



Technology Transition Workshop | *Jodee Steinberg, M.S.*

***Analysis of Genetic and Epigenetic
Variation Using MassARRAY[®]
Compact from Sequenom[®]***

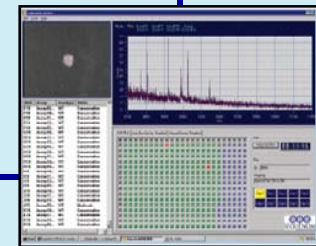
MassARRAY® Platform

Biochemistry

SpectroCHIP®

Mass Spectrometry

Data Analysis



Genotyping

Quantitative Gene Analysis

Comparative Sequencing

- hME (1-15 plex)

- iPLEX® (1-29 plex)

- iPLEX® GOLD (1-40 plex)

- Quantitative gene expression

- Allelotyping

- Copy number variation

- EpiTYPER™ (Methylation)

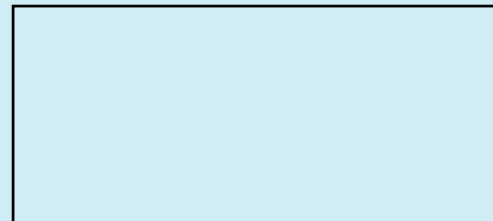
- iSEQ™ (Pathogen typing)

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Matrix Assisted Laser Desorption Ionization Time of Flight Mass Spectrometry (MALDI-TOF MS)

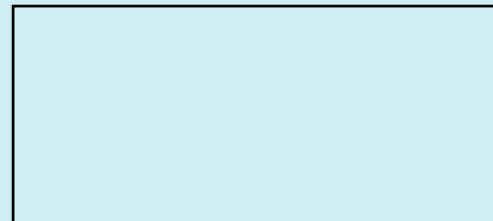
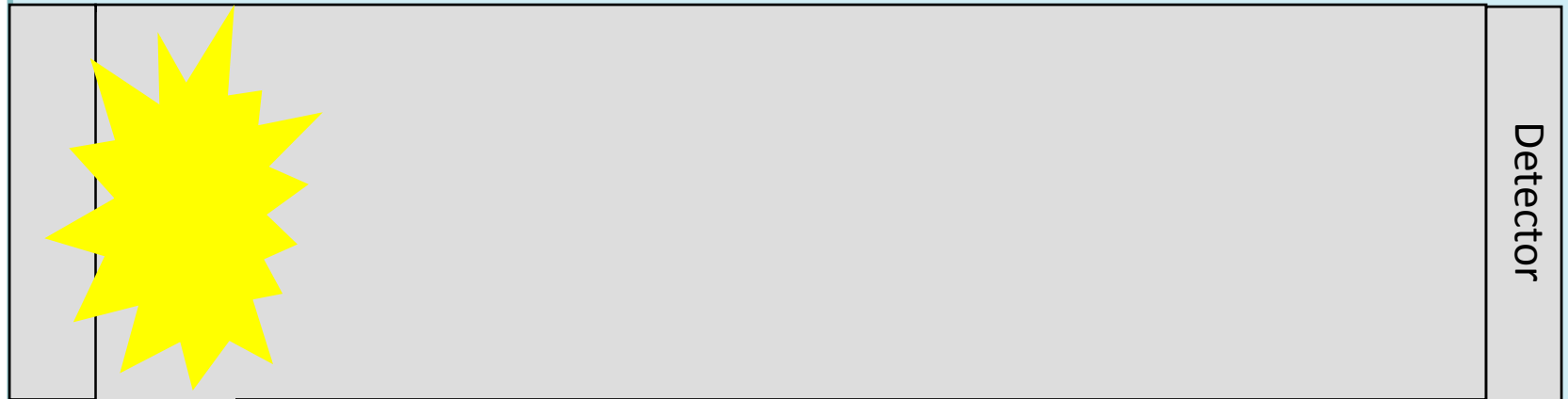
Matrix/Analyte



Mass Spectrum m/z

Matrix Assisted Laser Desorption Ionization Time of Flight Mass Spectrometry (MALDI-TOF MS)

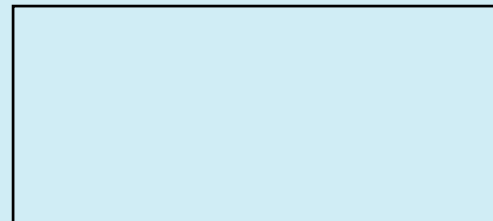
Laser Desorption and Ionization



Mass Spectrum m/z

Matrix Assisted Laser Desorption Ionization Time of Flight Mass Spectrometry (MALDI-TOF MS)

