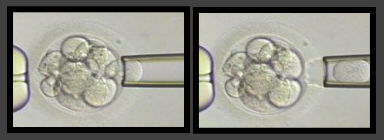
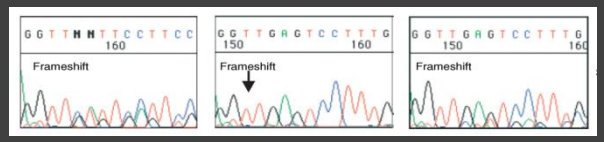
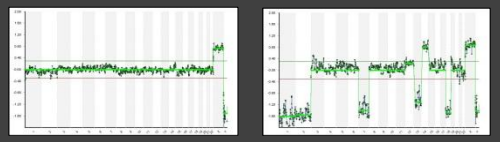
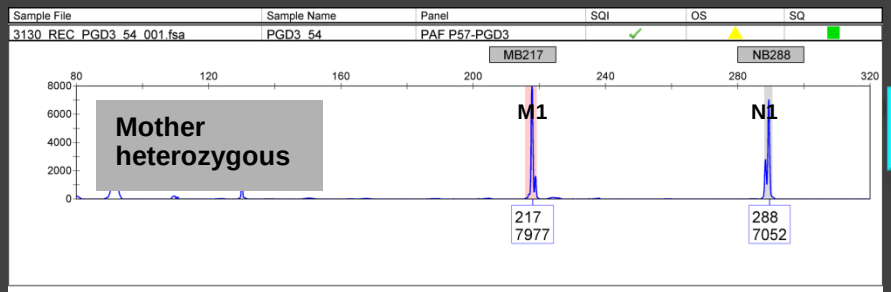
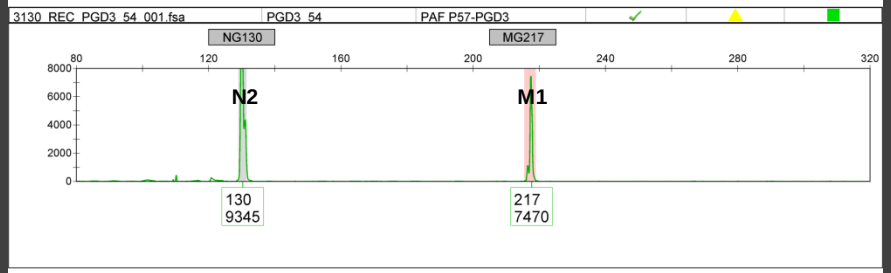


