

# イルミナシーケンサーの原理

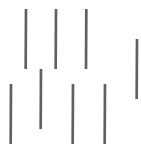


イルミナ株式会社  
営業部  
川島 佑介



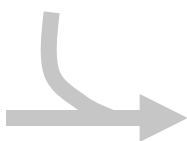
# サンプル調製とDNA分子の増幅(自動化)

ゲノムDNAおよびターゲットDNA  
を抽出後、断片化



DNA断片  
0.1 ~ 1µg

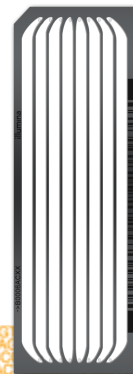
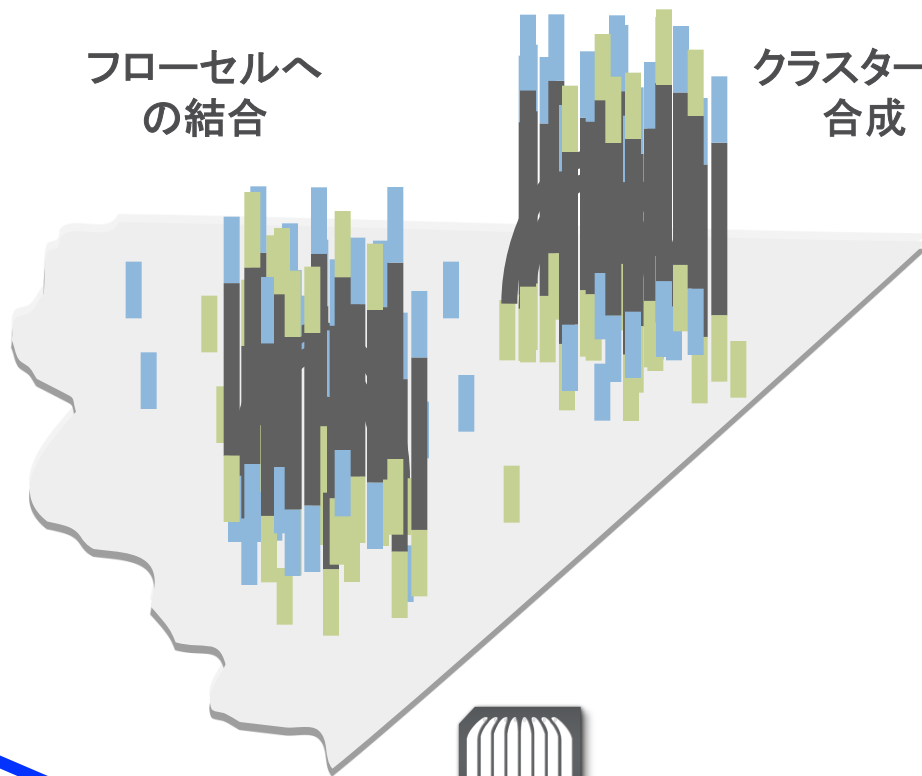
2種類の  
アダプター付加



