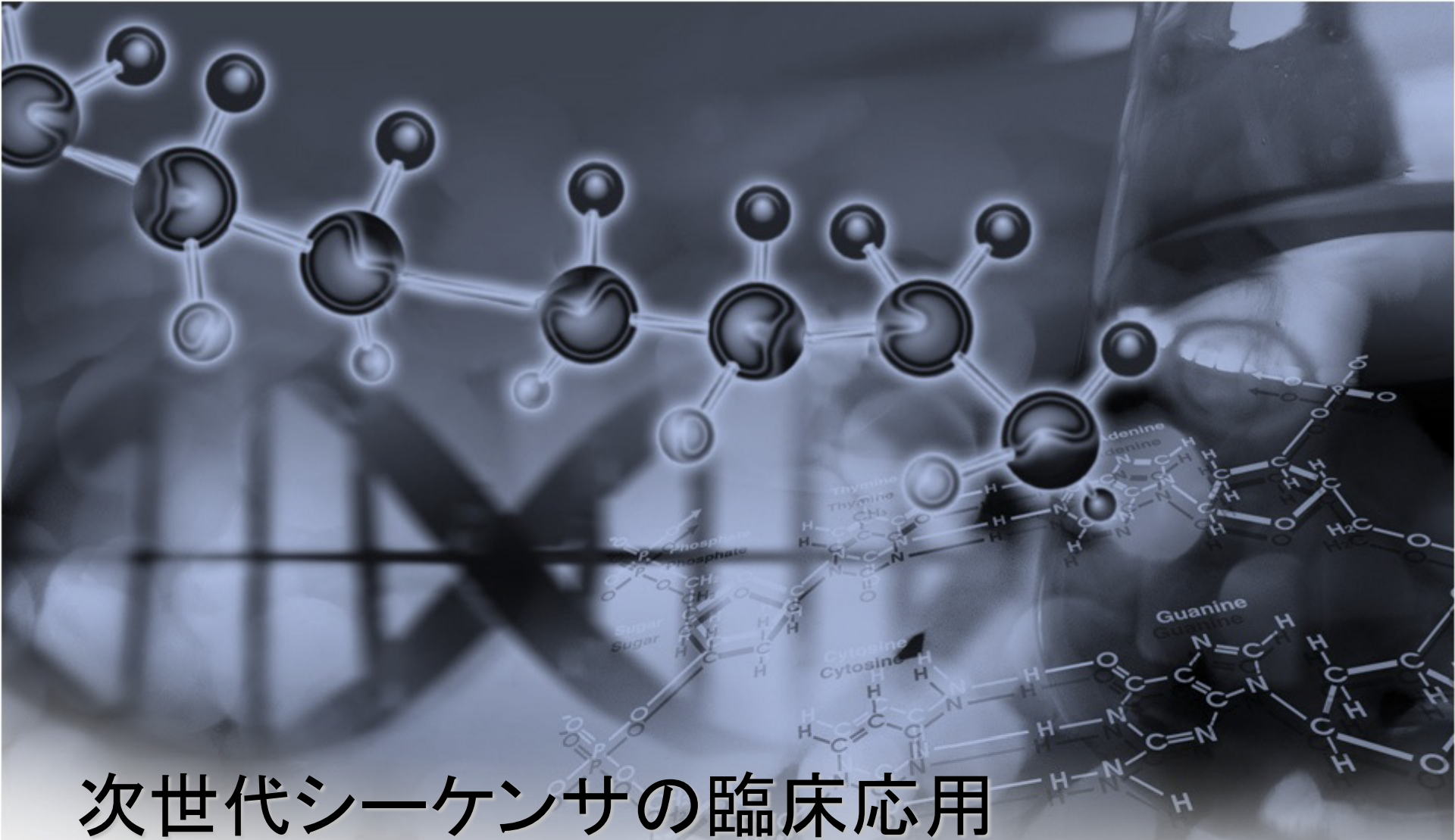

次世代シーケンサの臨床応用

ライフテクノロジーズジャパン

片山　稔