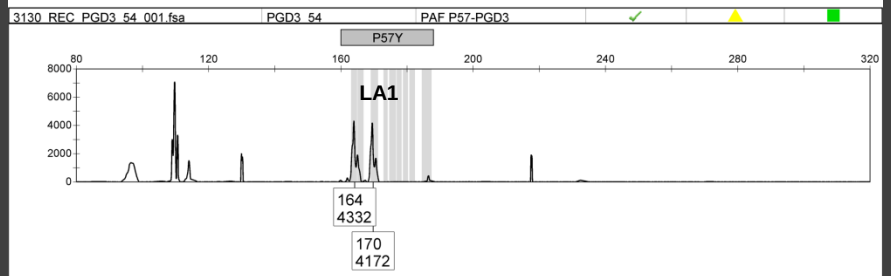


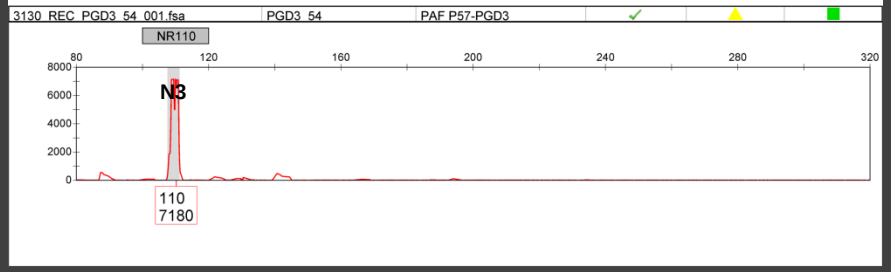


Mother heterozygous

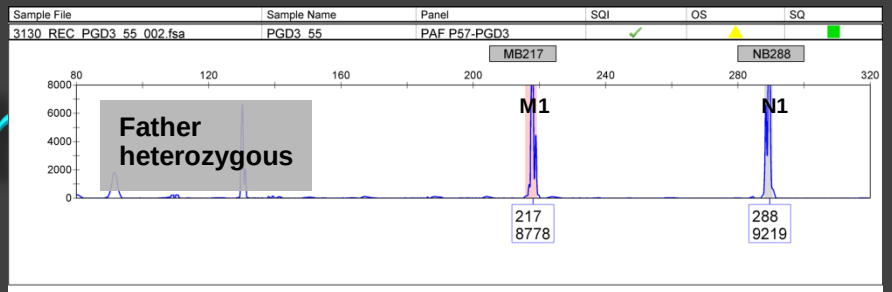


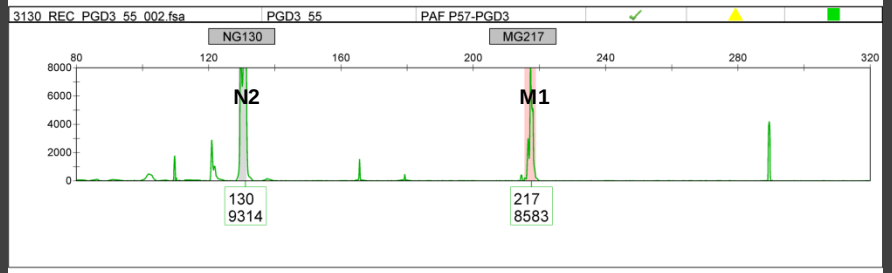


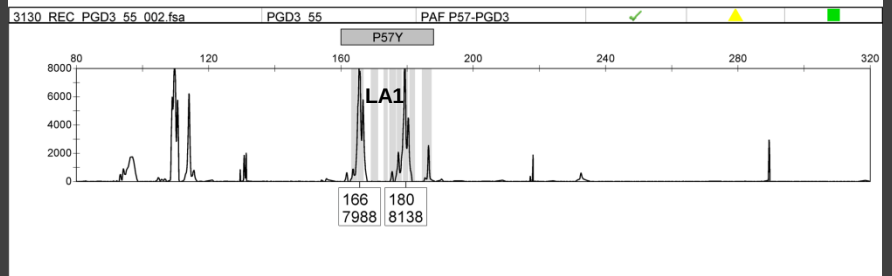


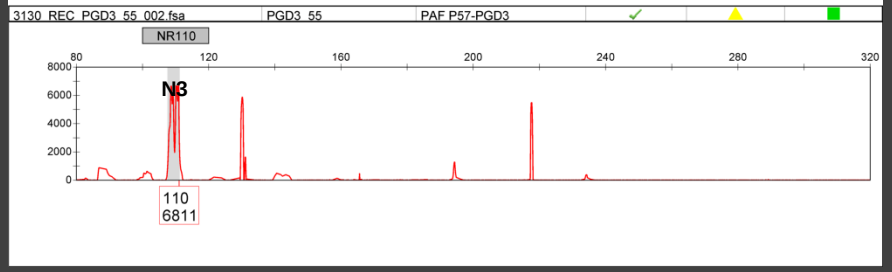


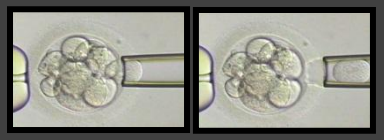
Father heterozygous

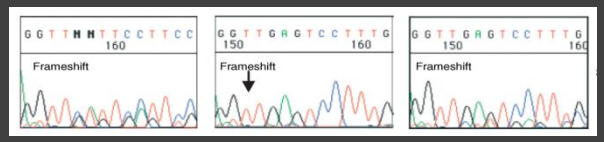


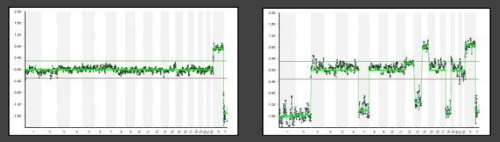




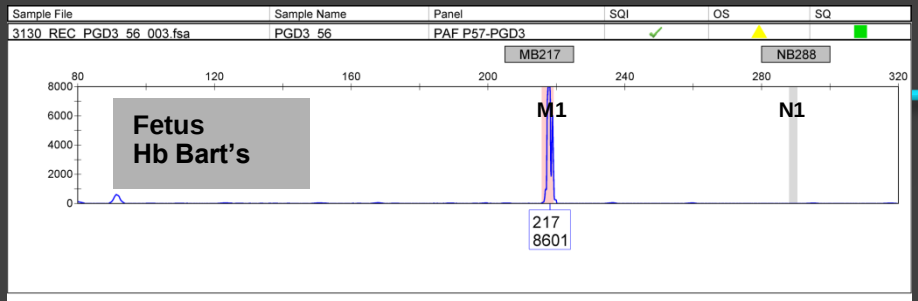


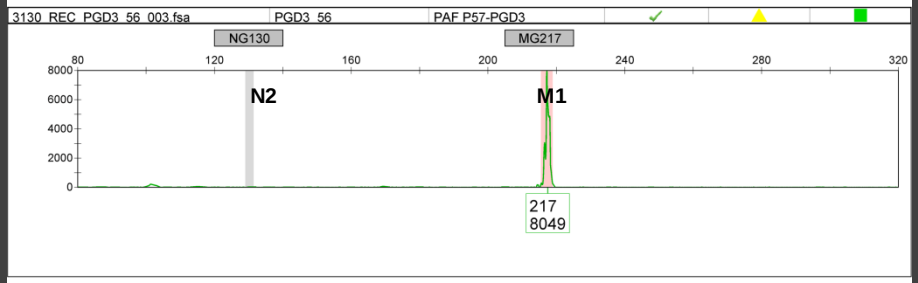


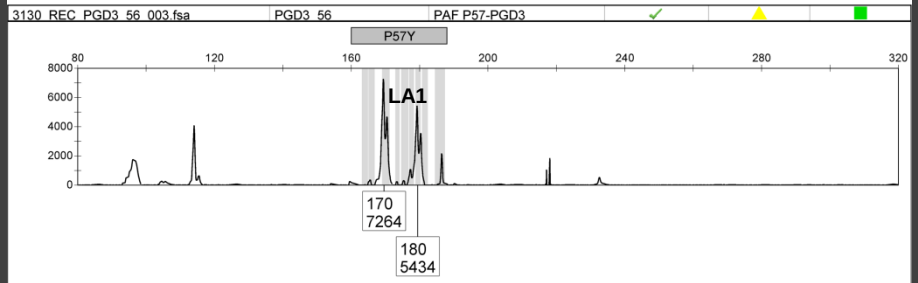


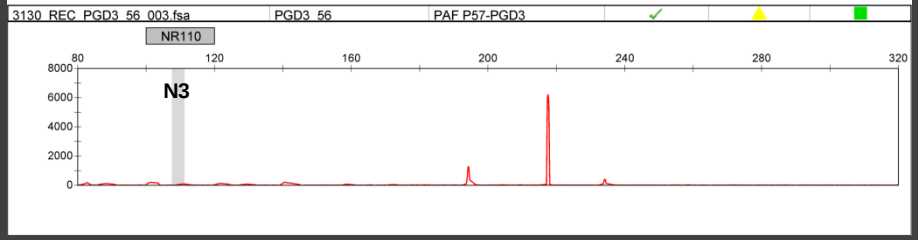


Sample File | Sample Name | Panel | SQI | OS | SQ
3130_REC_PGD3_56_003.fsa | PGD3_56 | PAF P57-PGD3

