



# Clinical implications of SNP Arrays & Next generation sequencing technologies

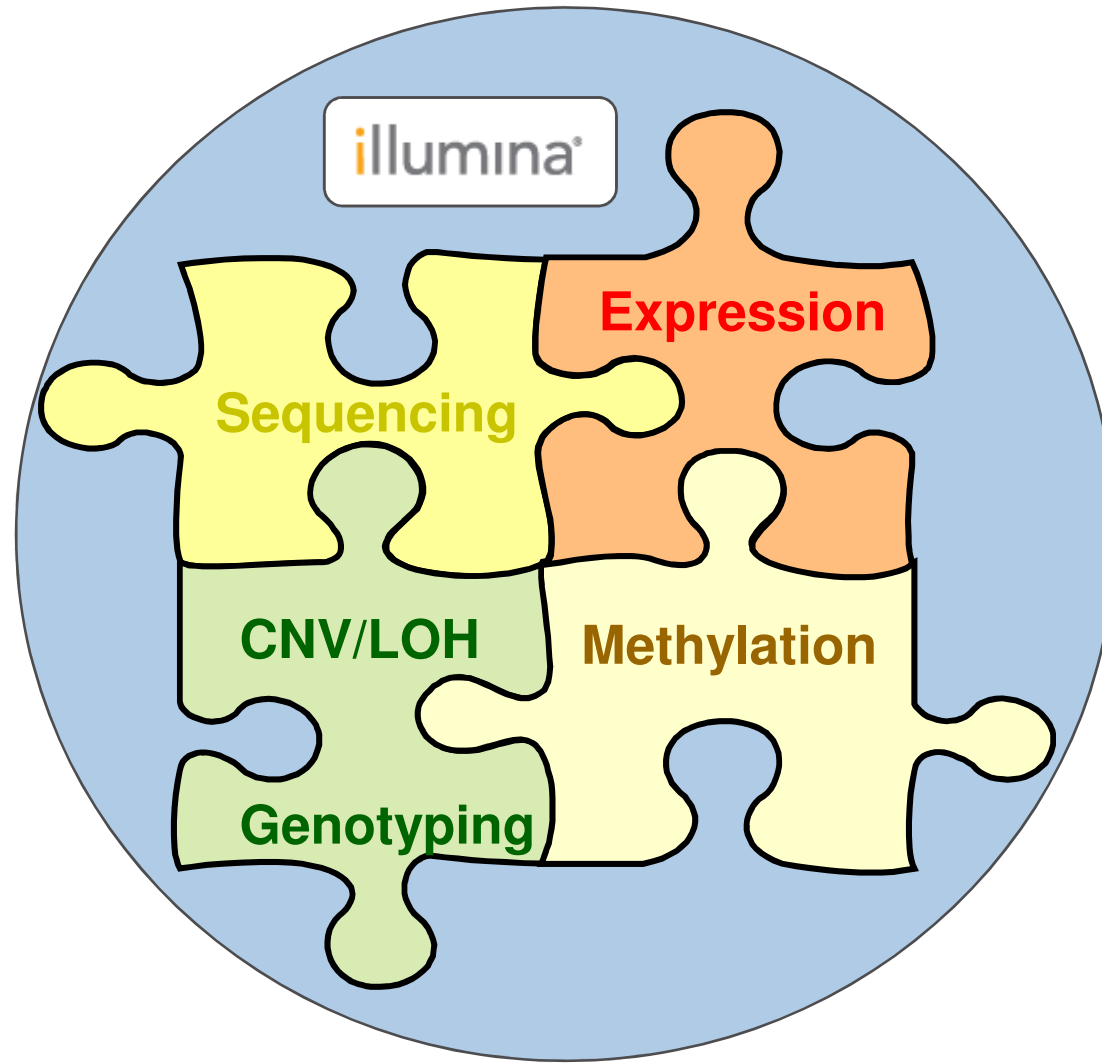
## SEMINAR SCHEDULE

09.15	Registration – Welcome Coffee
	Introduction to Agenda
10.00	Caroline Thureau, Diagnostics Illumina
10.10	What's New in Sequencing : A Sequencing System adapted to your Needs Marco Cappelletti, Illumina
	A genomic approach to Sudden Cardiac Death
10.50	Richard Redon, Ph.D, INSERM, Nantes France
11.30	Integrating Clonal Sequencing into a Healthcare Diagnostic Setting Graham Taylor, PhD FRCPATH, IMM Leeds UK
	Exome Sequencing as a promise for next generation clinical research
12.10	Emmanuel Martin, PhD, IntegraGen, Evry France
12.50	Buffet Lunch
	An overview of Illumina's Microarrays Product used in Clinical Settings
14.30	Caroline Thureau, Diagnostics Illumina
15.00	Can we use microarray-based profiling in a routine workflow for the diagnostic / prognostic of everyday patients with acute myeloid leukaemia? Philippe Guardiola, Ph.D., Transcriptome & Epigenomics Angers University Hospital, France
	Analysis of patients suffering from mental deficiency & epileptic encephalopathy using Illumina SNPs arrays