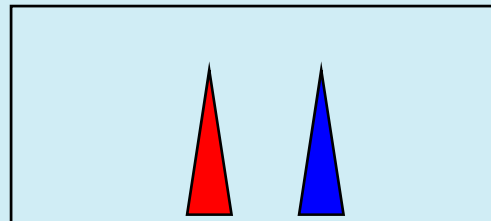
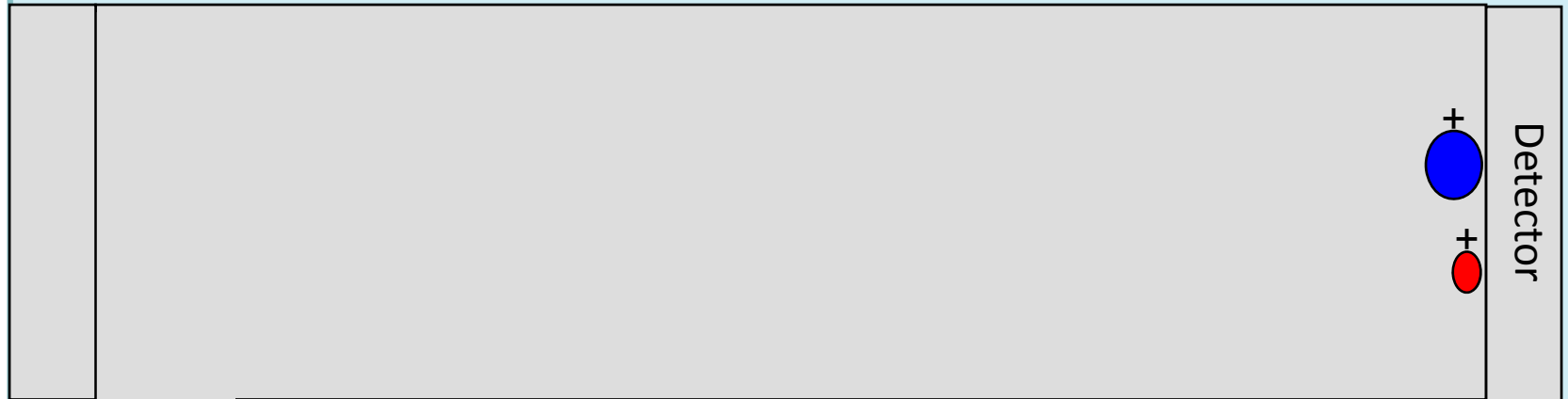
Laser Desorption and Ionization



Mass Spectrum m/z

Matrix Assisted Laser Desorption Ionization Time of Flight Mass Spectrometry (MALDI-TOF MS)