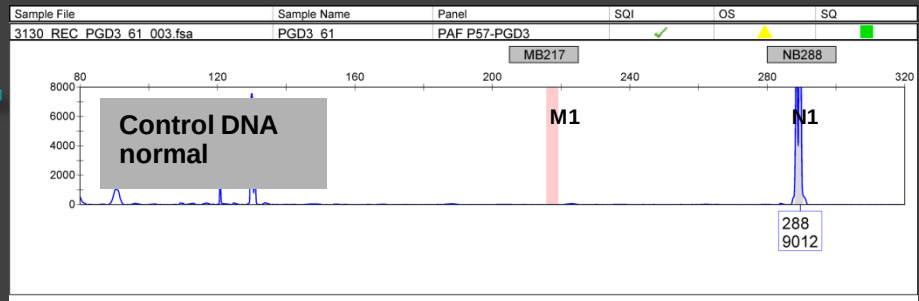


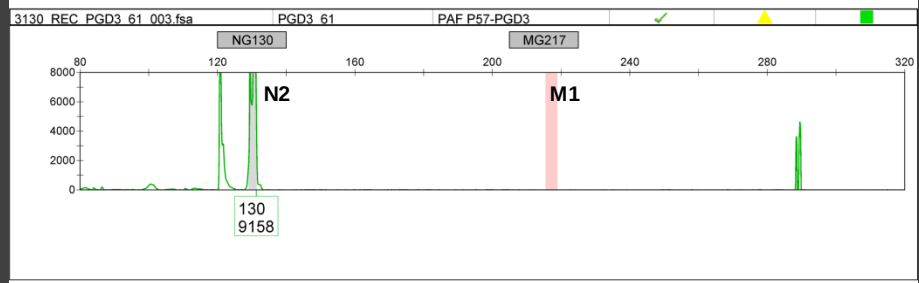


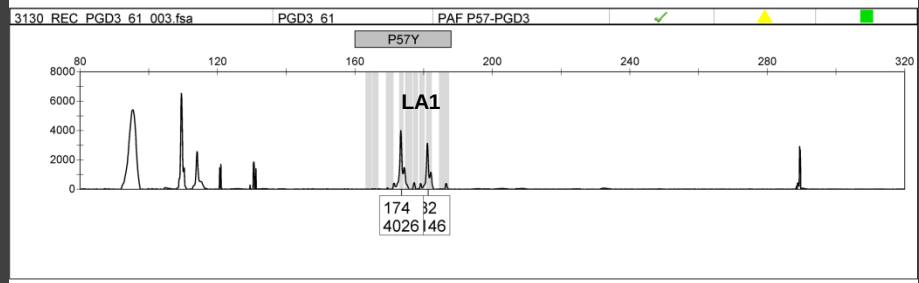


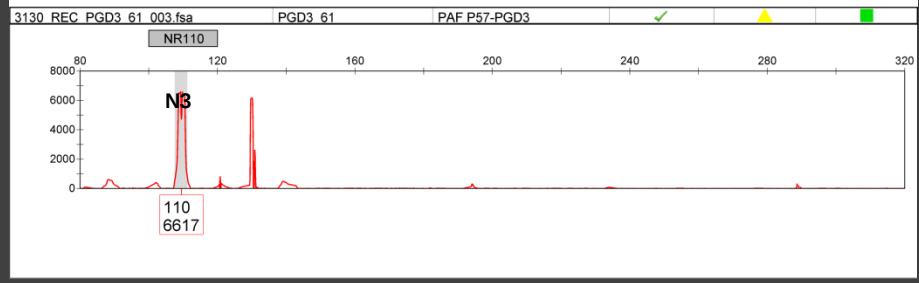


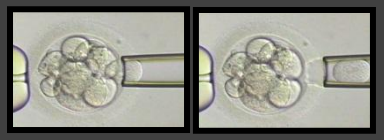
Sample File | Sample Name | Panel | SQI | OS | SQ
3130_REC_PGD3_61_003.fsa | PGD3_61 | PAF P57-PGD3