15.40	Boris Keren & Christel Depienne, Ph.D, Hospital Pitie Salpêtrière, Paris France
16.20	Conclusion





# One Provider – Solutions for Different Applications



# From discovery to single target Validation

## Sequencing & Arrays Platform Synergy

Discovery

Focused Research/Validation

Screening

$>10^9$

$10^8$

$10^7$

$10^6$

$10^5$

$10^4$

$10^3$

$10^2$

10

1

Whole Genome

Single Marker



Genome Analyzer

- WG Resequencing
- WG Gene Expr.
- miRNA Discovery & Profiling
- WG ChIP-Seq
- **Candidate Resequencing**
- **Chromosome sequencing**



iScan

- Gene Expr.
- WG GT
- CNV Screening
- Biomarker Discovery
- **HT Biomarker Validation**
- Custom GT
- Focused Expr.
- **FFPE**



Sequencing Enabled iScan



BeadXpress

- Low/mid density GT/GEX
- Protein applications
- **Biomarker panels: ADME**
- **Clinical Diagnostics**

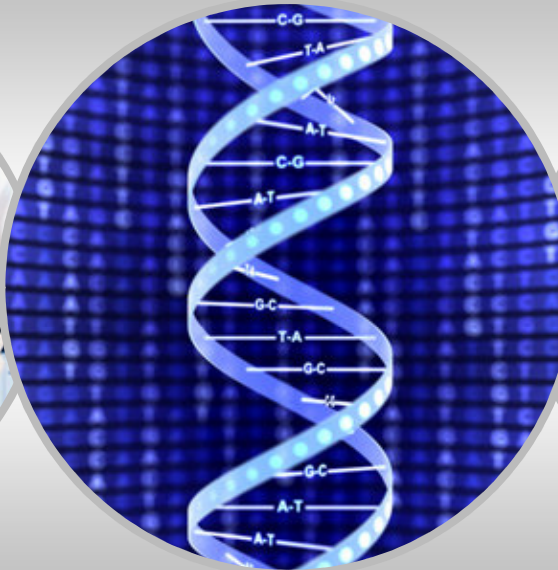




# Mission

*Innovating for the Future of Genetic Analysis*

From Genome Wide Discovery...



To Targeted Validation and Beyond

illumina's mission is to be the leading provider of integrated solutions that advance the understanding of genetics and health.