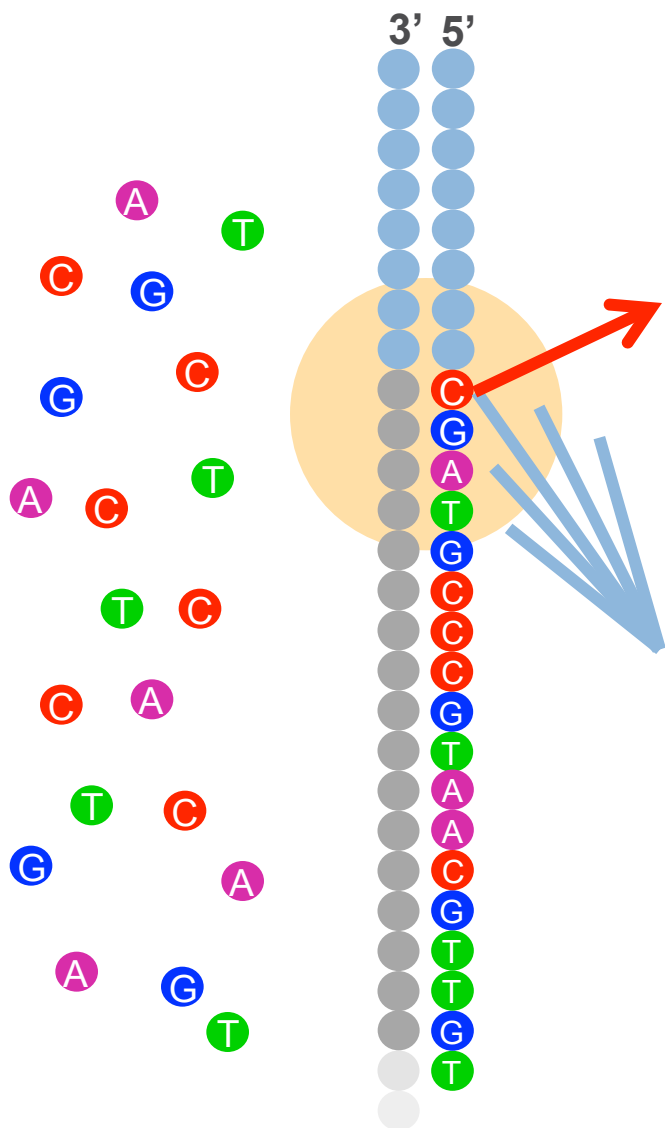
PCR後、  
濃度調整をして  
フローセルへ

フローセルへ  
の結合

クラスターの  
合成



# 可逆的ターミネーター法を用いた1塩基伸長反応



## Cycle 1

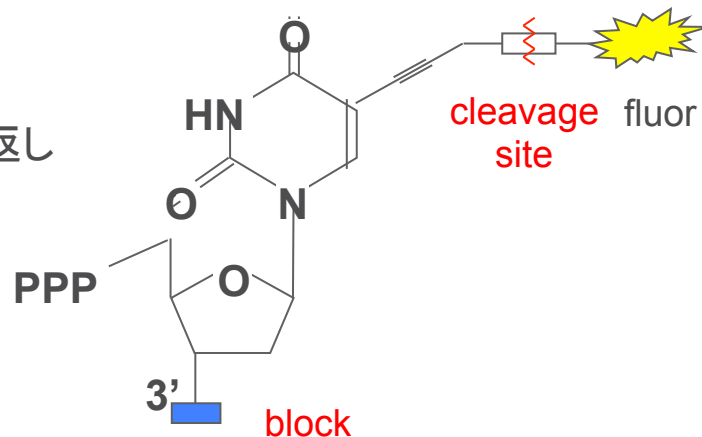
- ▶ シーケンス試薬の添加
- ▶ 1塩基伸長反応
- ▶ 未反応試薬の除去
- ▶ 蛍光シグナルの取り込み
- ▶ 保護基と蛍光の除去

