

21<sup>ST</sup>



# PGDIS CONFERENCE



6-8 May 2024  
Kuala Lumpur  
Malaysia

PGT and  
BEYOND...



# Quality in the PGT Laboratory – Assessing the Referring Clinic Performances

Mohamed Saleem, MSc (Haem); PhD (Med Genetics)



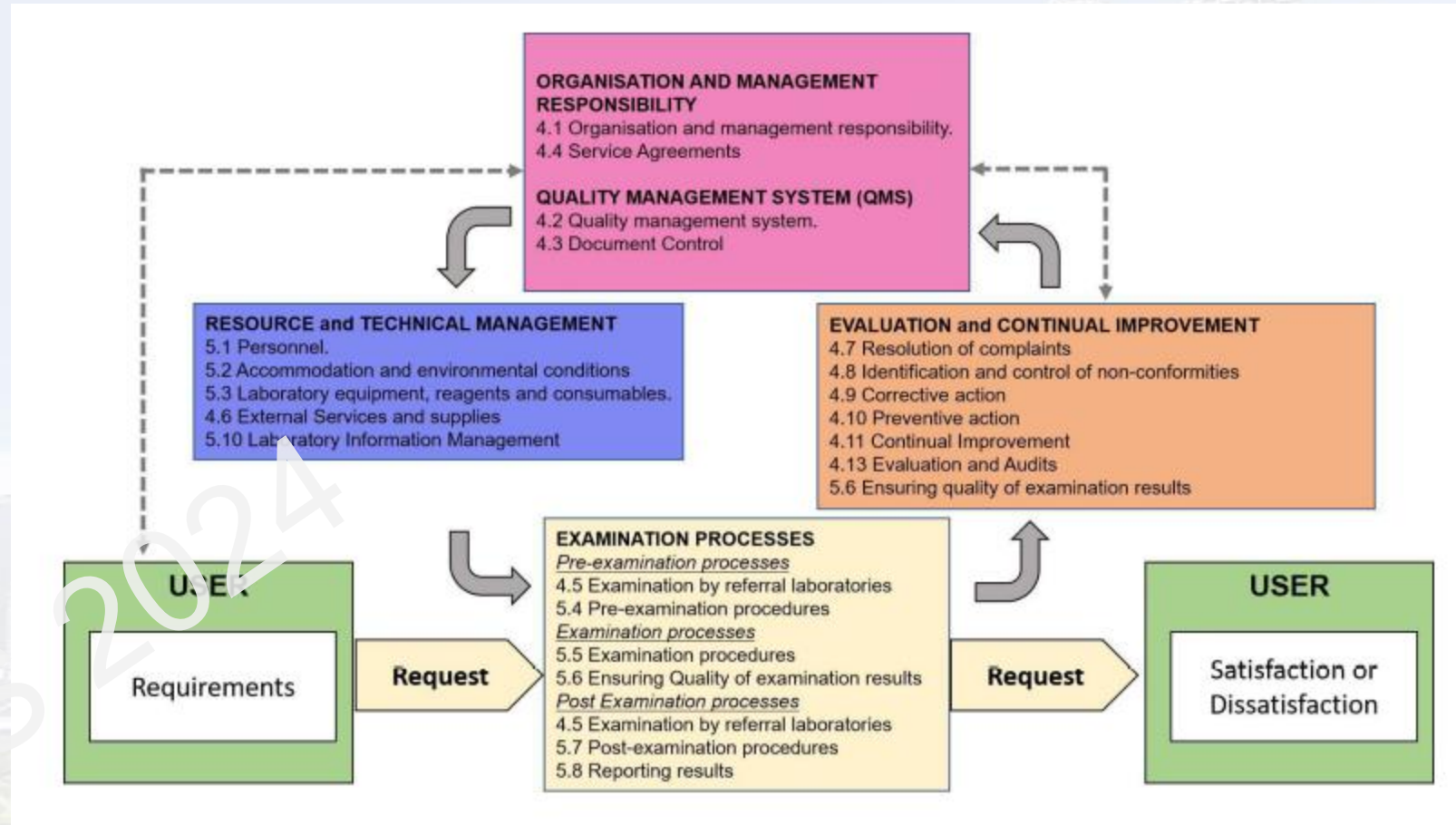


# Quality Management System

- Essential prerequisite – QMS

QMS consist of a series of processes, defined within a Quality Manual, which when executed in the correct sequence allows to meet the requirements of a Quality Policy to meet the requirement of the users.