Acceleration and Detection



Mass Spectrum m/z



iPLEX[®] Gold

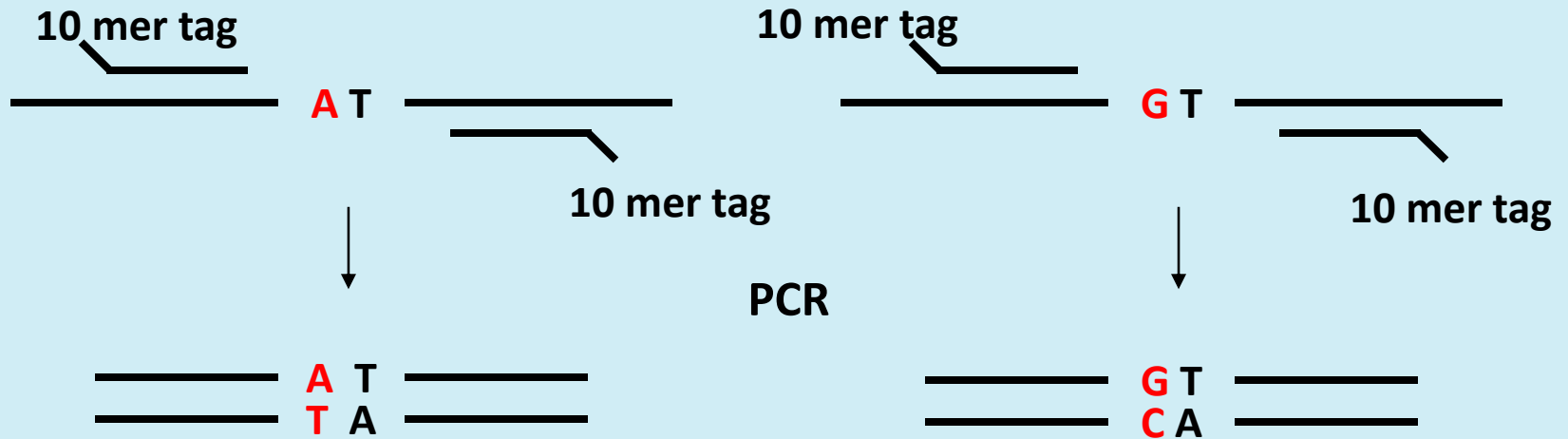
The Finest in Fine Mapping

SEQUENOM[®]