## Cycle 2

- 上記反応の繰り返し

## Cycle 3, 4, 5.....

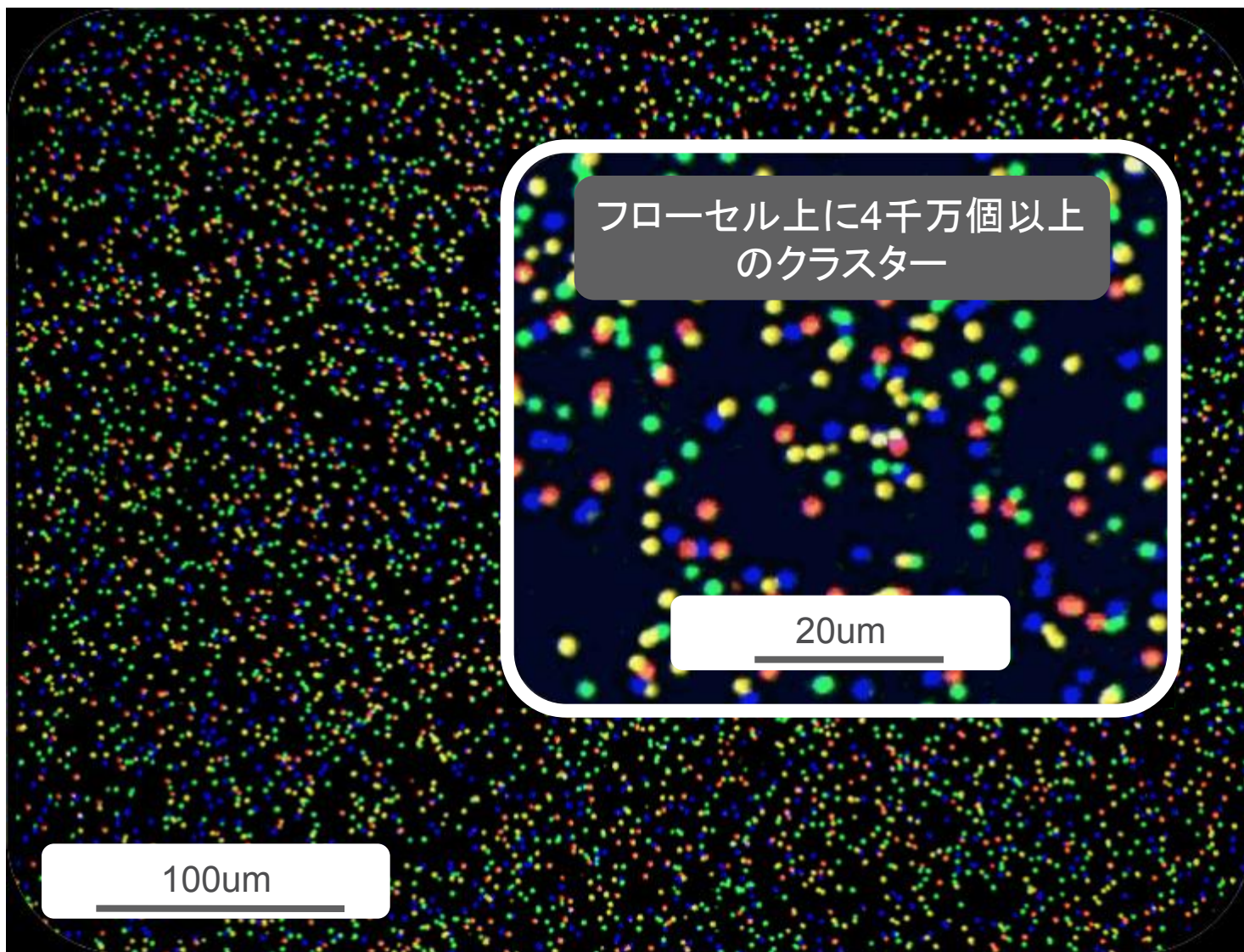
- 上記反応の繰り返し



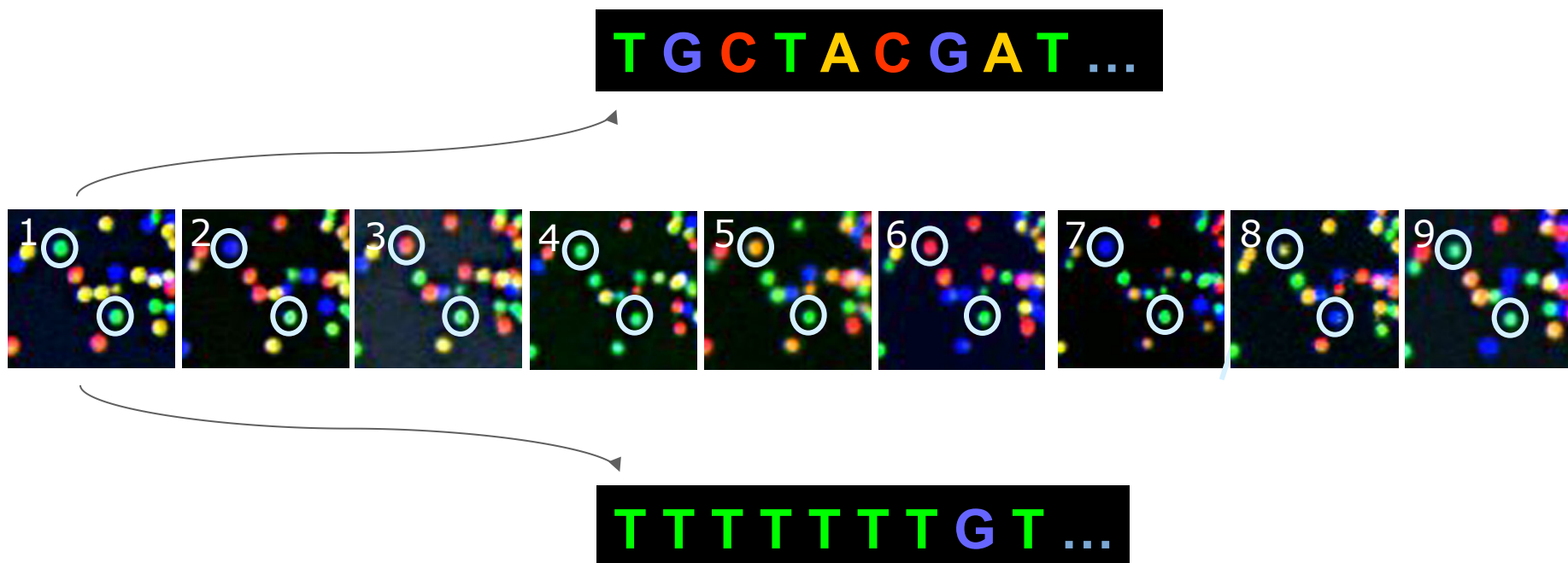
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 GGTAAACAGCITTCGTGTTAACCTTAAAGATTACITTCATCCACTGATTCAAGTACCGTAAAGTATAATTAACCGTACCATTAAAGAGCTACCGTGCACAGGACGAAAAAAGATGATA  
 CTTTCGACAGTAAACAGCITTCGTGTTAACCTTAAAGATTACITTCATCCACTGATTCAAGTACCGTAAAGTATAATTAACCGTACCATTAAAGAGCTACCGTGCACAGGACGAAAAAAGATGATA  
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 CTTTCGACAGTAAACAGCITTCGTGTTAACCTTAAAGATTACITTCATCCACTGATTCAAGTACCGTAAAGTATAATTAACCGTACCATTAAAGAGCTACCGTGCACAGGACGAAAAAAGATGATA