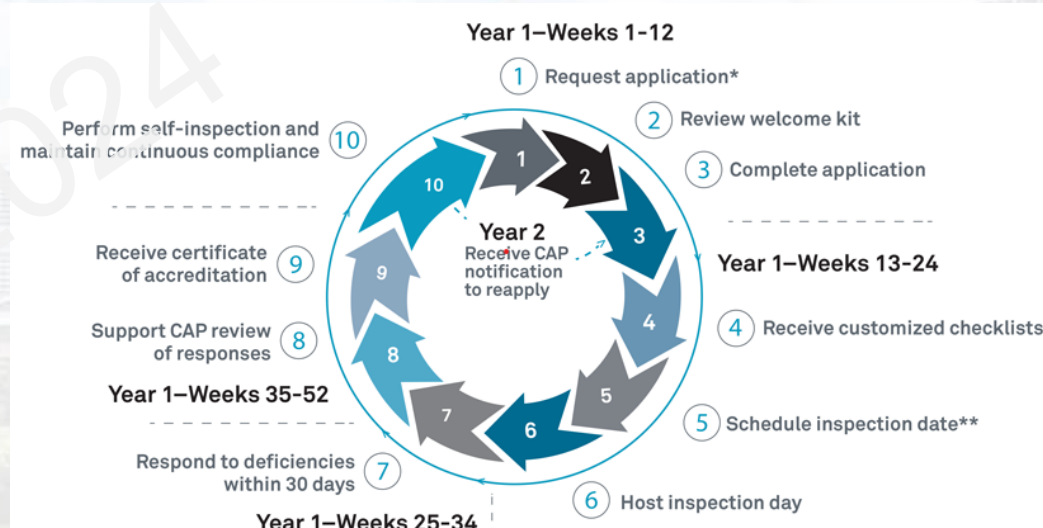
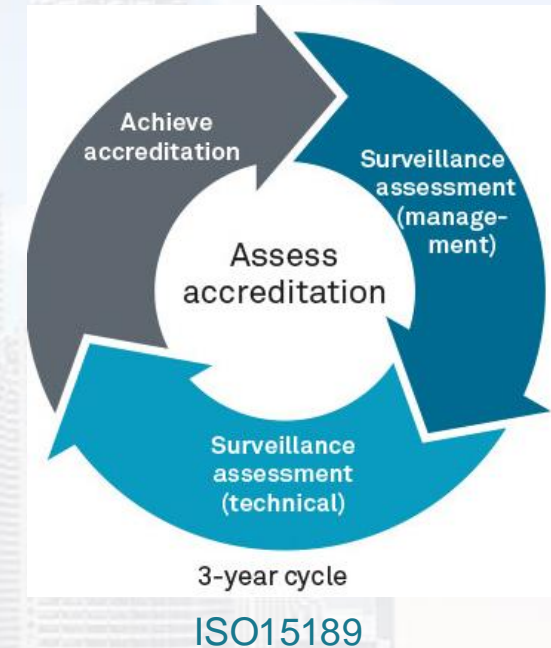
# Quality Management Processes



# Accreditation of the PGT/Genetic lab

Medical Laboratories—Requirements for quality and competency

- ISO15189
- CAP
- Any other



CAP- onsite inspection occurs every two years



# Proficiency Testing in PGT lab

- GenQA (~1500GBP) - UK
  - PGT-A
  - PGT-M
  - PGT-SR
  - Cycles x1
  - Submission of a structured report (ISO15189/ESHRE/PGDIS)
- AAB (USA)
  - PGT-A (USD490)
- ?PGDIS