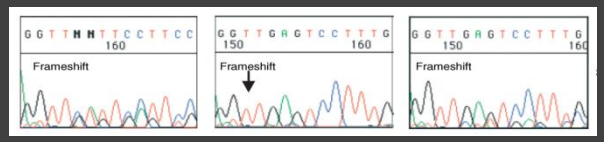


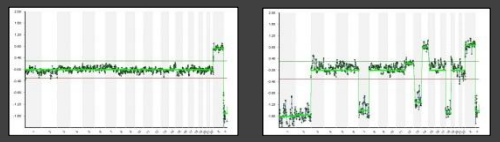




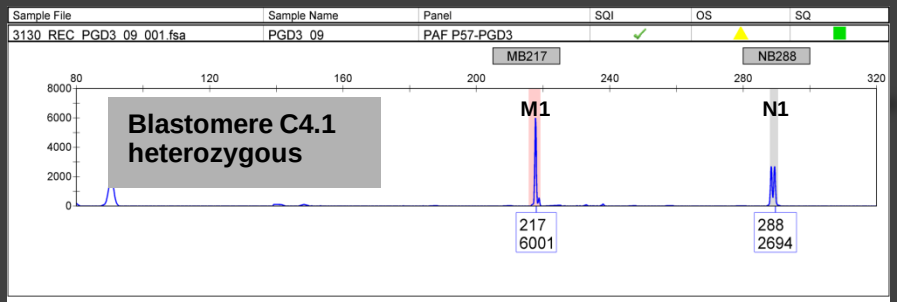


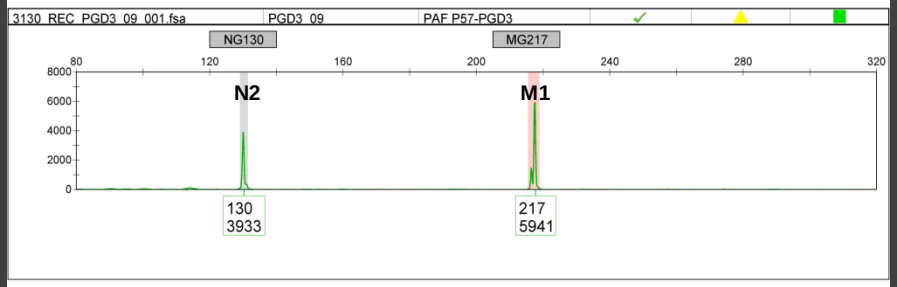


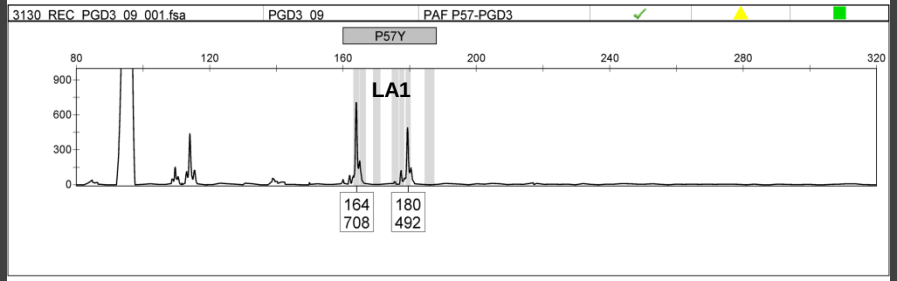


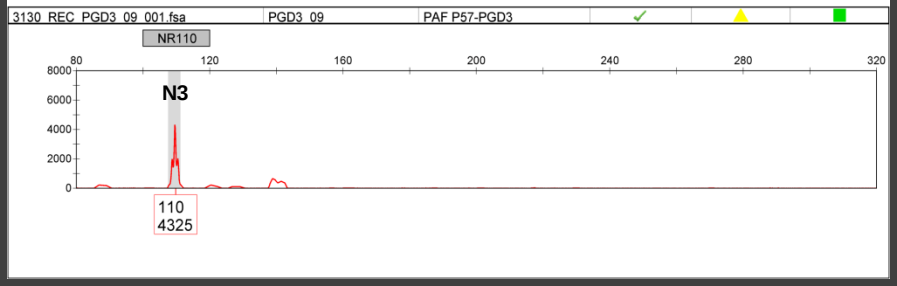


Blastomere C4.1
heterozygous

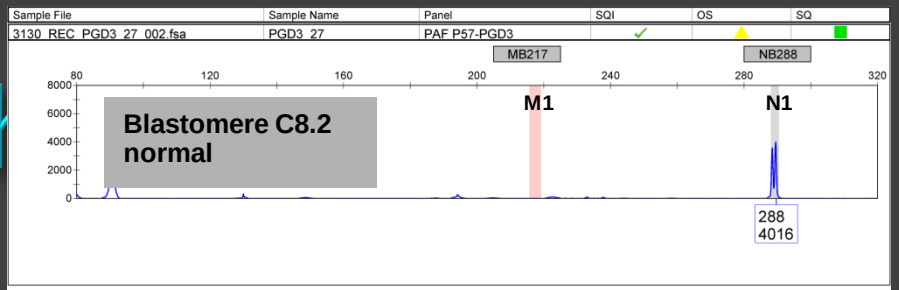


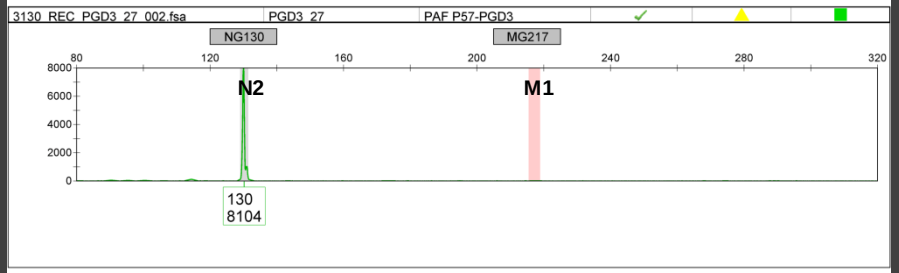


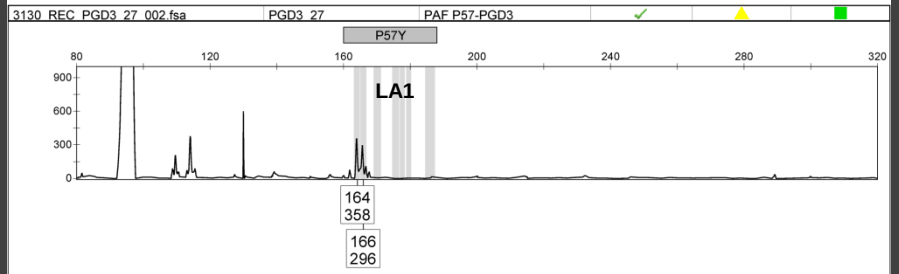


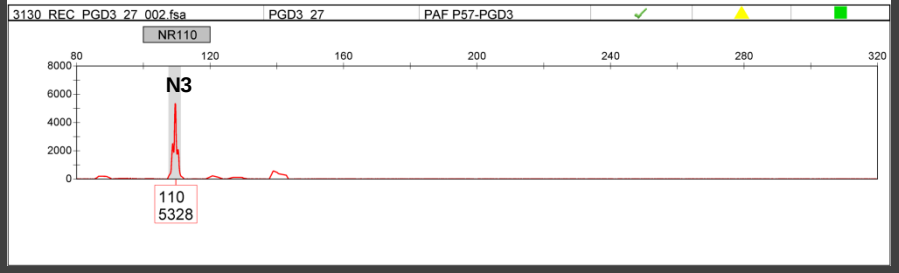


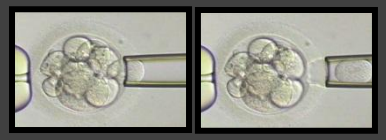
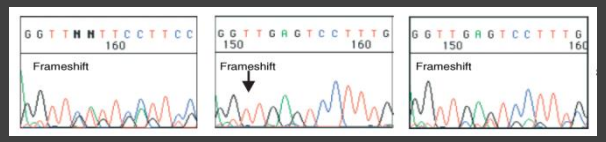
Blastomere C8.2