# 画像イメージの取り込み



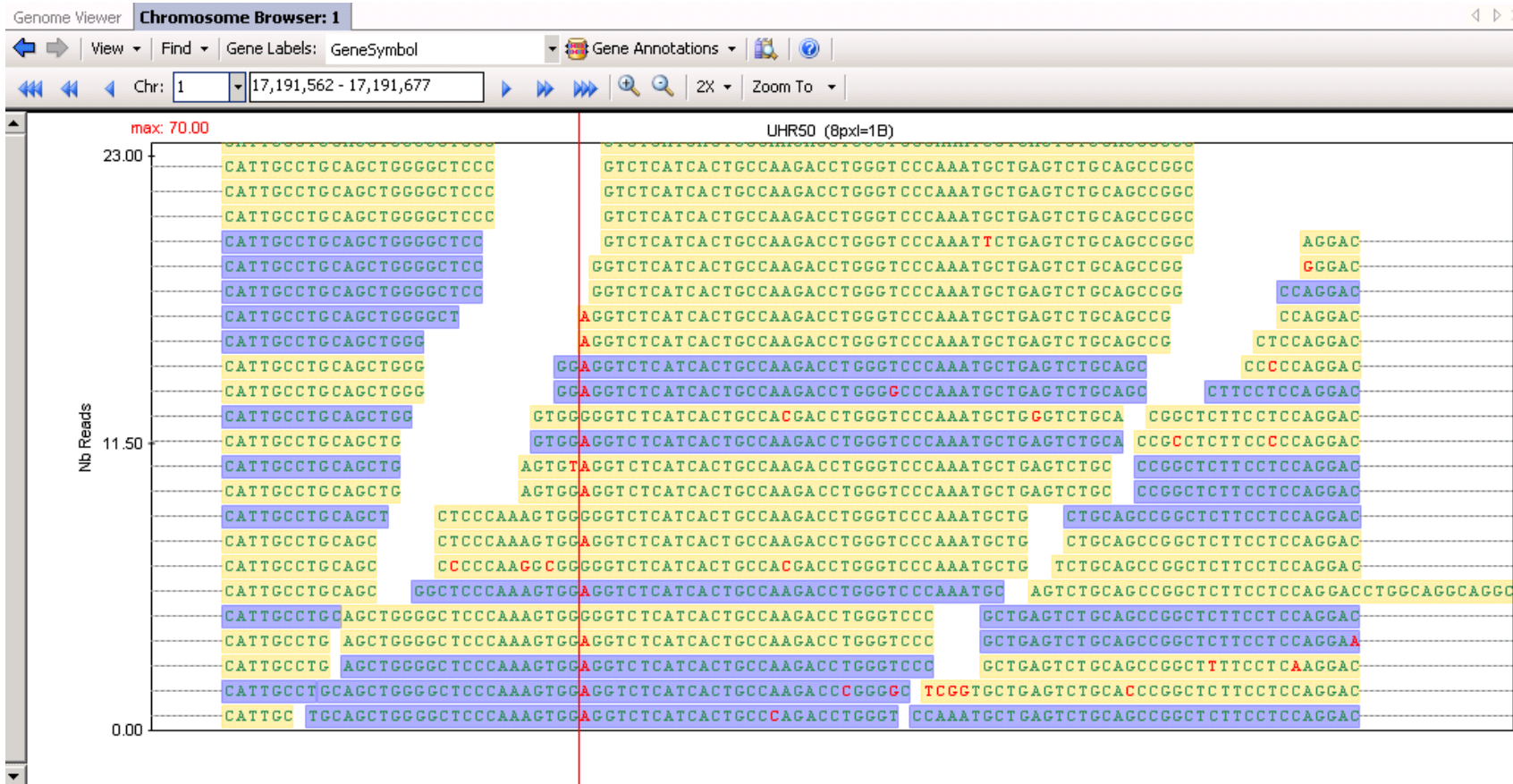
# 画像蛍光シグナルから塩基への変換



1塩基伸長反応ごとに蛍光イメージをとる  
→ 蛍光の色から各塩基を決定する



# リファレンス(既知のゲノム配列)へのマッピングと



Base Position	17,191,572	17,191,582	17,191,592	17,191,602	17,191,612	17,191,622	17,191,632	17,191,642	17,191,652	17,191,662	17,191,672
Cytogenetic Band	p36.13										
Sequence (+)	GCTCACCATGCGCTGCAGCTGGGGCTCCCAAAGTGGGGTCTCATCACTGCCAAGACCTGGGTCCTCCAAATGCTGAGTCTGCAGCCGGCTCTTCCTCCAGGACCTGGCAGGCAGGC										
brain50				A	G						C

# シングルリード法・ペアエンド法

## シングルエンド

- ▶ DNA断片の片側を100bp読み取り



## ペアエンド

- ▶ DNA断片の両端を100bp読み取り
- ▶ DNA断片の長さにより、さらに2つに区分
  - 200-500bp: ペアエンド法(イルミナシステムのみ)
  - 2-5kb: メイトペア法



ペアエンド法  
200-400bp

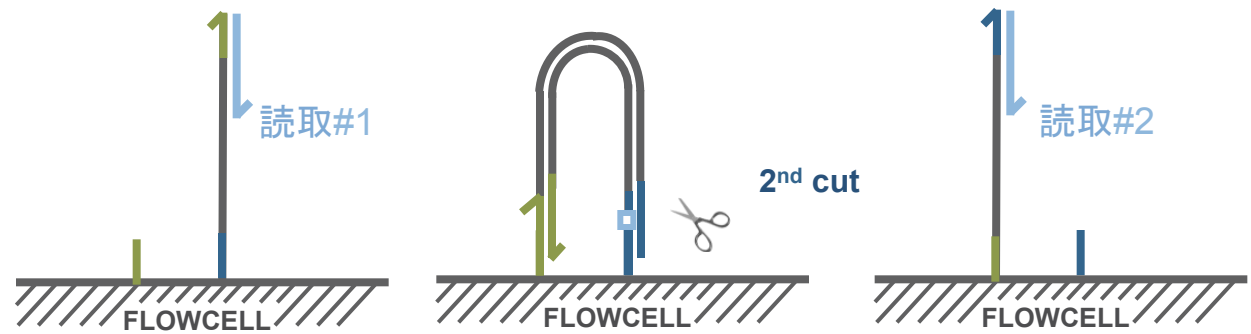


メイトペア法  
2-5kb

- ・両端を読むことで、データ量が2倍に
- ・アライメント時の効率アップ

# ペアエンド(両端読み)とは？

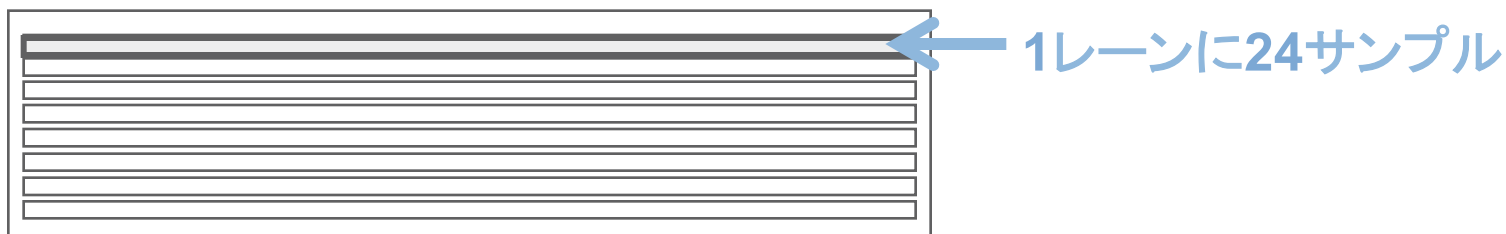
- ▶ 同一のDNA断片の両端を読む
- ▶ 片側を読んだ後、DNA断片をひっくり返してもう一方の端を読む