# Referral Services

## Setting and monitoring KPI

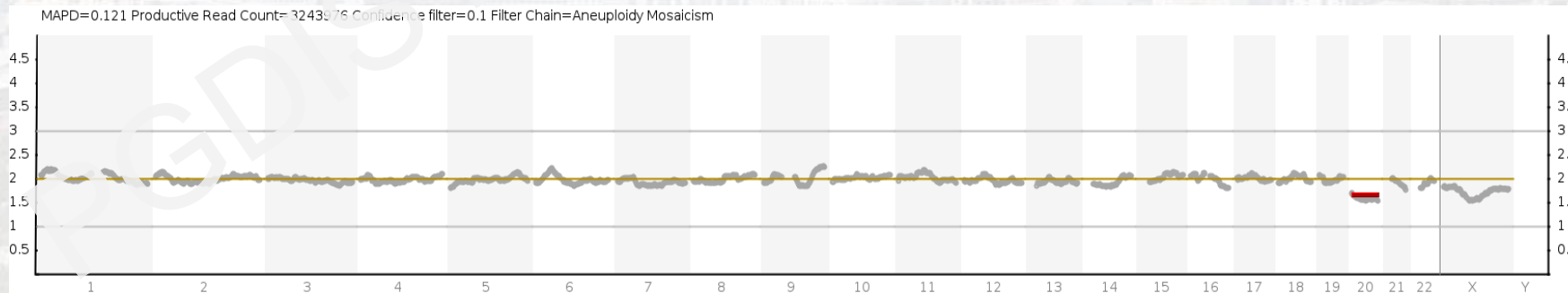
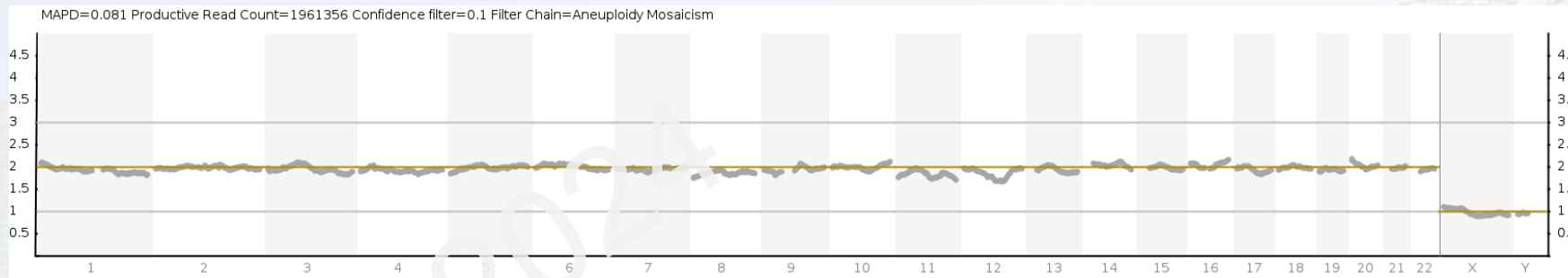
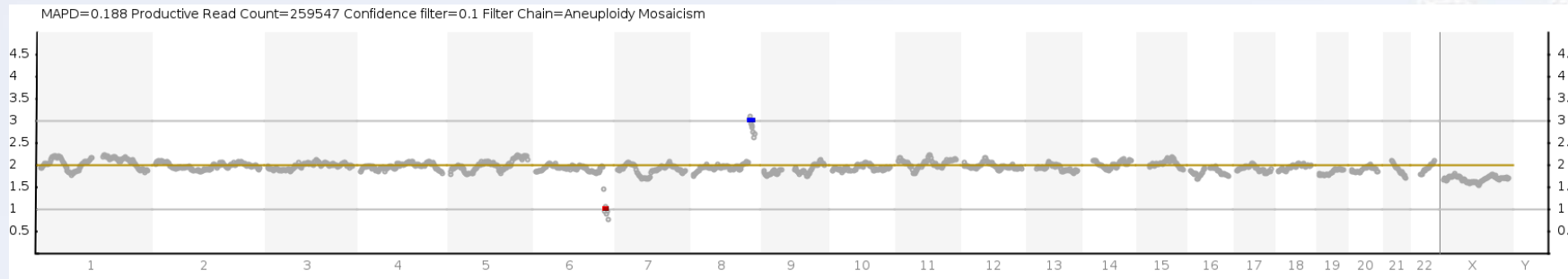


# Shipping validation

- How is it done?
- Source of cells – cultured amniocytes/ cultured cells from POC
  - KT and LPWGS confirmed (Euploid and Aneuploid) cells
  - Cell mixing - euploid + aneuploid amniocytes in 40-60% proportions (process controls – mosaicism)
  - Return courier - 48-72 hours
  - Run PGTA protocol