normal

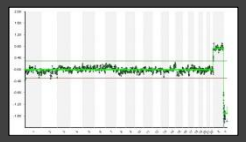
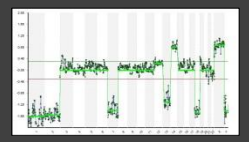






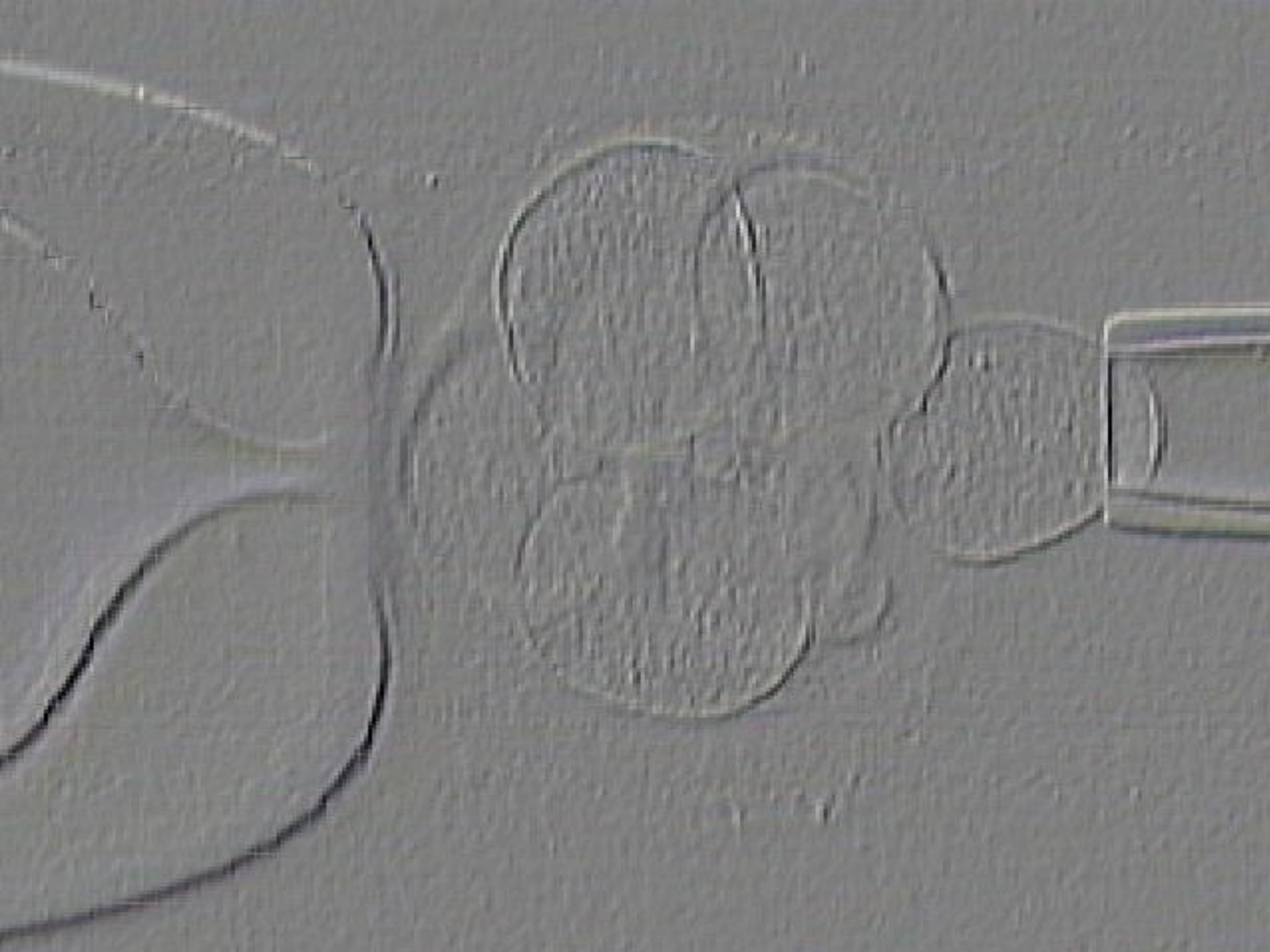


PGD of Thalassaemia at CHIANG MAI UNIVERSITY

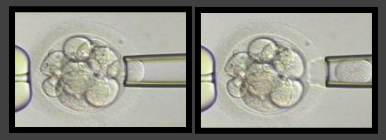
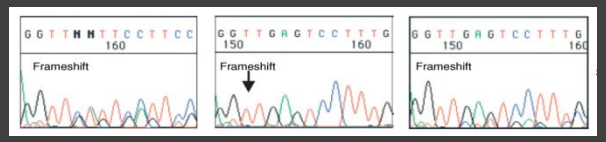
	cycles	pregnancies
beta-Thal	2	1
alpha-Thal	26	9 (10)
beta-Thal-HbE	6	2
Total	34	12 (13)

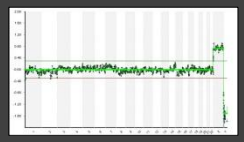
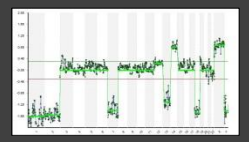


MedCMU

PGD Center
Department of OB&GYN

Faculty of Medicine, Chiang Mai University, Chiang Mai, THAILAND.

