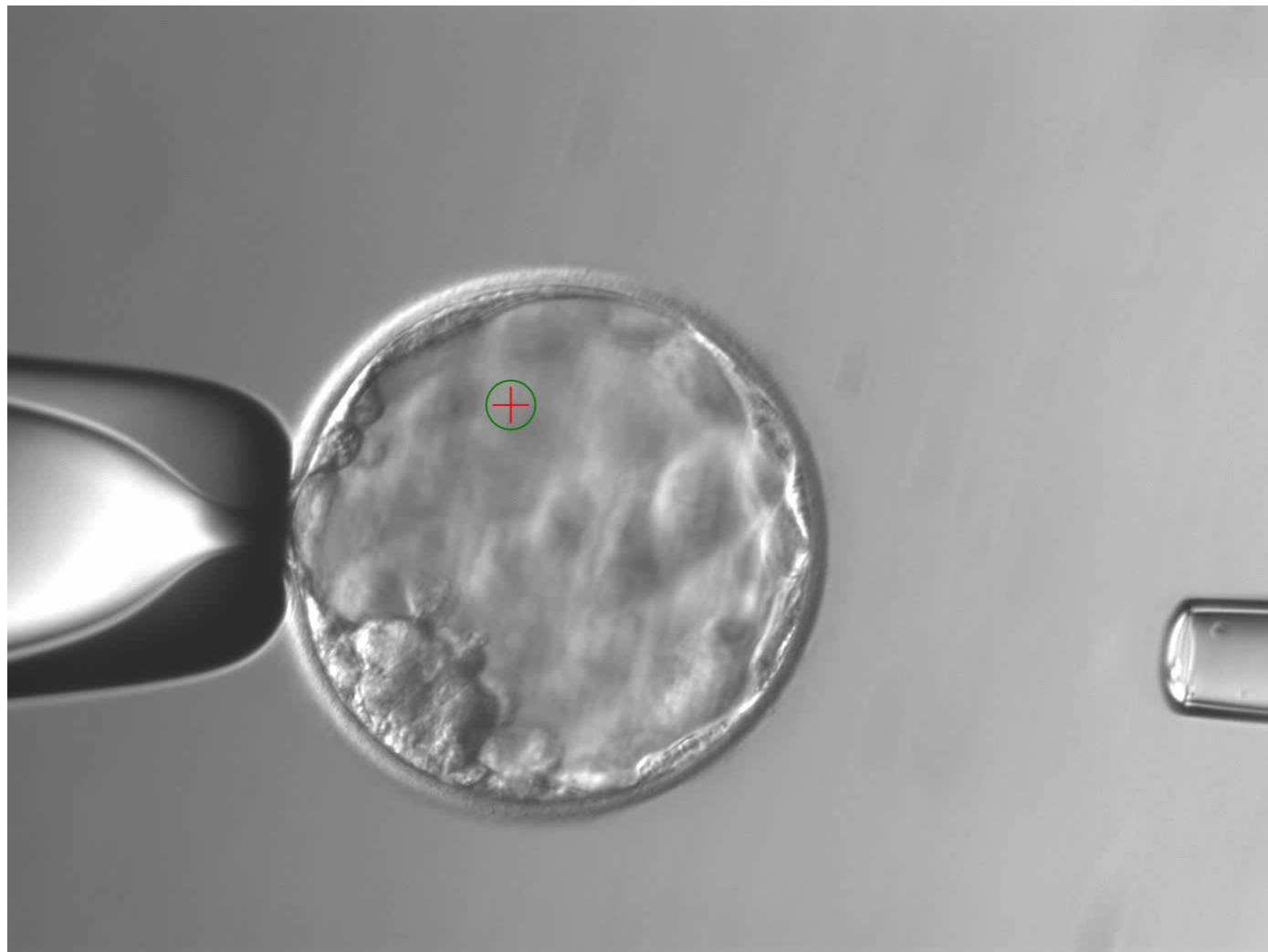
Figure 1 Trophectoderm biopsy protocol for human expanded blastocysts at the cleavage stage of embryo development. This figure shows the clinical protocol used to biopsy good quality human blastocysts on Day 5, Day 6 or Day 7 of development to obtain multicellular samples for DNA amplification and genetic testing. The protocol is suitable for both hatching and non-hatching blastocysts and requires no breaching of the zona pellucida during cleavage stages or the period of culture prior to biopsy. (A) Expanded good quality human blastocyst positioned on the holding pipette to give a clear view of the ICM at 7 o'clock, $\times 20$ magnification. (B and C) Biopsy pipette expelling medium through the breach in order to release the TE cells from the internal surface of the zona, $\times 40$ and $\times 20$ magnification, respectively. (D) TE cells are aspirated into the biopsy pipette with gentle suction and several laser pulses are directed at the junctions between cells to disconnect the aspirated cells from the body of the embryo, $\times 40$ magnification. (E) Blastocyst after TE biopsy, $\times 20$ magnification. (F) TE biopsy cell sample released into the biopsy drop, $\times 20$ magnification.