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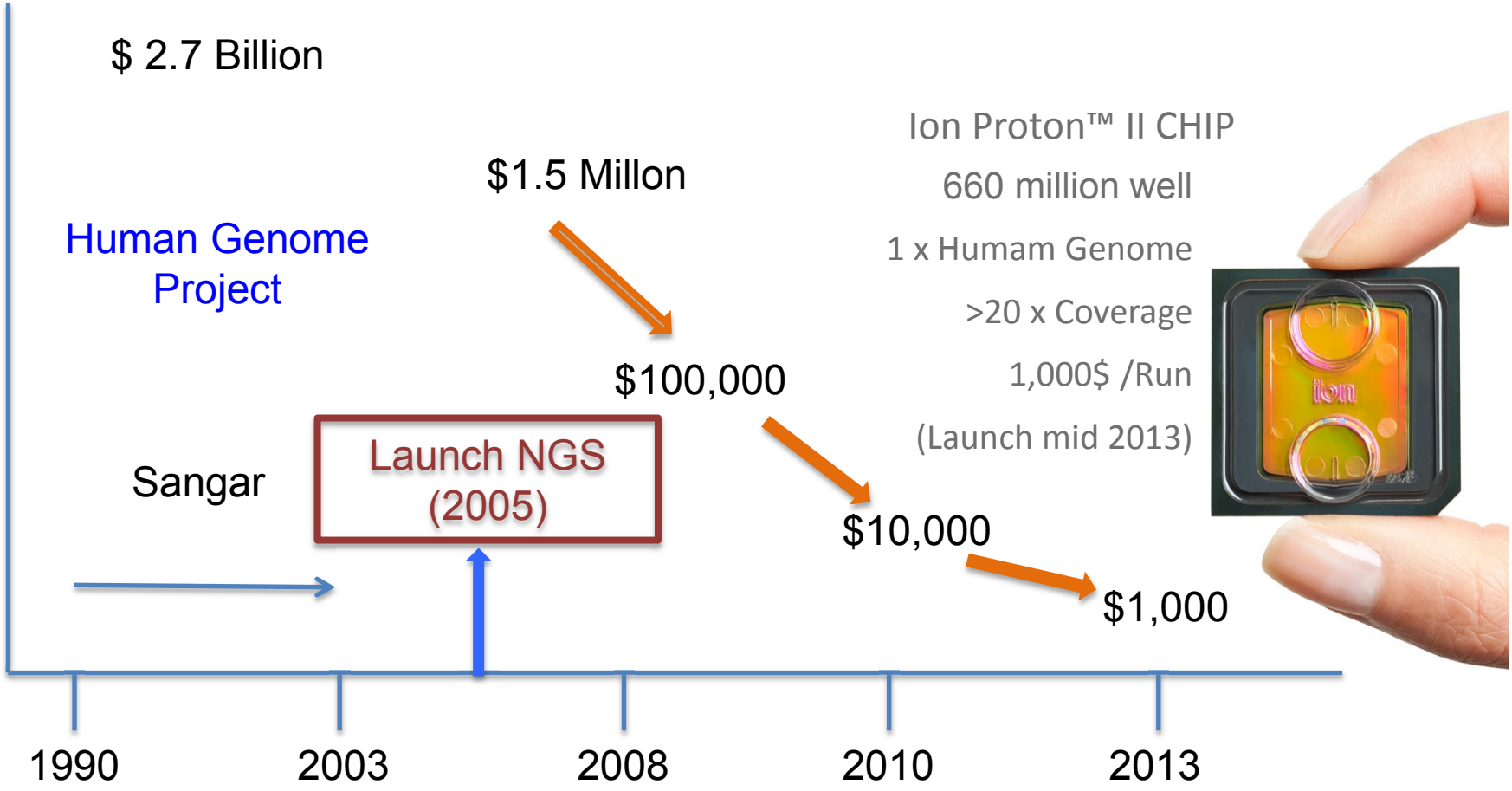
Cost for Human genome sequence