# Ovarian Cancer Will be Our Entry Point

To Be the Leader in  
Cancer Translational Diagnostics

## Focus Core Technologies on Cancer

- Next Generation Sequencing
- Whole Transcriptome Analysis
- Methylation Profiling



# Oncology Discovery

## Ovarian Cancer

### Deadly

- 204,449 New cases annually; 124,860 deaths

### Incurable

- Less than 40% are cured

### Difficult Dx

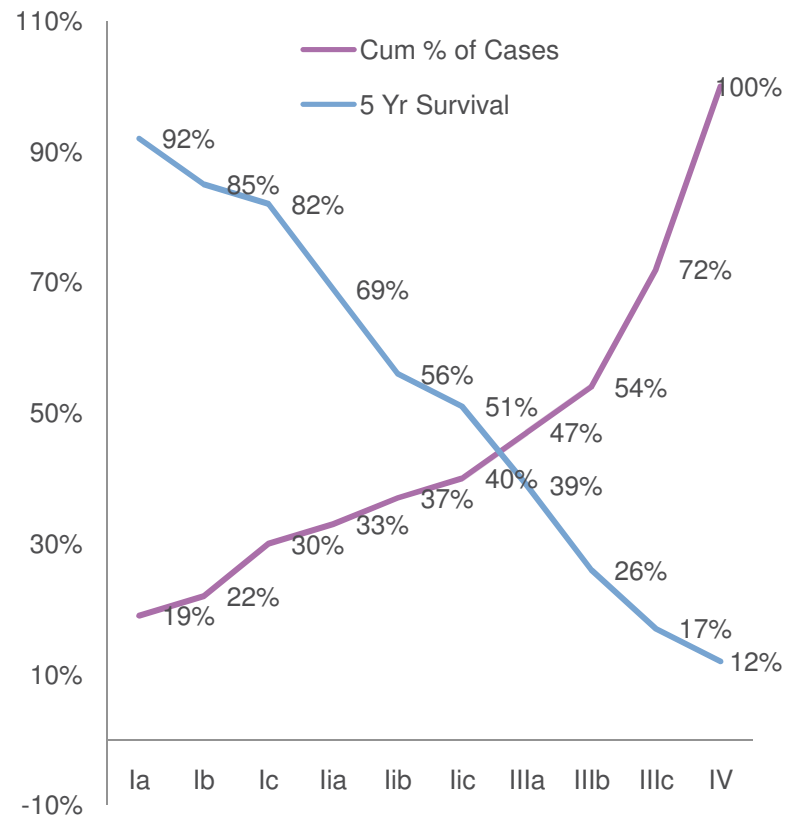
- Patients present with a suspicious/palpable mass

### Early Dx is Key

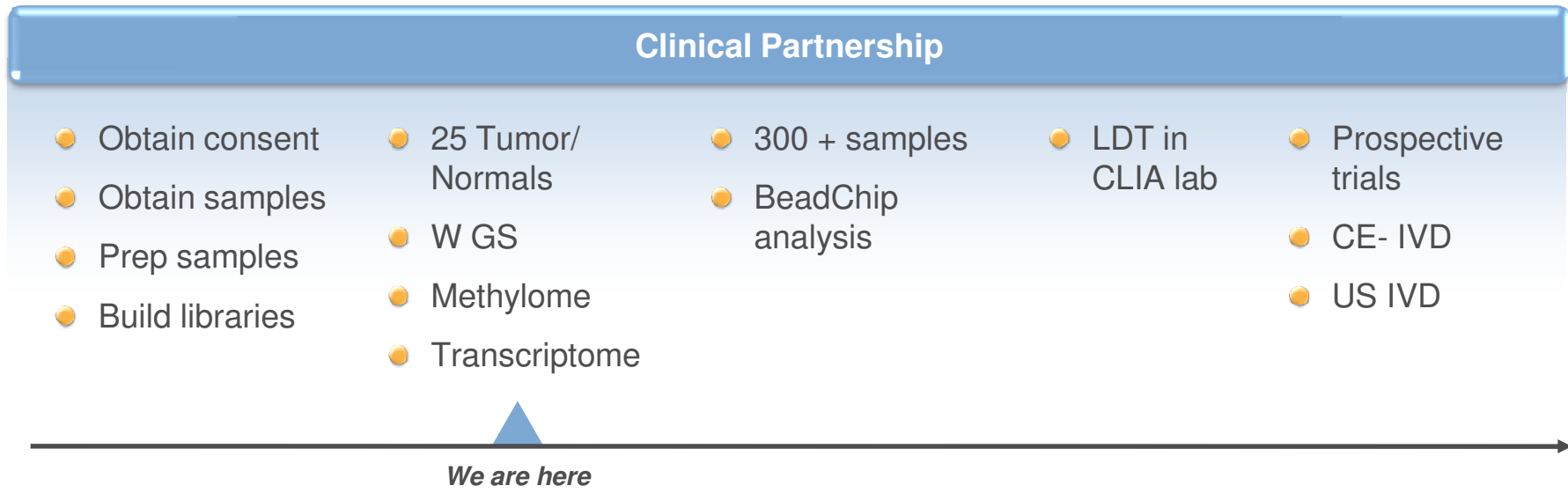
- Five year survival is good if diagnosed early, but most patients are diagnosed late stage