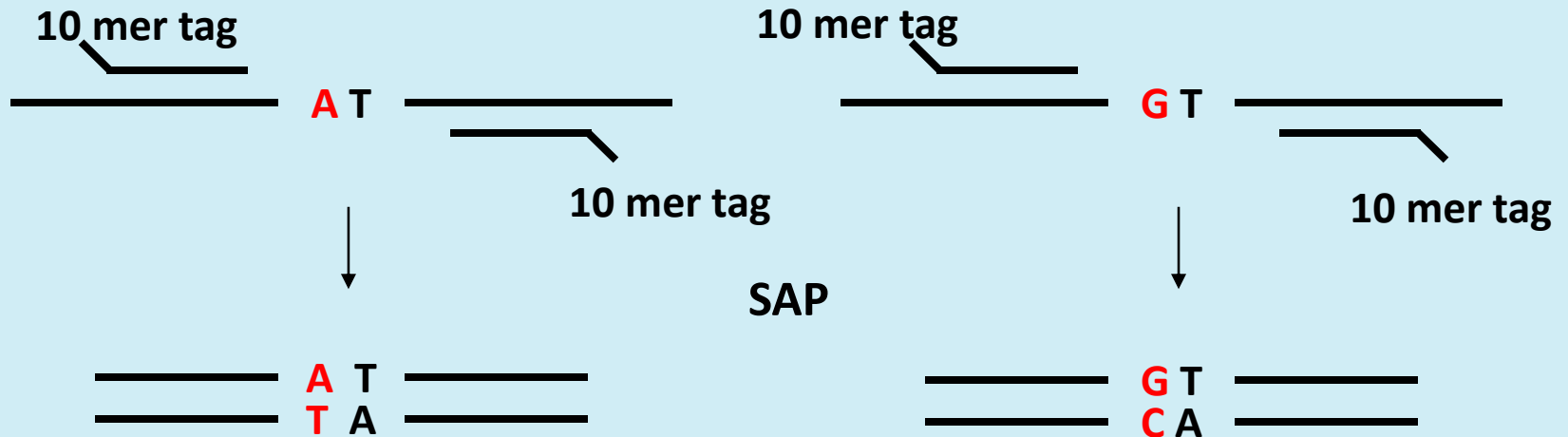
www.sequenom.com

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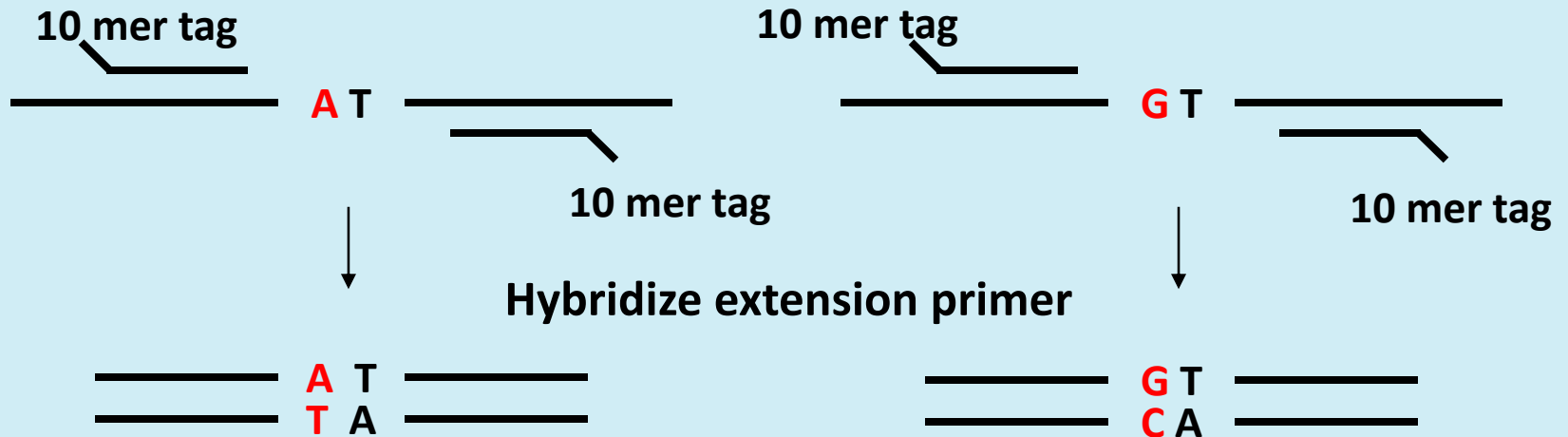
iPLEX[®] **Gold** Genotyping Assay