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次世代シーケンサが身近に

Ion PGM™

1Gbの、さらにその先へ

Ion Proton™

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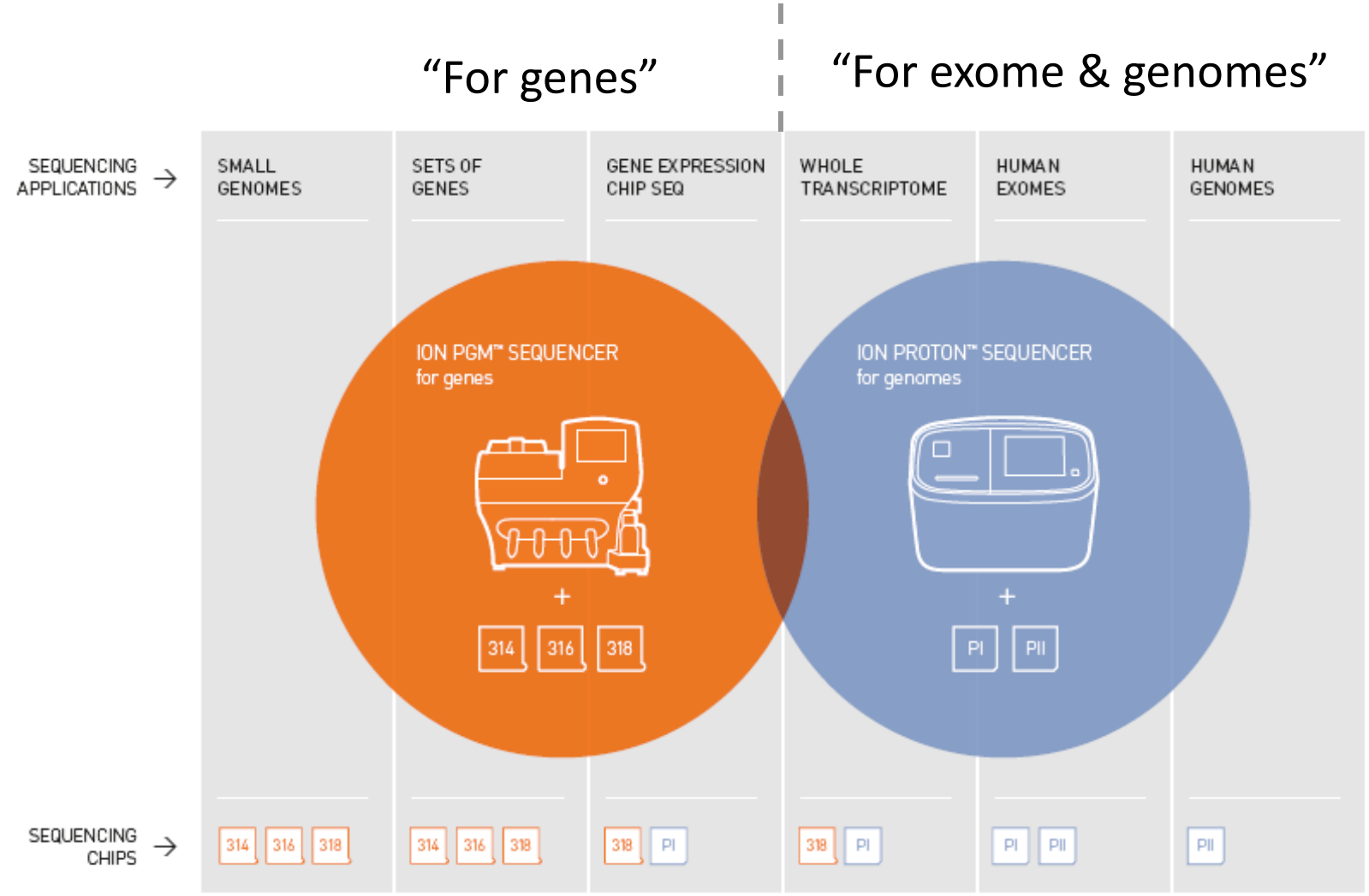
Gene to Genome by Next generation sequencer