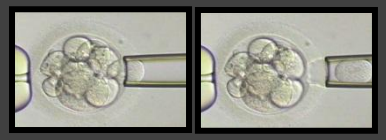
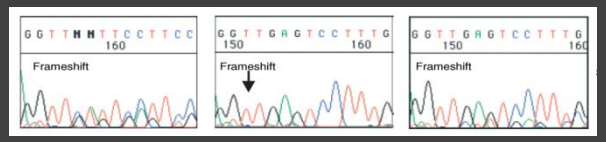
PGD Protocol

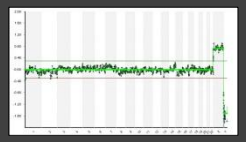
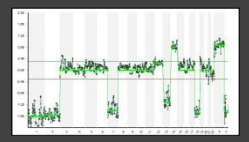
Single Cells

Cell Lysis

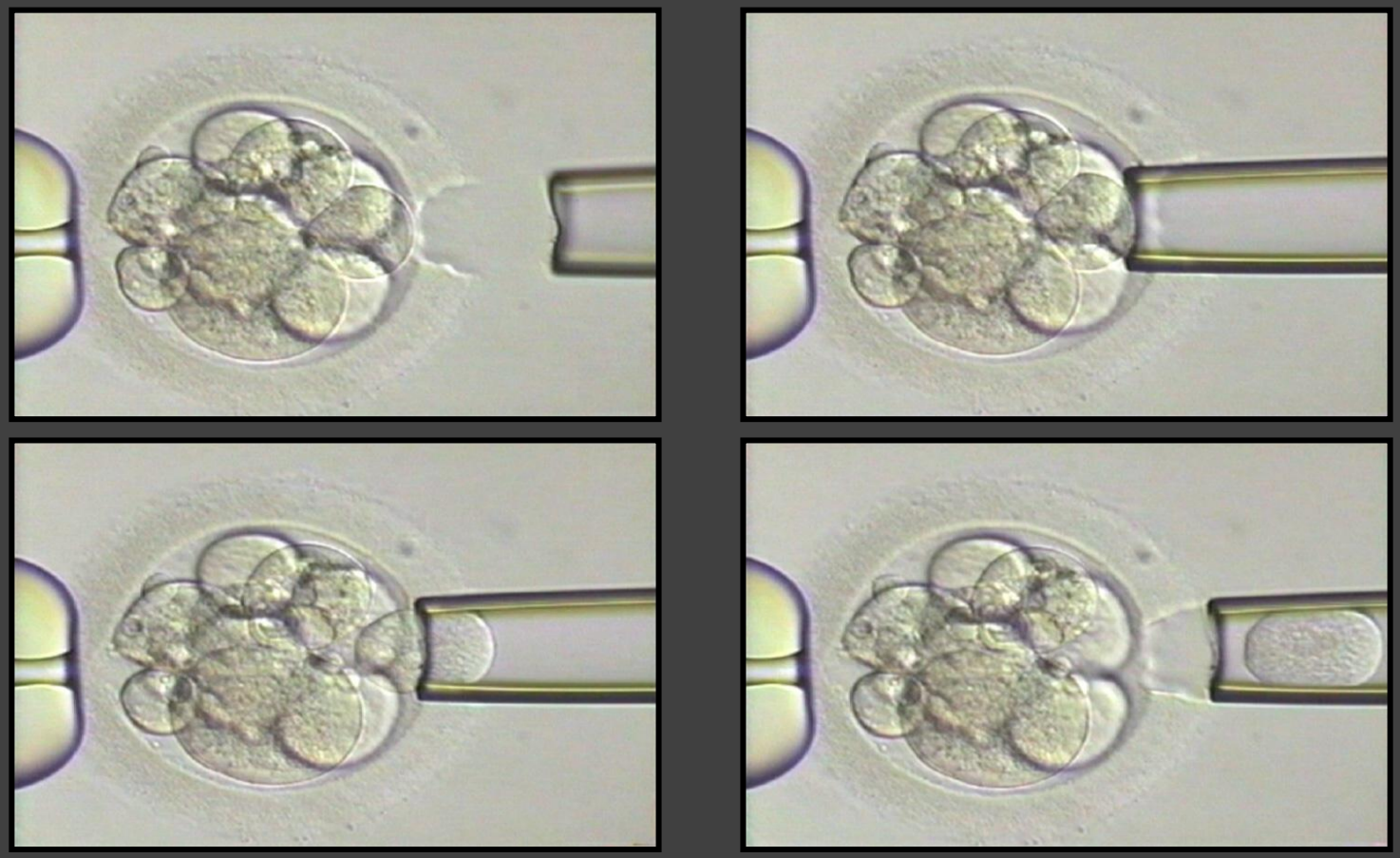
Multiplex PCR

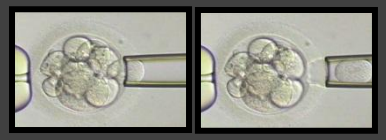
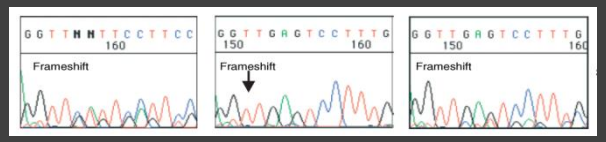
F-PCR Analysis