Capalbo et al. HR 29:1173-1181



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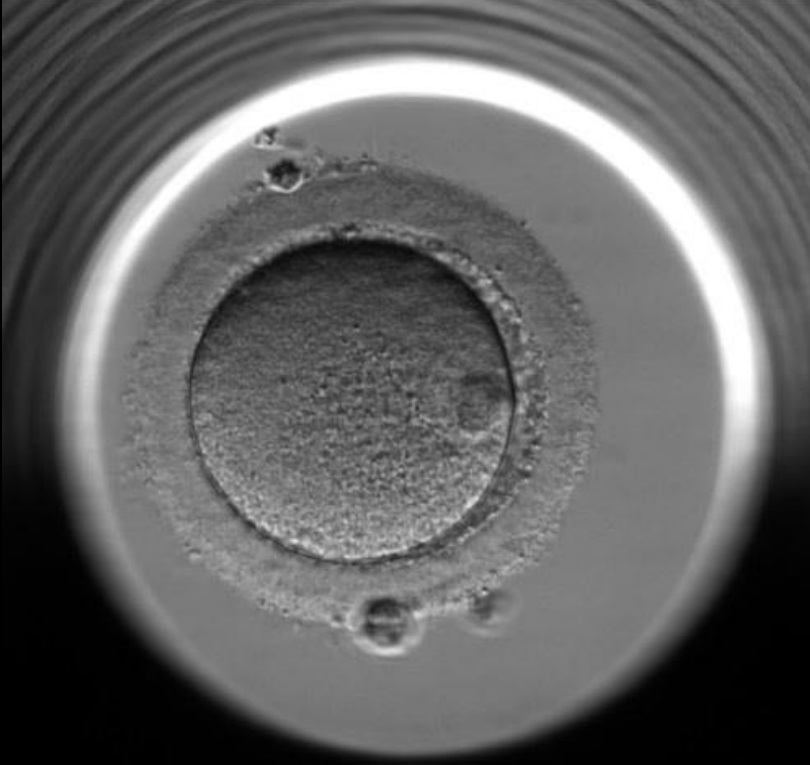
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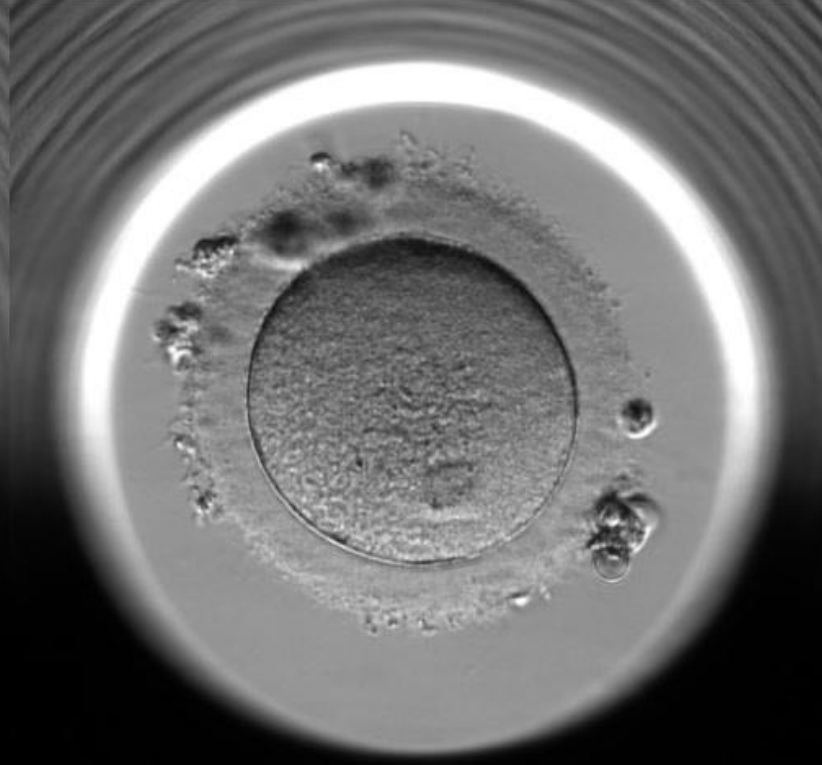
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TE Biopsy