### Illumina Solution

- Develop a diagnostic assay which will diagnose ovarian cancer at an early stage



# Oncology Discovery Approach



# Illumina's First FDA Cleared In-Vitro Diagnostic Device

- The BeadXpress System is an **FDA 510(k) cleared In-Vitro Diagnostic Device**
- FDA Cleared BeadXpress System includes:
  - BeadXpress Reader
  - VeraScan Software
- The Intended Use Statement:
  - The BeadXpress® System is an In-Vitro Diagnostic Device intended for the simultaneous detection of multiple analytes in a DNA sample utilizing VeraCode holographic microbead technology. The BeadXpress System consists of the BeadXpress Reader and VeraScan software.



# Enabling Clinical Early Adopter



## FDA cleared BeadXpress System

Regulated device ensures quality and performance standards for clinical laboratories



## General Purpose Reagent Beads

Enables customers to develop their own assays using royalty-free beads



## Path to FDA Submissions

BeadXpress clearance and GMP reagents create a platform for follow-on diagnostic development



# Enabling Diagnostic Partners



*Signed agreements with EraGen:*

## Expanded Menu

*EraGen can port content to BeadXpress*

EraGen potential transfer menu includes:

- CFTR, Factor II/IV, MTHFR
- Respiratory Viral Panel
- CYP2C9 & VKORC1

## Improved Workflow

*Faster chemistry on BeadXpress*

- Reduced labor costs
- Faster turnaround
- Enables further menu expansion in infectious disease

***BeadXpress 510(k) Clearance Removes a Significant Hurdle for development of diagnostic tests by Partners***



# Supporting Illumina Diagnostic Assays in Development

- Multi-Drug Resistant Organism (MDRO) panel on BeadXpress using EraGen chemistry
  - MRSA, MSSA, VRE, KPC2, KPC3, CTX-M, Clindamycin resistance
  - **Clinical trials** start at end of year
- Viral Transplant Panel on BeadXpress using EraGen chemistry
  - **Herpes Panel** : CMV, EBV, VZV, HHV6, HSV 1 & 2
  - **Respiratory Viral Panel**: Flu A and B; PIV1, -2, -3, -4a and -4b; RSV A and B; HRV; CoV 229E, NL63 and OC43; Ad B, C, and E; HMPV
  - **Clinical trials** start at end of year



***Clinical trials and FDA submission included in development plans***



# Portfolio for Cytogenetic Analysis

*iScan*

- Q4 '09 Illumina submits Pre-IDE for Cytogenetics
- Package includes iScan platform, arrays, reagents and software
- Intended use
  - *The Infinium HD Cytogenetics Test is intended to be used as an aid in the postnatal diagnosis of chromosomal abnormalities known to be associated with developmental delay and mental retardation, including Prader-Willi, Angelman, Down, DiGeorge/VCS, and Klinefelter syndromes, in conjunction with other information currently used in postnatal diagnosis, consistent with professional standards of practice.*

KaryoStudio



HumanCytoSNP-12



- Optimized for cytogenetics

HumanOmni1-Quad



- High resolution detection of micro-deletions
- Efficient four-sample format