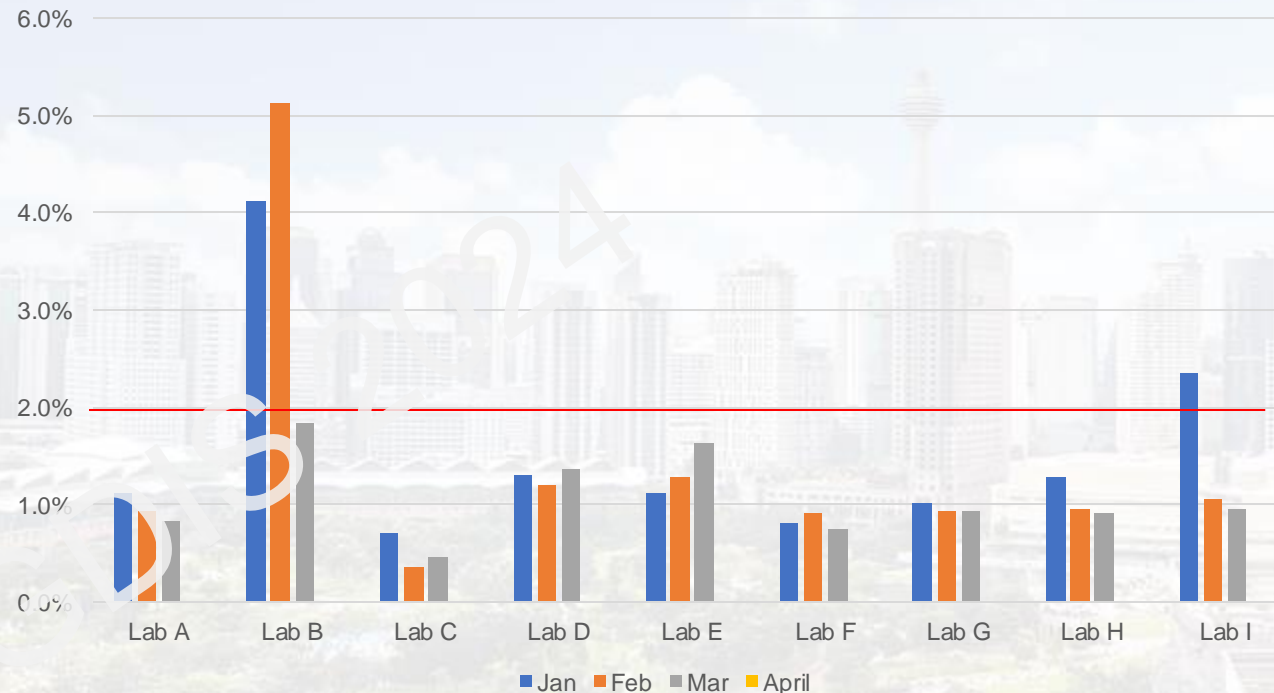
# Amniotic fluid culture cells – Returned Courier



# Amplification Failure - KPI

- Inappropriate tubing manipulations/degraded DNA can lead to amplification failure or inconclusive results