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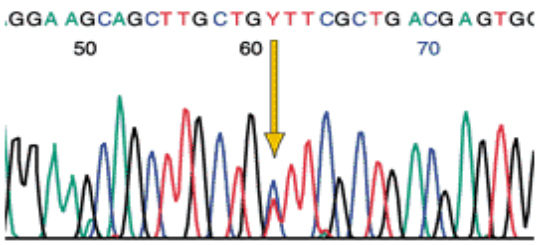
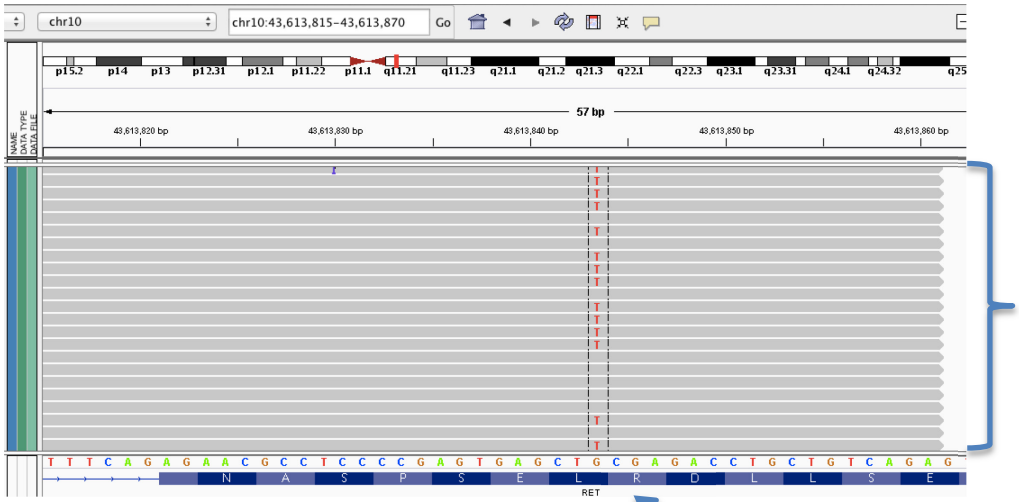
NGS potential as a diagnostic tool:



Limitation:
15~20 % mutation



Ref	Variant	Var Freq	Coverage	Ref Cov	Var Cov
G	T	59.66	24152	9739	14409

<u>On sequence variant detection</u>

NGS can get both of qualitative and quantitative data in same time!
AND low mutation can detect than Sanger sequence!!

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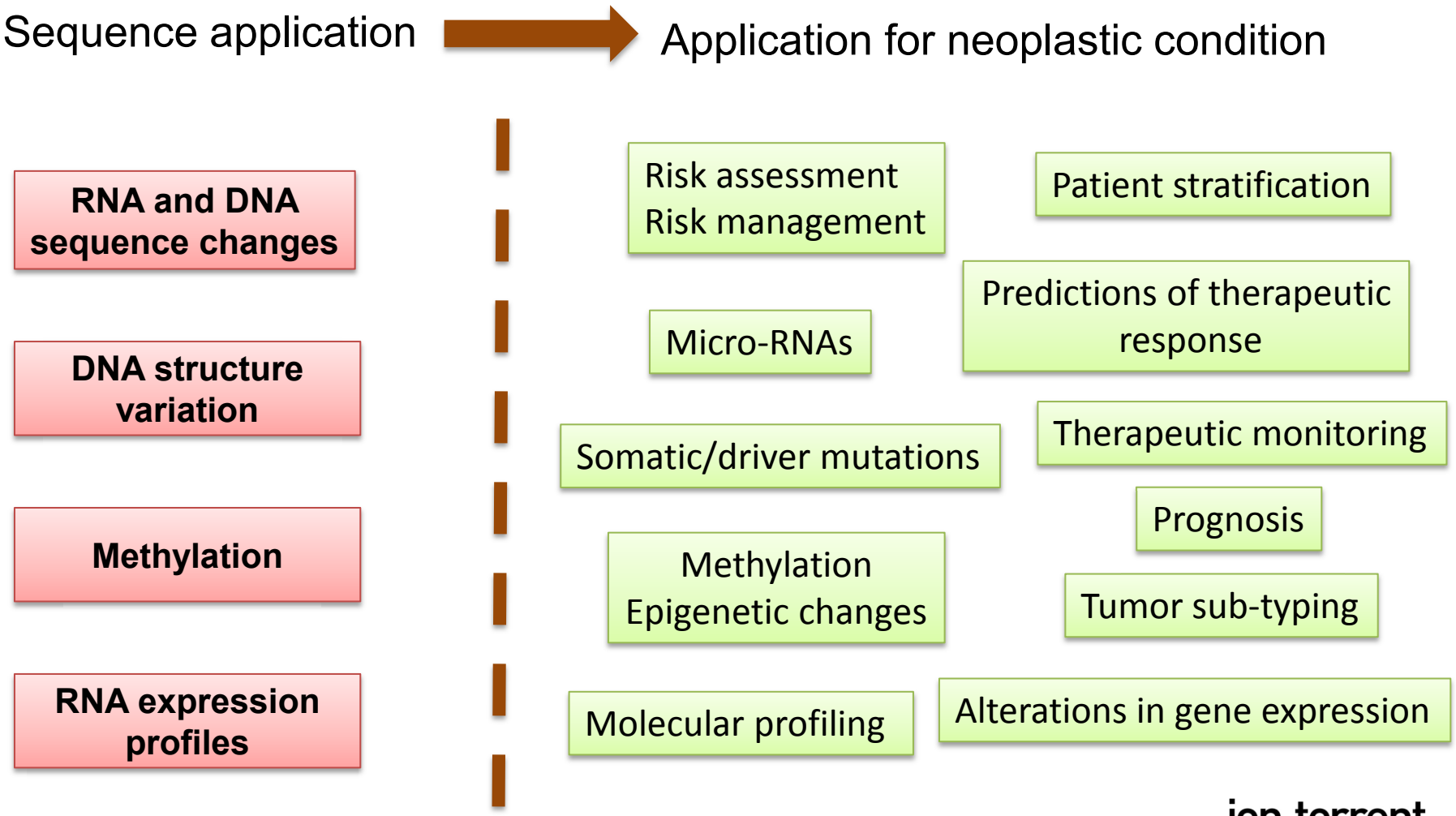
Examples for NGS Application