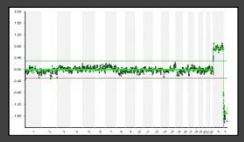
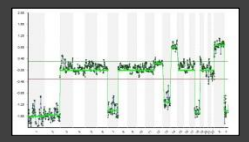



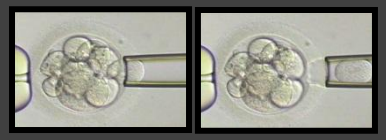
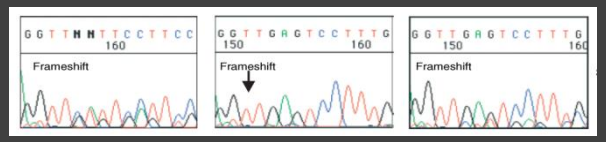
CLEAVAGE STAGE EMBRYO BIOPSY

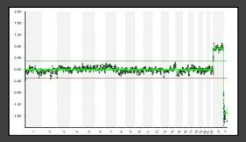
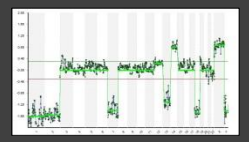


CLEAVAGE STAGE EMBRYO BIOPSY

PGD of beta-thalassaemia codon41-42



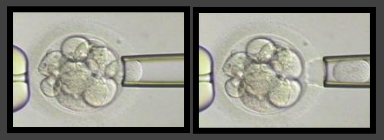
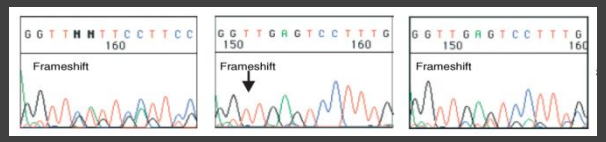


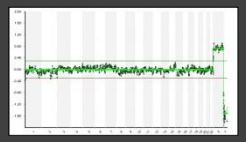
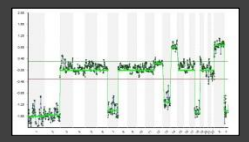






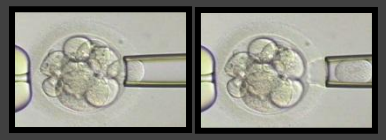
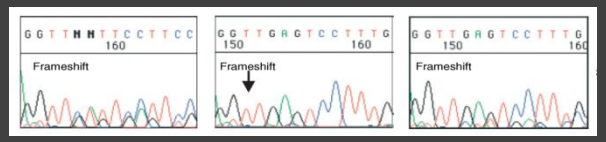

normal	heterozygous	affected	normal
ET	ET		ET

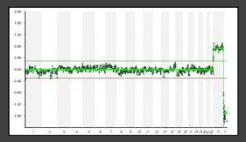
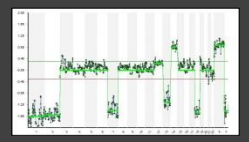



PGD of beta-Thalassaemia codon 41-42 couple A

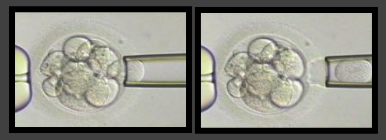
- 14 oocytes collected
- 8 embryos biopsied
- 2 normal - ET
- 1 clinical pregnancy resulted

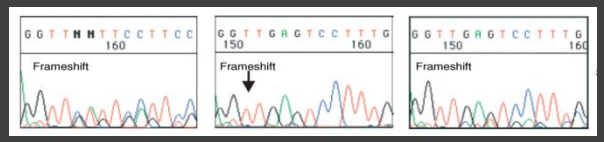



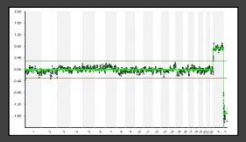



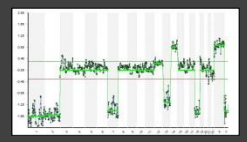
PGD of beta-Thalassaemia codon 41-42 couple B

- 20 oocytes collected
- embryo freezing due to OHSS
- resume PGD
- 9 embryos biopsied
- 1 normal + 1 heterozygous - ET