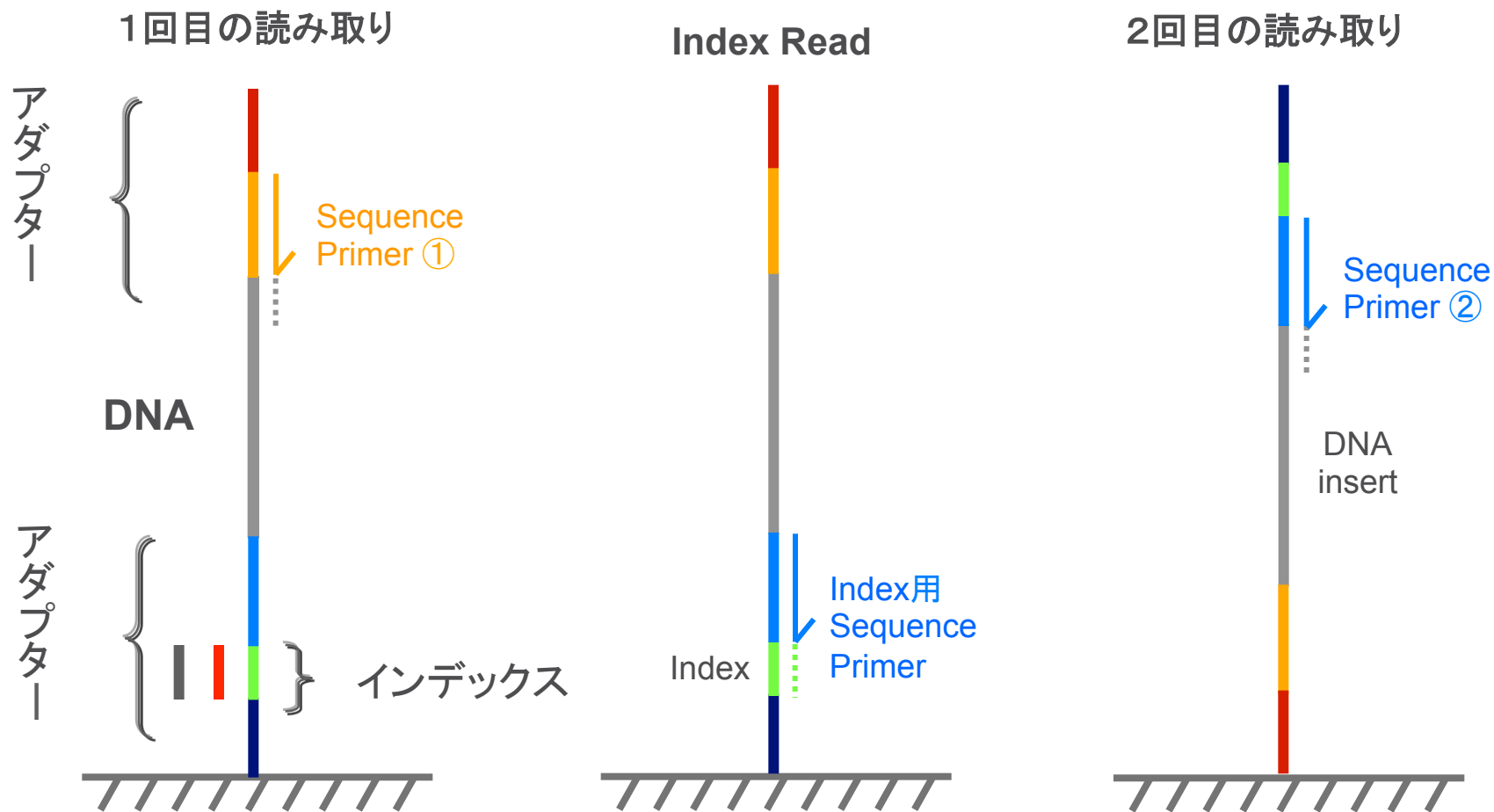


# マルチプレックス

- ▶ 多サンプルをシーケンスしたい
- ▶ 1レーンに複数サンプルを混ぜてシーケンス
- ▶ 現在1レーンあたり最大24サンプル可能、1枚のフローセルでは最大168サンプル (1レーンはコントロールに使用)



# マルチプレックスの反応経路





# 最近の出来事

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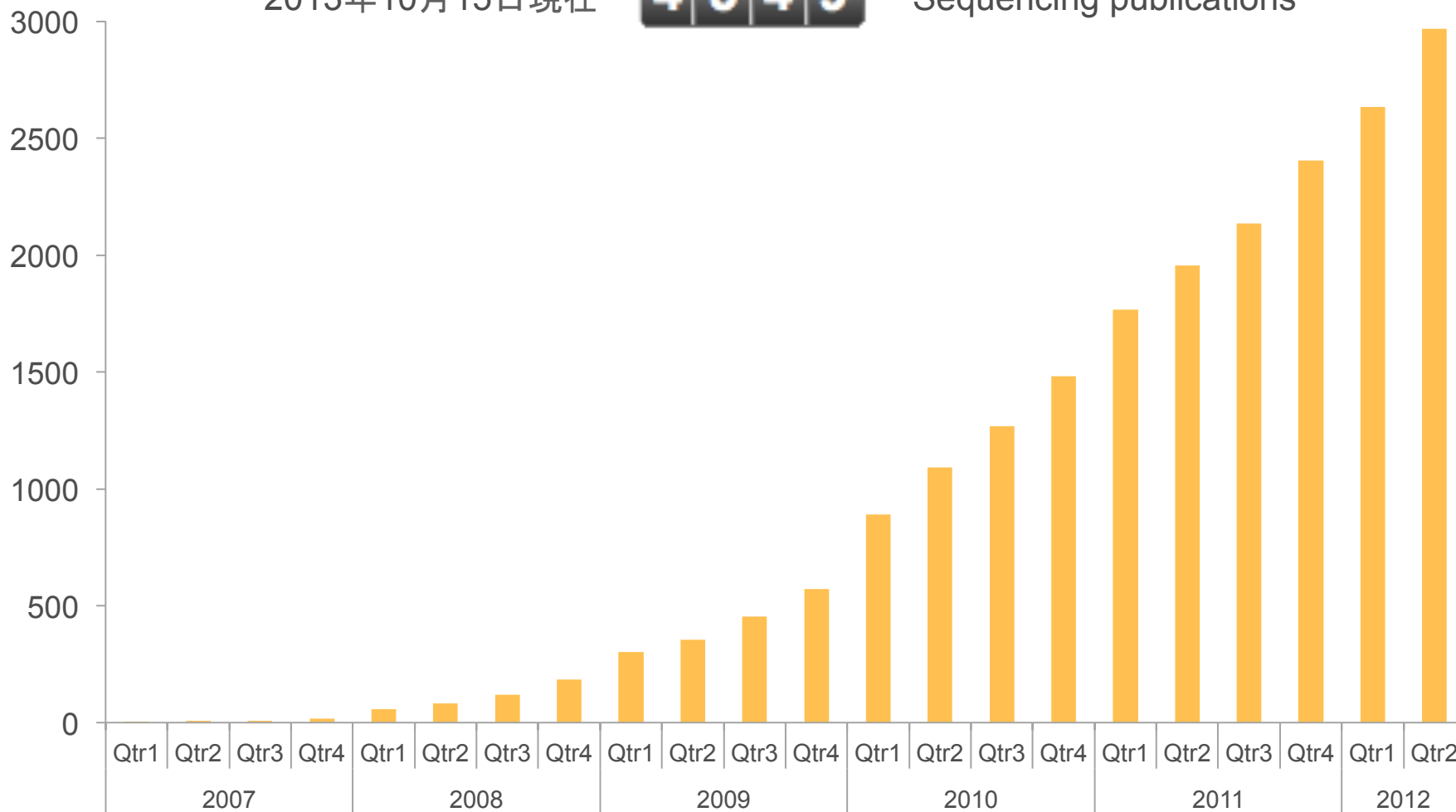
# SBSケミストリーの実績