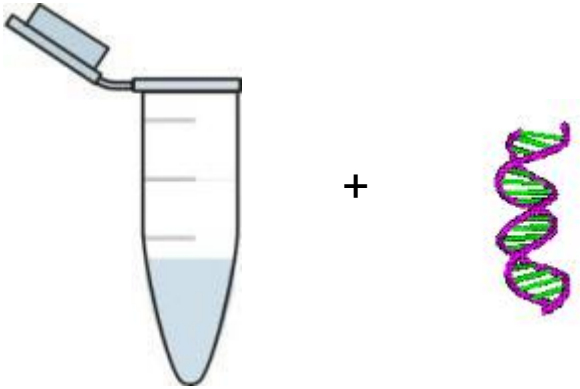


Ion AmpliSeq™ Technology: As Simple As PCR

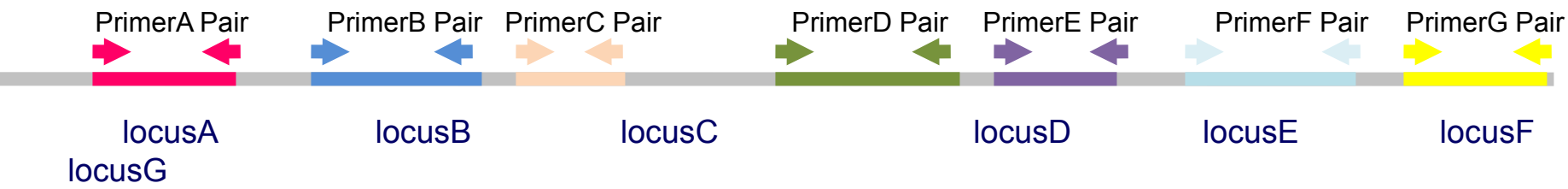
Single-tube, ultra-high multiplex for Targeted Resequencing

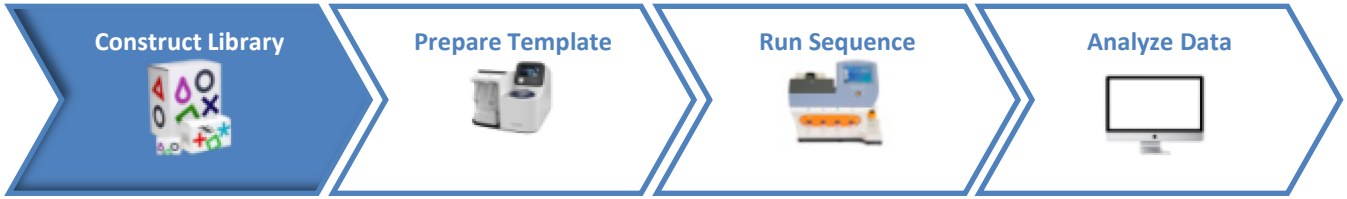
Up to 3072 primer pairs

10ng DNA



Multiplex PCR: Maximum 3072 amplion from 1 tube reaction





3.5 hours

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AmpliSeq™ panels

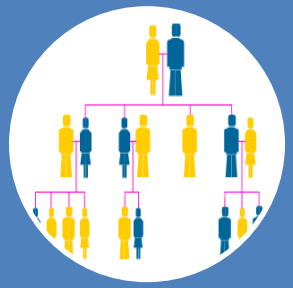
Cancer Primer Pool

46 genes

739 mutations

Comprehensive Cancer Panel

409 genes



Inherited Disease Panel

328 genes

>700 diseases

CUSTOM made

Ion AmpliSeq™ Designer

www.ampliseq.com

24 to 3,072 amplicons/tube

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Example for AmpliSeq Custom panel