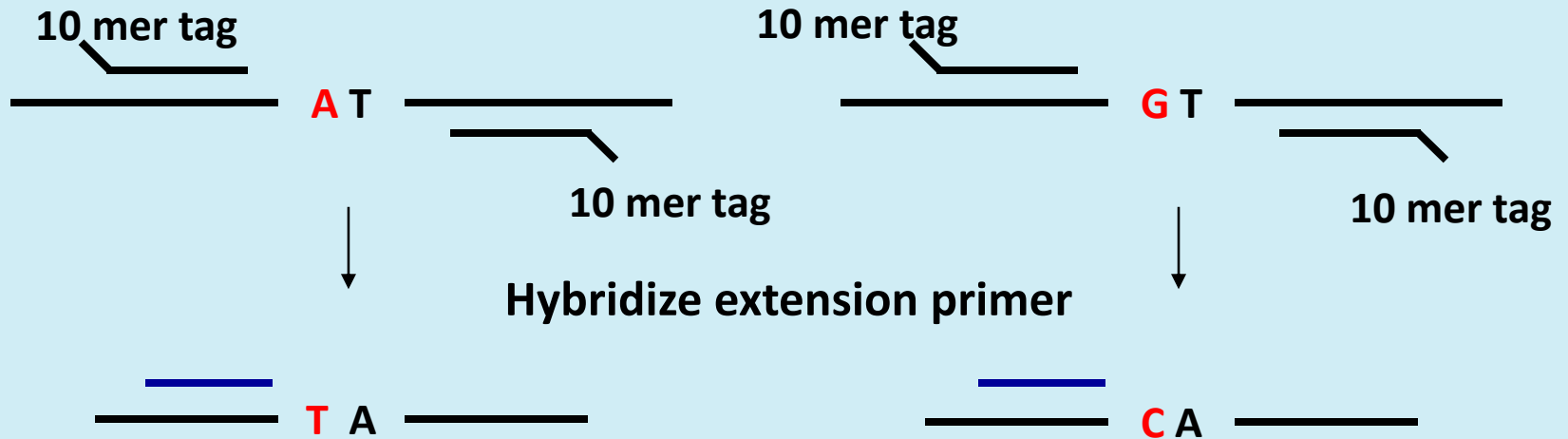
iPLEX[®] **Gold** Genotyping Assay



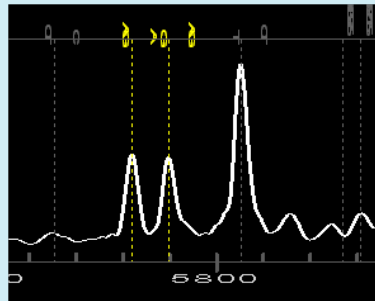
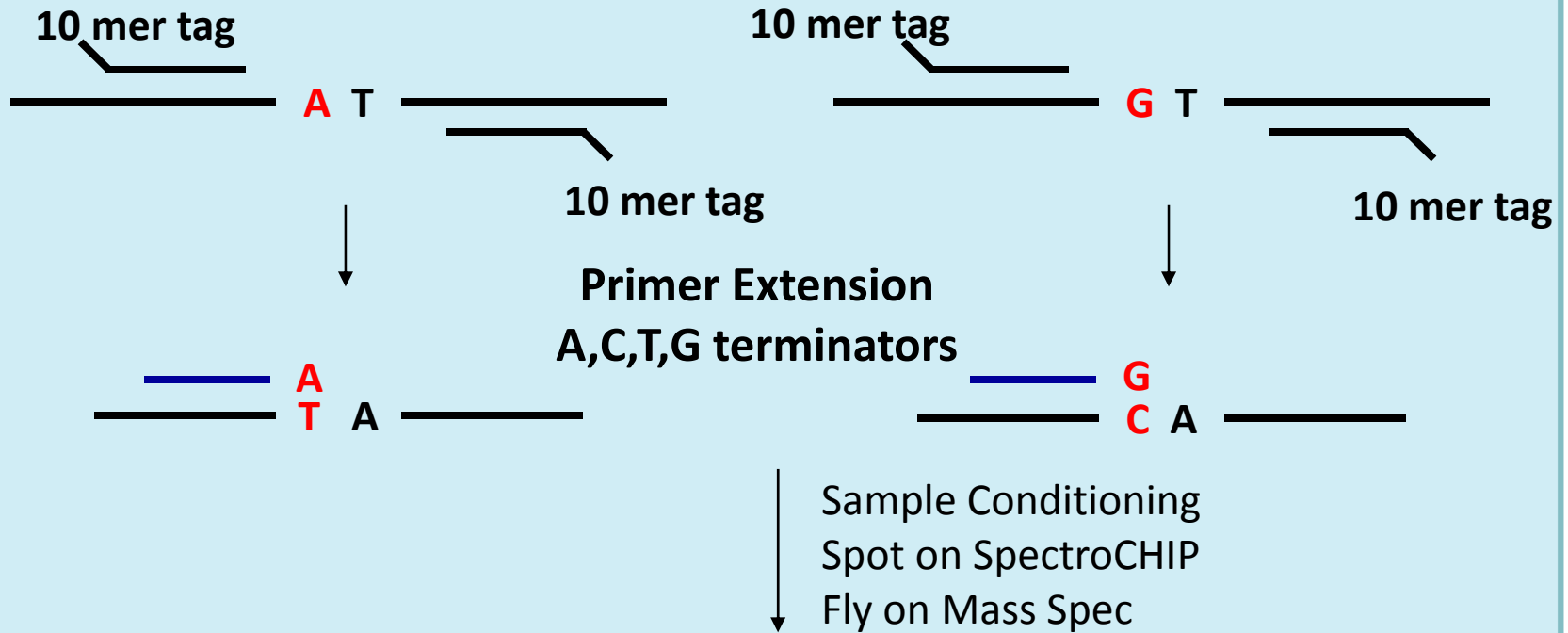
iPLEX[®] **Gold** Genotyping Assay