## イルミナ次世代シーケンサー利用文献数

2013年10月15日現在

4549

Sequencing publications



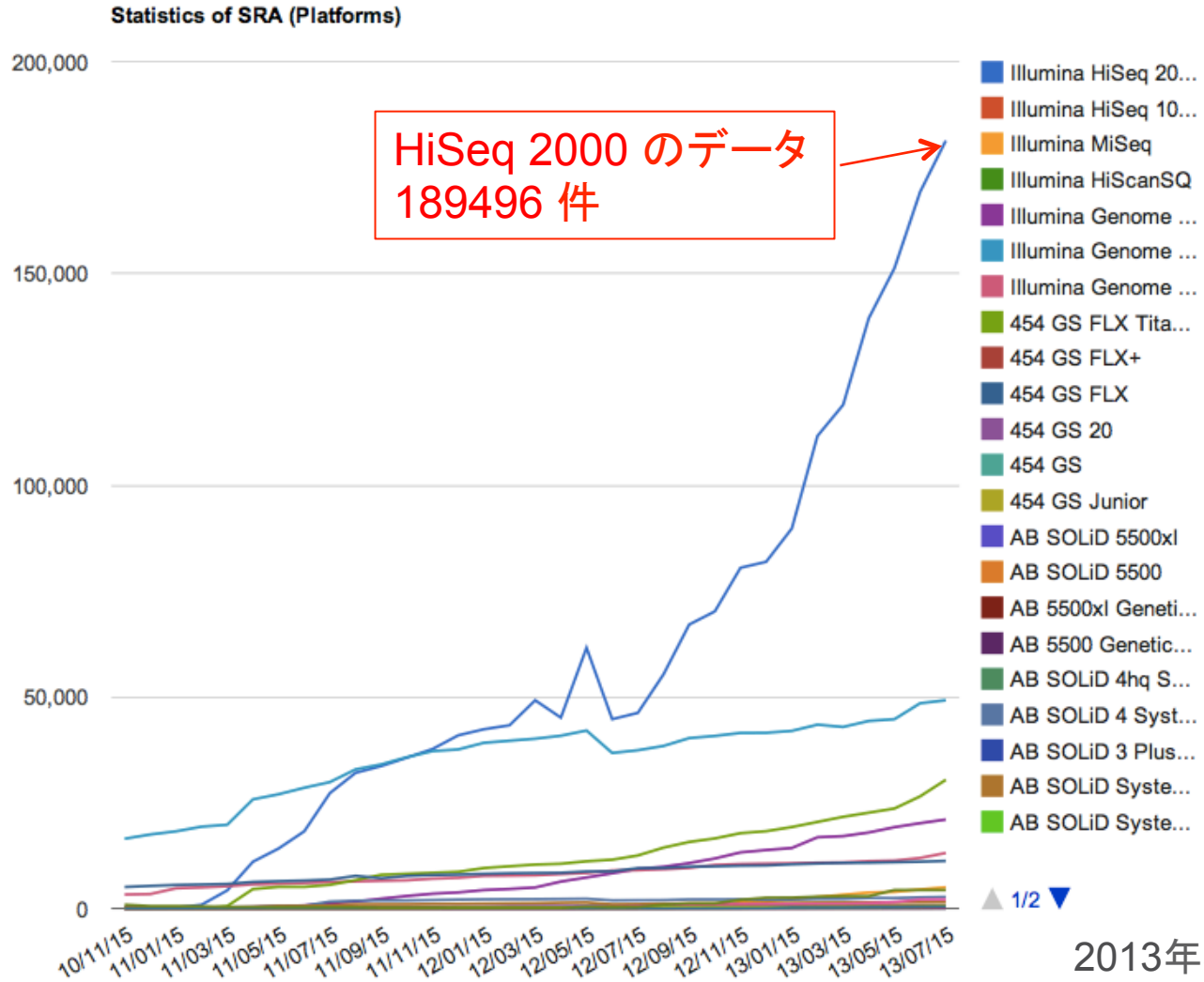
<http://www.illumina.com/publications/>



# 国際データベースに登録されている次世代シーケンサーのデータ

<http://sra.dbcls.jp/>

Sum of "Short Read Archive" entry on NCBI, EBI and DDBJ



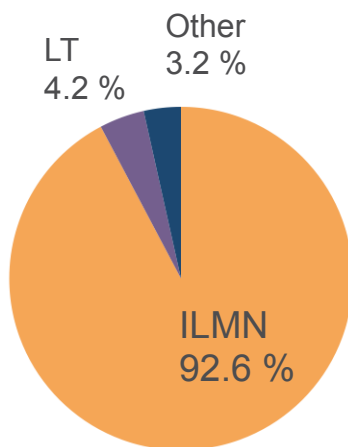
# 国際データベースに登録されているヒト解析データ

90%以上がイルミナプラットフォームからのデータ

- データ取得元 NCBI Sroht Read Archive <http://www.ncbi.nlm.nih.gov/sra>
- NCBI, EBI, DDBJ に登録された次世代シーケンサーのデータを集計したサイト