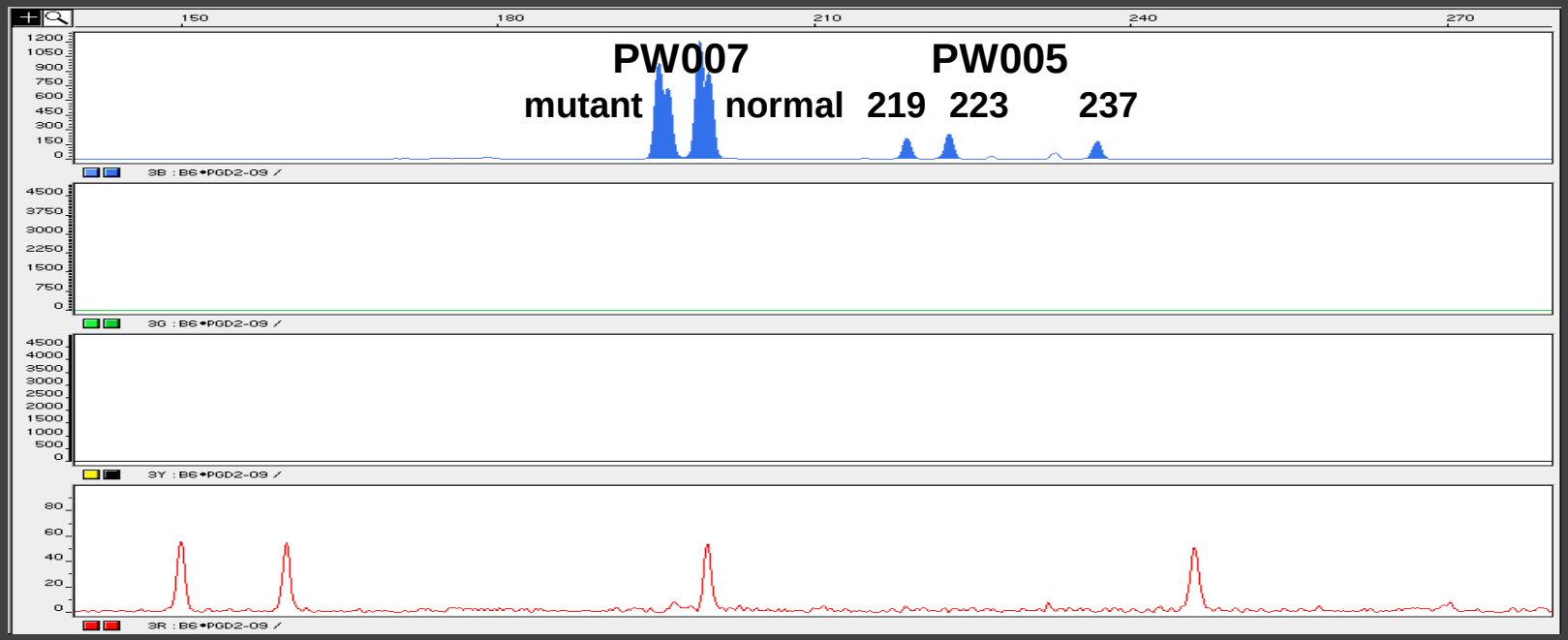




PGD of beta-Thalassaemia codon 41-42 Family B

Embryo 3 - 8-cell

: Heterozygous with trisomy 21



FIRST BIRTH FOLLOWING PREIMPLANTATION GENETIC DIAGNOSIS IN THAILAND: SUCCESSFUL DIAGNOSIS OF BETA-THALASSAEMIA AND SIMULTANEOUS DETECTION OF DOWN'S SYNDROME USING MULTIPLEX FLUORESCENT PCR

Wirawit Piyamongkol*, Teraporn Vutyavanich, Sirivipa Piyamongkol, Dagan Wells, Chairat Kunaviktikul, Theera Tongsong, Somsak Chaovisitsaree, Rattika Saetung, Torpong Sanguansermsri

A Successful Strategy for Preimplantation Genetic Diagnosis of beta-Thalassemia and Simultaneous Detection of Down's Syndrome Using Multiplex Fluorescent PCR

Wirawit Piyamongkol MD, PhD*,
Teraporn Vutyavanich MD, MMedSc*, Sirivipa Piyamongkol PhD**,
Dagan Wells PhD***, Chairat Kunaviktikul MD*,
Theera Tongsong MD*, Somsak Chaovisitsaree MD*,
Rattika Saetung****, Torpong Sanguansermsri MD****

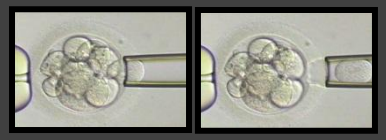
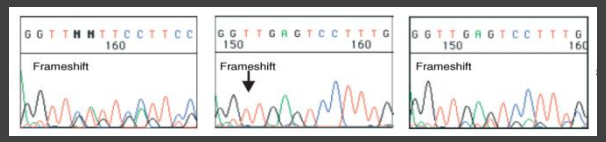
* *Department of Obstetrics and Gynaecology, Faculty of Medicine, Chiang Mai University, Chiang Mai*
** *Department of Pharmaceutical Sciences, Faculty of Pharmacy, Chiang Mai University, Chiang Mai*
*** *Department of Obstetrics, Gynecology & Reproductive Sciences, Yale University, New Haven, USA*
**** *Department of Pediatrics, Faculty of Medicine, Chiang Mai University, Chiang Mai*

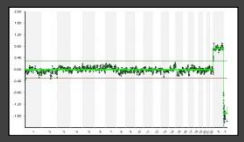
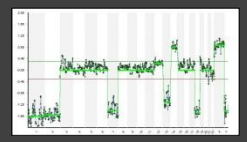
Objectives: *Preimplantation Genetic Diagnosis (PGD) is an alternative to prenatal diagnosis providing couples the chance to start a pregnancy with an unaffected fetus. The objective of the present study was to develop and apply quick, sensitive and accurate single cell PCR protocols for PGD of beta-thalassemia and Down's syndrome detection.*

Material and Method: *Two couples carrying beta-thalassemia codon41-42 mutation underwent routine IVF procedures. Embryo biopsy was performed on Day-3 post-fertilisation and single cell multiplex fluorescent PCR was employed for mutation analysis, contamination detection and diagnosis of trisomy 21 cases.*

Results: *Seventeen embryos were tested in two clinical PGD cycles. This resulted in the first birth following PGD for a single gene disorder in Thailand and South East Asia, confirmed by prenatal testing. Two embryos were shown to be affected by Down's syndrome.*

Conclusion: *Successful strategy for PGD of beta-thalassemia and Down's syndrome detection using multiplex fluorescent PCR was introduced.*

PGD of alpha-Thal^{-SEA} : 1st cycle

- 14 oocytes collected
- 14 embryos biopsied
- Results:
  - 4 normal (2 suggestive of Ht by LA1)
  - 2 heterozygous (Ht)
  - 6 affected (1 suggestive of Ht by LA1)
  - 2 with no result
- 1 normal + 2 heterozygous - ET
- no pregnancy resulted