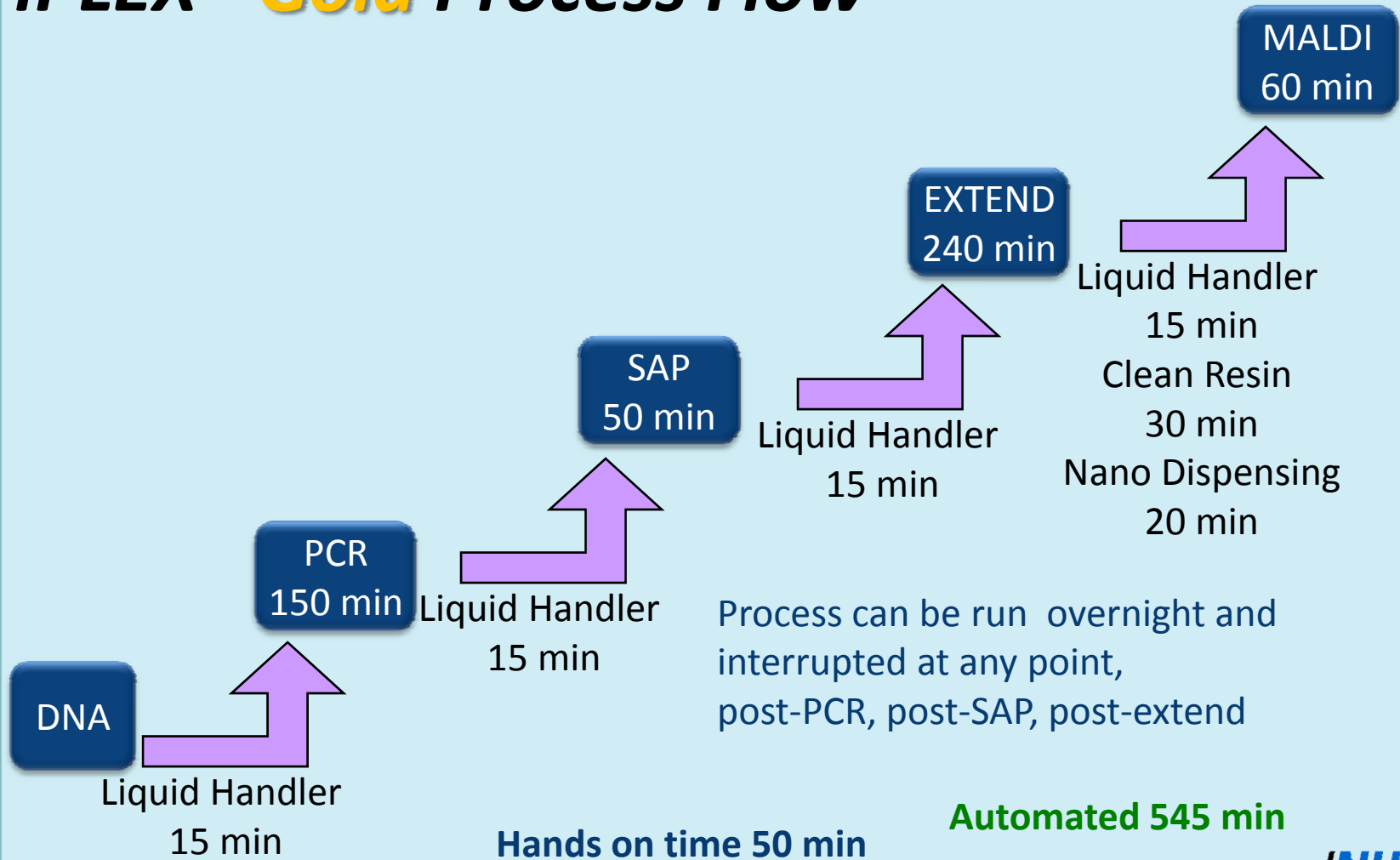
iPLEX[®] **Gold** Genotyping Assay



iPLEX[®] Gold Genotyping Assay



iPLEX[®] Gold Process Flow



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DNA Sample Types Genotyped by iPLEX®

If you can successfully PCR your template, our technology can successfully analyze it.

- **DNA extracted from blood**
- **DNA from formalin fixed and frozen tissue samples**
- **DNA isolated from blood plasma and serum**
- **DNA from FTA cards**
- **Whole genome amplified DNA**
- **Buccal cells**
- **DNA from ear punches**
- **DNA from semen**
- **DNA from plants**

iPLEX[®] Gold Throughput

- 1 CHIP every hour
- 384 samples per CHIP
- Up to 40 SNPs per sample

		CHIPS					
		1/384	1	2	6	8	10
Plex Level Genotypes/Rxn	6	6	2,304	4,608	13,824	18,432	23,040
	12	12	4,608	9,216	27,648	36,864	46,080
	24	24	9,216	18,432	55,296	73,728	92,160
	36	36	13,824	27,648	82,944	110,592	138,240

Flexibility of platform allows the running of partial CHIPS for small projects

Flexibility of iPLEX[®] Gold Genotyping Platform

Panel selection

Select SNPs for panel



Assays multiplexed 32-40 plex

85%-95% conversion rate



Plex 1



Plex 2



Plex 3



Reconfigure panel

Flexibility of iPLEX[®] Gold Genotyping Platform

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Reconfigure panel

Remove old assays

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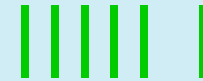
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Reconfigure panel

