Reference Materials for Genetic Testing

Gonda Verpooten, R&D Group Manager Genetic Testing
Molecular Genetic Testing Products
Polymorphic genes / Different (rare) mutations

Test Principle
Sample/DNA extraction
PCR
Hybridization/detection

Transplantation
- LiPA HLA-A
- LiPA HLA-B
- LiPA HLA-C
- LiPA HLA-DRB
- LiPA HLA-DQB
- LiPA HLA-DPB
- LiPA HLA-DQA

Cystic Fibrosis
- LiPA CFTR36
- LiPA CFTR Italian
Development of Diagnostic Tests

• Assurance of safety and effectiveness
  – Quality and validity: analytical performance
  – Accuracy (right answer), precision (right every time)

• Time to market

♫ Accessibility of appropriate and verified reference material
Analytical Performance

• Hybridization
  – secondary structures
  – concentration
  – heterozygous/homozygous
  Synthetic samples, (Real samples)

• PCR
  – whole genome
  – concentration, purity
  – matrix, inhibitors
  Real samples, (Synthetic samples)
Reference Material: Wish List

- Samples (in decreasing order of needs)
  - Real samples
  - Cell lines
  - DNA
- (Synthetic) matrices
- Databank
  - IEC/IRB approval
  - Informed consent (as many genes as possible)
  - Well-characterized samples
  - Accessible