## 実験 (Run) 別に集計\*

- 登録数 100,837 件
- ILMN 93,411 件 (92.6%)



\* データ取得元 <http://www.ncbi.nlm.nih.gov/sra> (2013年8月23日取得)

ILMN検索条件 : txid9606[Organism:exp] と HiSeq, Genome Analyzer, MiSeq のいずれか

LT 検索条件 : txid9606[Organism:exp] と SOLiD, 5500, PGM, Proton のいずれか



# HiSeq関連の出来事関連の出来事

# HiSeq 2500を発表： 1台のシステムで2つのモード搭載



マウスでクリック選択

標準モード  
11日間で600Gb

Fast Runモード  
1ゲノムを約1日で解析



# HiSeq2500を発売:

## 1台のシステムで2つのモード搭載

- ▶ 2つのコンフィグを設定
  - High Outputモード
  - Fast Runモード
  
- ▶ Fast Runモード
  - 1日で100bp x2ランを終了
  - クラスター形成も装置内で自動化  
(※標準モードではcBotが必要)
  - MiSeqケミストリーによる迅速サイクル
  - 150bp x2も可能に  
(※Fast Runモードのみ適応)
  - 新フローセルを使用

HiSeq 2500 システムパフォーマンス*			
	標準モード	Fast Runモード	
リード長	2 x 100	2 x 100	2 x 150
データ量 (Gb)	~600	~115-130	~170-195
ラン時間	~11日	~27時間	~39時間
% > Q30	> 80	> 80	> 75
パスフィルターリード	> 90%	> 90%	> 90%
フローセル数	2	2	2
レーン数/フローセル	8	2	2
クラスター形成	cBot	装置内 自動化	装置内 自動化

\*Anticipated performance at launch for new HiSeq 2500 systems. Official specifications will be set at launch.

# 新生児集中治療患者の迅速なシーケンスとアノテーション

5ヶ月の乳児 -  
発達遅延、発作など

サンプル調製後  
HiSeq 2500でシーケ  
ンス解析

銅代謝に関連する新  
規の変異を同定

Menkes 症の  
診断を確認



gene machines

Science  
Translational  
Medicine

d cause of

RESEARCH ARTICLE

DIAGNOSTICS

## Rapid Whole-Genome Sequencing for Genetic Disease Diagnosis in Neonatal Intensive Care Units

Carol Jean Saunders,<sup>1,2,3,4,5\*</sup> Neil Andrew Miller,<sup>1,2,4\*</sup> Sarah Elizabeth Soden,<sup>1,2,4†</sup>  
Darrell Lee Dinwiddie,<sup>1,2,3,4,5†</sup> Aaron Noll,<sup>1</sup> Noor Abu Alnadi,<sup>4</sup> Nevene Andraws,<sup>3</sup>  
Melanie LeAnn Patterson,<sup>1,3</sup> Lisa Ann Krivohlavek,<sup>1,3</sup> Joel Fellis,<sup>6</sup> Sean Humphray,<sup>6</sup> Peter Saffrey,<sup>6</sup>  
Zoya Kingsbury,<sup>6</sup> Jacqueline Claire Weir,<sup>6</sup> Jason Betley,<sup>6</sup> Russell James Grocock,<sup>6</sup>  
Elliott Margulies,<sup>6</sup> Emily Gwendolyn Farrow,<sup>1</sup> Michael Artman,<sup>2,4</sup> Nicole Pauline Safina,<sup>1,4</sup>  
Joshua Erin Petrikin,<sup>2,3</sup> Kevin Peter Hall,<sup>6</sup> Stephen Francis Kingsmore<sup>1,2,3,4,5†</sup>