Knight cancer institute

KNIGHT CANCER INSTITUTE
Oregon Health & Science University

KNIGHT DIAGNOSTIC LABORATORIES

Pioneering Personalized Diagnostics

About Us | Genetic Disorders | Infectious Diseases | Oncology | Research & Development | Client Services

Search All Tests | Search Tests | Alphabetical | by Department

Home / Search Results

Test Search Results - *ampliseq*

GeneTrails GIST Gene Panel

Test Code: 4500 • **Department:** Solid Tumors

Test Synonyms: GIST Cancer Gene Panel • **AmpliSeq** GIST Panel • Ion Torrent GISH Panel • AKT1 • AKT2 • AKT3 • ATM • BRAF • CDKN2A • HRAS • KIT • KRAS • MAP2K1 • NF1 • NRAS • PDGFRA • PIK3CA • PTEN • PTPN11 • SDHA • SDHAF1 • SDHAF2 • SDHB • SDHC • SDHD • TP53 • RAS • RAF

View Test >

GeneTrails NSCLC Panel

Test Code: 5120 (Sequencing) 7600 (FISH Panel) • **Department:** Solid Tumors

Test Synonyms: Lung Cancer Panel • **AmpliSeq** NSCLC Panel • Ion Torrent Lung Panel • AKT1 • ALK • BRAF • CDKN2A • DDR2 • EGFR • ERBB2 • HRAS • JAK2 • KDR • KRAS • MAP2K1 • NOTCH1 • NRAS • NTRK1 • NTRK2 • NTRK3 • PIK3CA • PIK3R1 • PIK3R2 • PTEN • PTPRD • TP53 • ROS1 • RET • MET • RAS • RAF

View Test >

Contact Us: 855-535-1522

http://www.knightdxlabs.com/home/search-results?q=ampliseq

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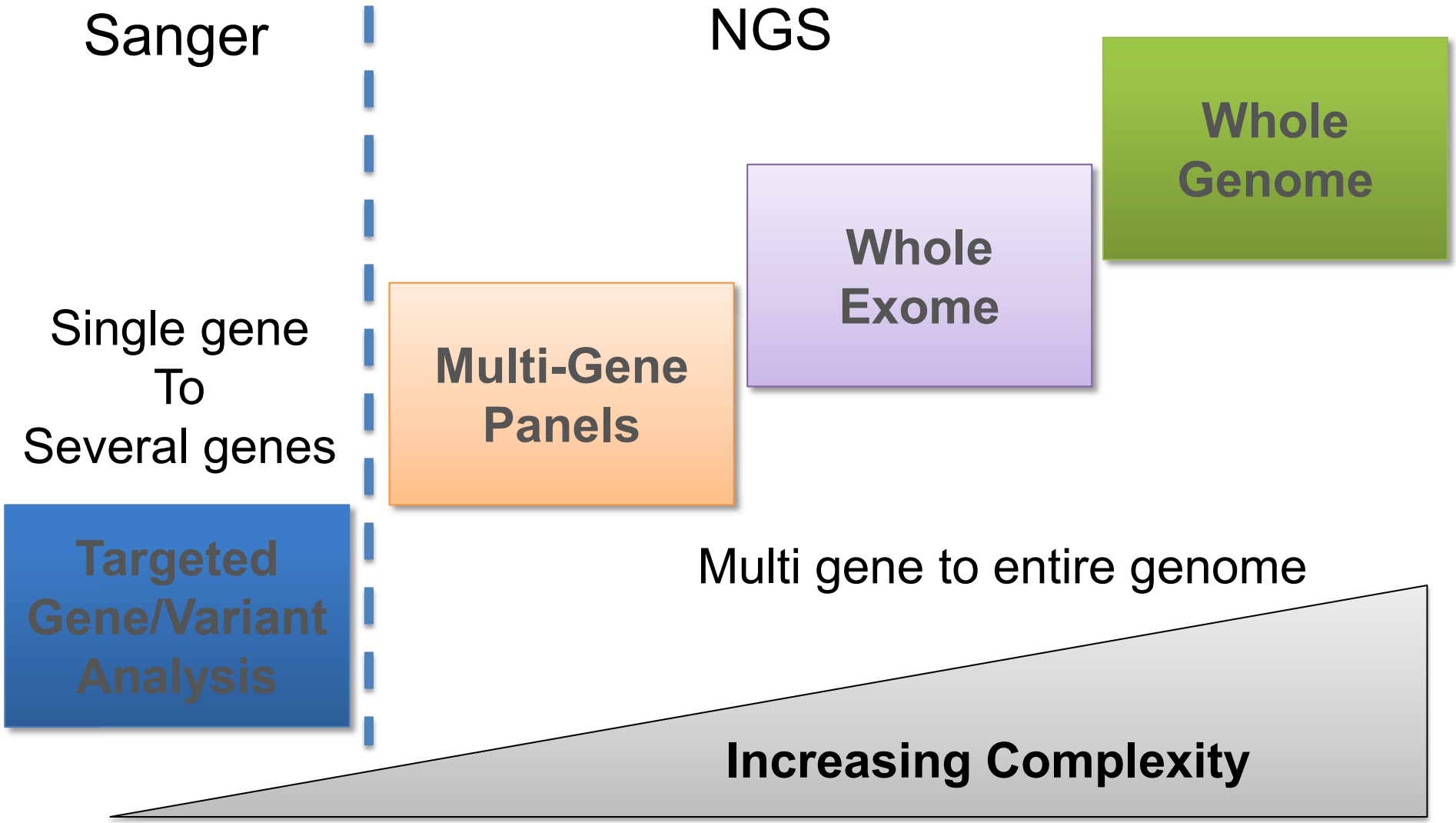
Diagnostic Spectrum