Remove old assays

Add additional new assays



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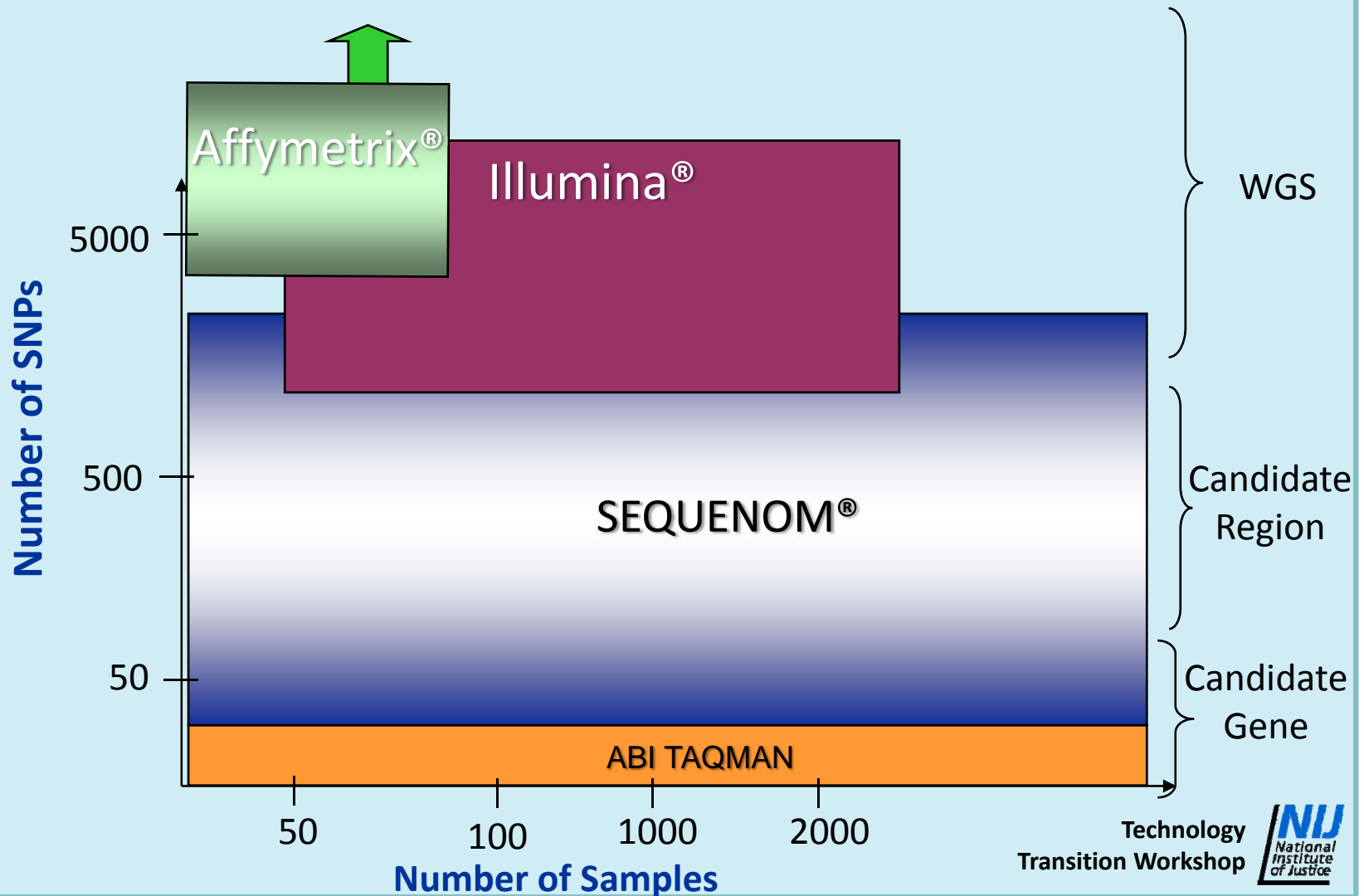
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Genotyping Analysis Technology Dynamics



MassARRAY iPLEX[®] Gold Advantages

- **Accurate/reliable**
 - 95 to 99% call rate on optimized assays
 - 99.9% accuracy
- **Flexible**
 - Assay design user controlled
 - Assays can be reshuffled and exchanged
- **Ease of use**
 - Universal assay conditions and reagents
 - Single well homogeneous assay

MassARRAY iPLEX[®] Gold Advantages

- **Cost effective**
 - Low cost per genotype at from 5 to 40 plex level
 - Unmodified primers – low sample number
- **Works on low quality DNA (e.g. formalin-fixed paraffin-embedded tissue)**
- **Quantitative genotyping**

Questions?

Contact Information

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Note: All images are courtesy of Sequenom®.