Amplification Failure 2024

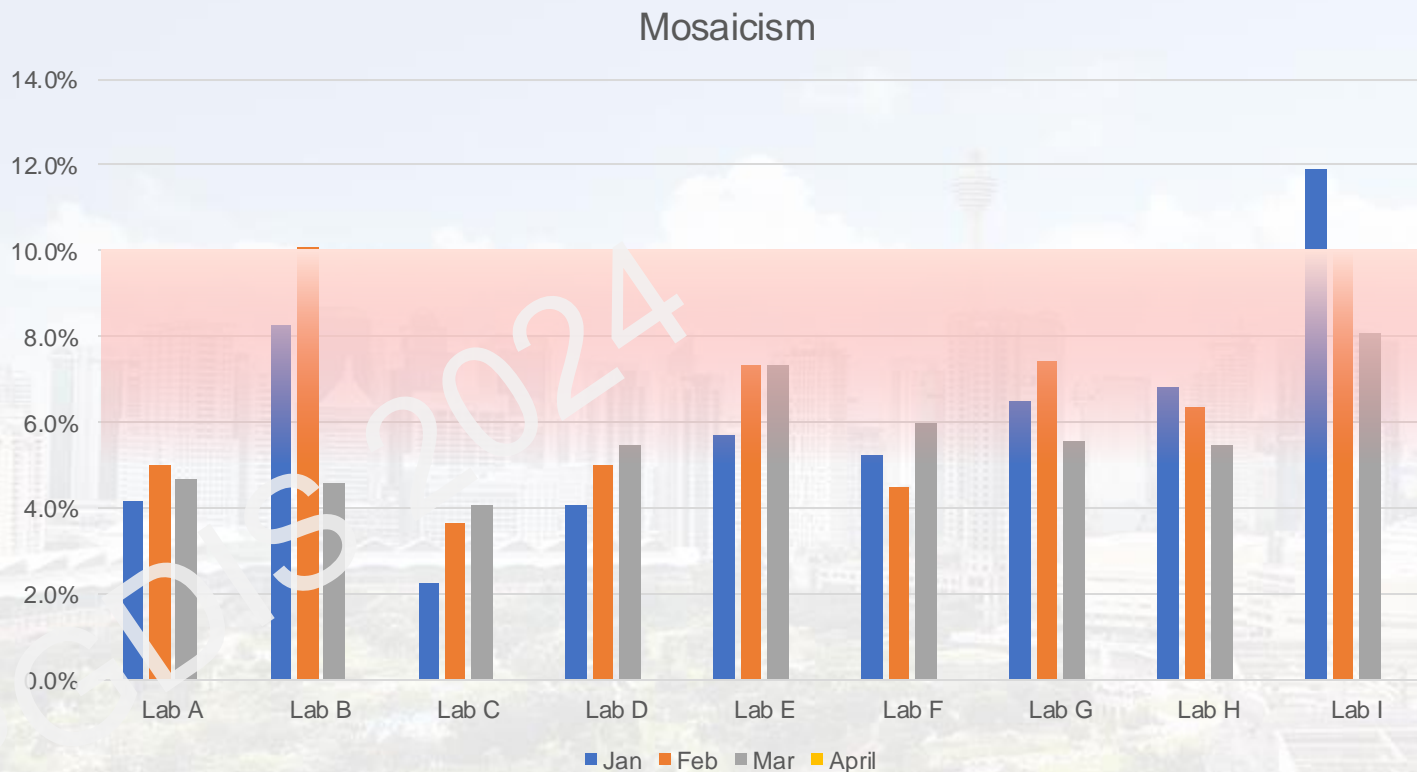


- Failed amplification <2%
- Re-biopsy recommended if required



# Mosaicism - KPI

- The rate of mosaic diagnoses in clinical testing of trophectoderm is estimated to be 3% to 20% depending on the specific NGS platform used.



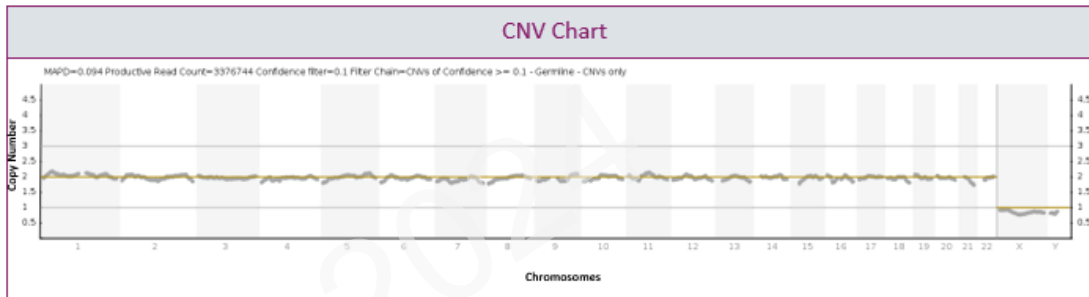
- Mosaic KPI <10%
- Investigation shows single operators take longer time >30 minutes to wash TE biopsy, tube and freeze the biopsy sample

# Reporting Error -KPI

## Pre-Implantation Genetic Testing-Aneuploidy (PGT-A) By NGS - Detail Report

Embryo ID	DM1	No of cells Biopsied	~8
Sample Type	Trophectoderm	Embryo Grade	5BA

Result	46,XY
Transfer Recommendation	Transfer Recommended



Chromosomes	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	X	Y
Imbalances	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N	N

N — Normal, G — Gain; L — Loss; M — Mosaic; SL — Segmental Loss; SG — Segmental Gain; I — Inconclusive

Interpretation/Comments:	Aneuploidy NOT detected. Transfer recommended.
--------------------------	--

- Error <0.1%
- Where possible follow ISCN2020 guideline



# Reporting

- IVF Success rate varies between Centres
- What are the factors and KPIs
- Help to Improve

# Summary

- Comprehensive QMS is essential
- Accreditation of the Genetic lab is essential
- A set of KPI and PI required to assess referring laboratory



# THANK YOU

**PGT and  
BEYOND...**