Well 2



0.3 h

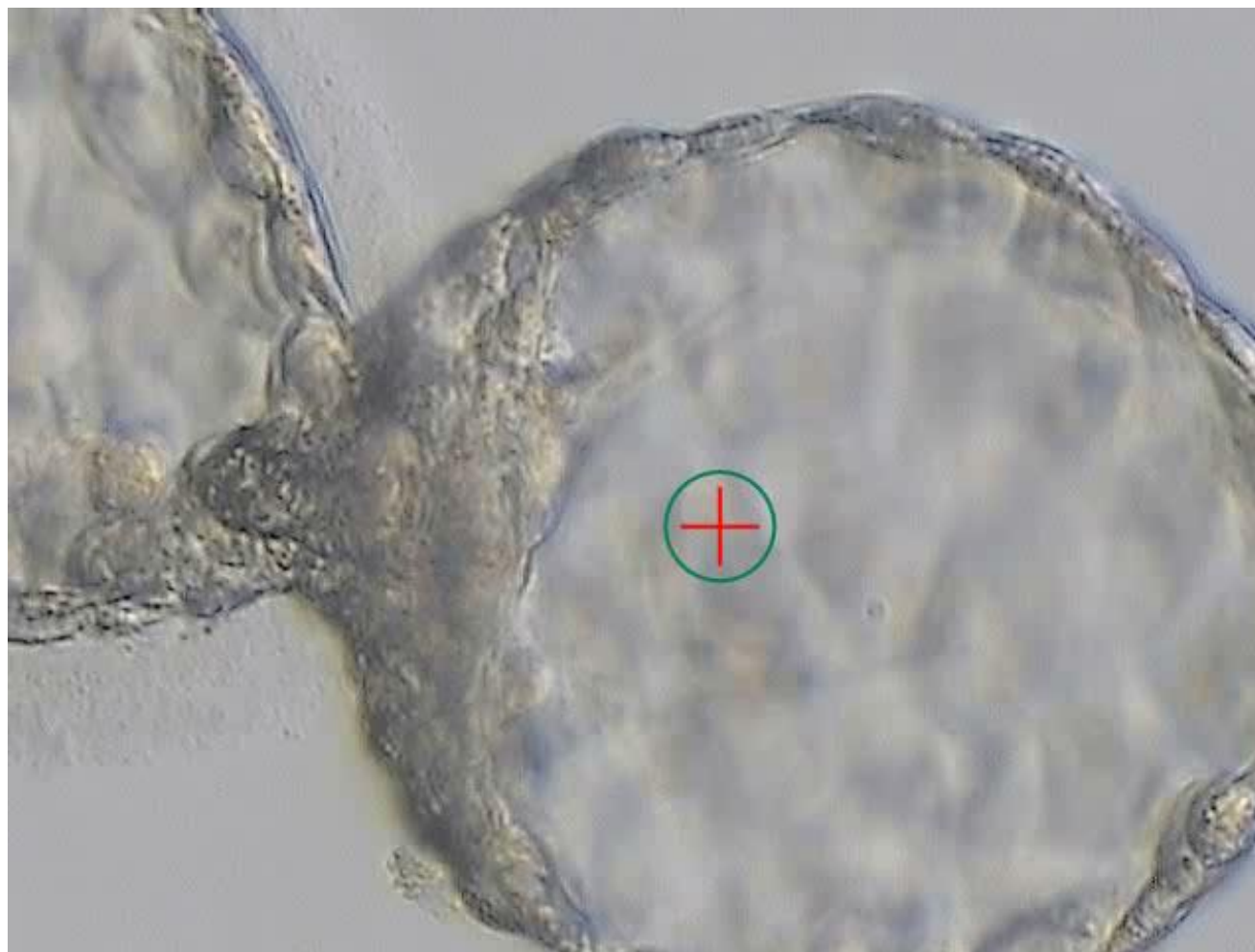
Well 8

0.3 h



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PGD – 2016 Experience

- N=30 (consistent with FA's historical utilisation; 1% of cycles)
 - 6 Huntington
 - 5 Translocations
 - 3 Muscular
- 58% pregnant
 - 3 of 30 with no unaffected embryos to transfer
 - 71% pregnant of those with a transfer



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Thank You!

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