Clinical implementation of NGS

- Division of Laboratory Science and Standards (CDC)
- Genetic Testing Reference Material Coordination Program (Get-RM) (CDC)

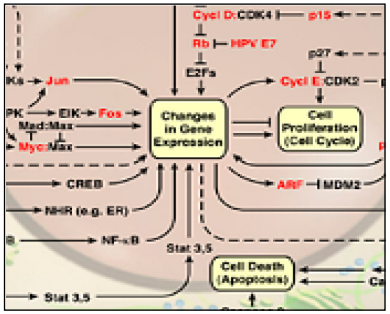
  http://www.cdc.gov/dls/genetics/rmmaterials/default.aspx
- Clinical Laboratory Standards Institute (CLSI)
- American College of Medical Genetics (ACMG)
- College of American Pathologists (CAP)
- Association For Molecular Pathology (AMP)

Clinical decision support systems

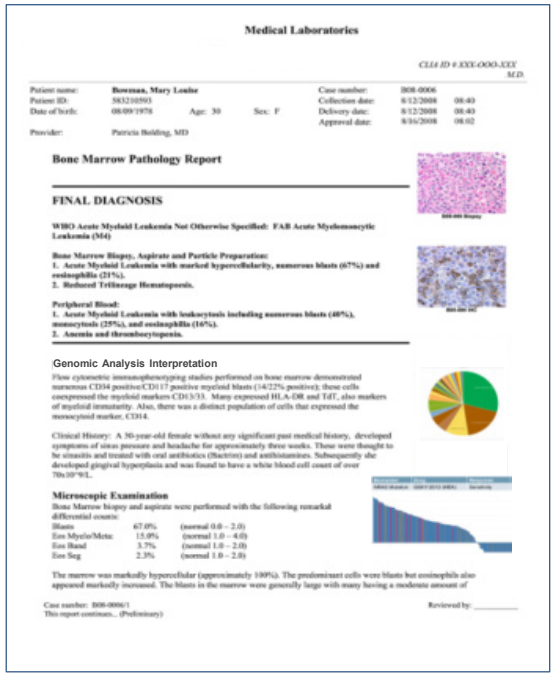
Example
- Expert interpretation and guidance
- Proficiency testing and alternative assessment are challenging

Ion Torrent PGM*

From sequences…



… to biological understanding…



Medical Laboratories

Bone Marrow Pathology Report

FINAL DIAGNOSIS

Genomic Analysis Interpretation

Microscopic Examination

…to actionable information

Ion Semiconductor Sequencing

Rapid, Benchtop Sequencing for All

PGM™ for genes.
Proton™ for genomes.
Sequencing for all.

Ion PGM™ Sequencer

Ion Proton™ Sequencer

For more information, visit http://lifetechnologies.com/proton

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