# 新生児集中治療患者の迅速なシーケンスとアノテーション

- ▶ 発達遅延、低血圧症、発作などを示す5ヶ月の新生児
- ▶ Rapidランによる全ゲノムシーケンス
  - DNAから変異検出まで50時間以内

Unrelated

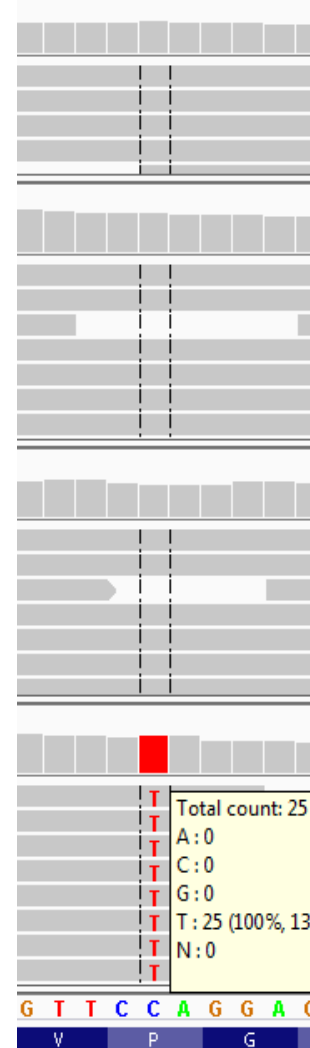
Unrelated

Unrelated

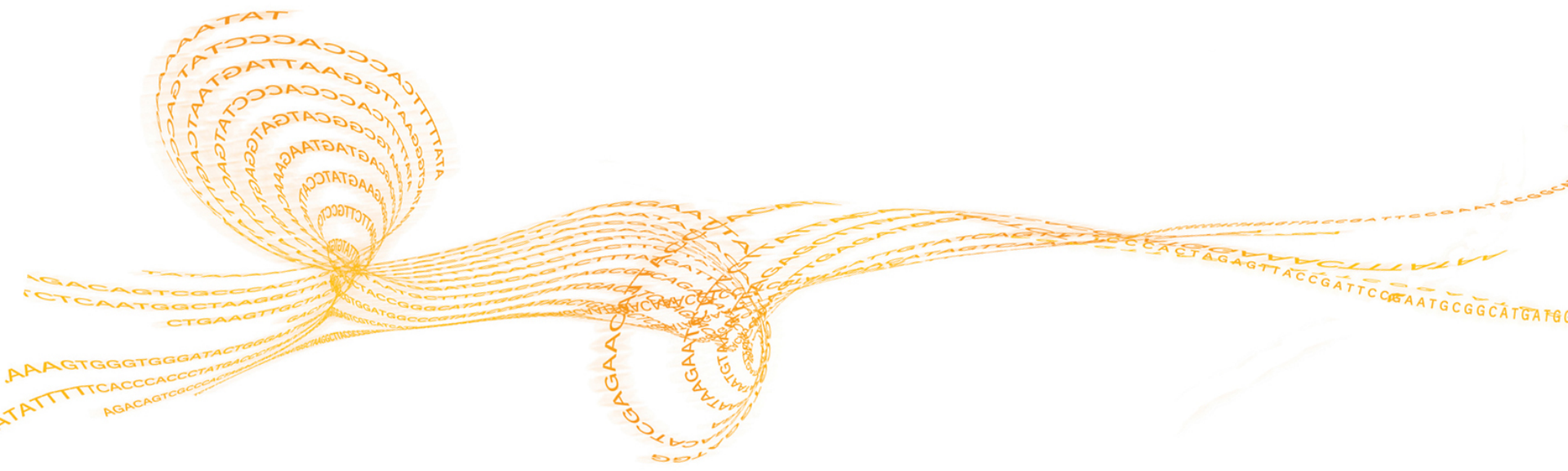
Menkes

変異コール数:	3.54M
コーディング領域の変異:	16,180
影響があると思われる予測変異:	449
稀な変異:	24
進化上保存された変異:	5
表現型で変異されている遺伝子	1

- ▶ 新しいヘミザイガスの変異を発見；銅代謝を妨げる遺伝子
- ▶ メンケス病と診断



Saunders, et al. Rapid Whole-Genome Sequencing for Genetic Disease Diagnosis in Neonatal Intensive Care Units, *Sci. Trans. Med.*, 2012



# 医療への進展



# BlueGnome社の買収 (2012年)

- ▶ ウェルカムトラストとケンブリッジ大学からの資金により、2002年にケンブリッジ大学からスピンアウトして創立した会社
- ▶ 分子細胞遺伝学分野のリーダー
  - 着床前遺伝スクリーニング (PGS: Preimplantation Genetic Screening) で #1の市場地位を確立
  - その他の細胞遺伝学市場 (出生前、出生後、癌) では #3
- ▶ 200を超える顧客数
- ▶ 2004年から利益
- ▶ マイクロアレイのソリューションを提供
  - マイクロアレイ、試薬、FISHおよびソフトウェア (BlueFuse)
- ▶ 57名の社員
  - 細胞遺伝学、統計的シグナル処理、ソフトウェア、分子生物学
  - 営業チームは主に体外受精センター (IVF: in vitro Fertilization) をターゲットとして活動
- ▶ 本社はイギリスのフルボーン (ケンブリッジの近く)
  - その他、シンガポール、アメリカバージニア州にも社員あり



# 着床前遺伝スクリーニングで正常な胚を選択

- ▶ BlueGnomeの 24sure というテクノロジーは、体外受精プロセスの中で、移植に最適な胚を選ぶための迅速なスクリーニング
- ▶ 最初の文献 24sure RCT (Yang et al, Journal of Molecular Cytogenetics 2012)
  - 24sure を用いた場合、20週における妊娠率は65%増加
- ▶ 着床前遺伝スクリーニングは、今後の体外受精において標準化される期待がある

**MOLECULAR CYTOGENETICS** Official Impact Factor 2.41

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**methodology**  
**Selection of single blastocysts for fresh transfer via standard morphological assessment alone and with array CGH for good prognosis IVF patients: results from a randomized pilot study**  
 Zhilong Yang, Jian Liu, Gary S Collins, Shaia A Salera, Xiaohong Liu, Sarah S Lytle, Alison C Peck, and Rifkat O Sultan

**ESHRE CLINICAL TRIAL**  
**Green light for polar body microarray CGH trial**

Following the successful completion of its proof of principle study of polar body biopsy and microarray analysis, ESHRE is now ready for the next step: a randomised controlled trial (RCT). This trial has two primary aims among women with advanced maternal age: first, to estimate the likelihood of having no euploid embryos in future ART cycles; and second, to improve live birth rates. Among women aged 36 to 40 years planning three IVF cycles, does microarray analysis of all chromosomes in the first and second polar body compared with no intervention increase the likelihood of a live birth within one year? And in women with no fresh or frozen embryos. Second outcomes include live birth rate, specific good prognosis group, pathway outcome (implantation rates, ongoing pregnancy rates, abortion rates), genetic outcome (proportion of aneuploidy, affect chromosomes), diagnostic efficacy (embryo outcomes, and adverse events).

The total study will involve 6 couples, allowing for possible 3 with 300 in each arm. The trial will begin soon and the Cambridge company BlueGnome will again be our partner

Human Reproduction, Feb 28, 2011, pp. 1-12, 2011  
 Advance Access publication on 20K 1000K doi:10.1093/hrop/k90

human reproduction ORIGINAL ARTICLE Rep

**PGD for reciprocal translocations using genomic hybridisation**

F. Fiorentino<sup>1</sup>\*, L. Spizzicci<sup>1</sup>, L. Ruzza<sup>1</sup>, F.M. Ubaldi<sup>1</sup>, E. ...

<sup>1</sup>GNOME—Molecular Genetics Laboratory, Via P. Agazzi, 1, 40138 Bologna, Italy; \*Correspondence address: E-mail: fiorentino@gnomegenetics.it

Submitted on December 17, 2010; accepted on February 17, 2011; accepted on March 1, 2011

**BACKGROUND:** Fluorescence in situ hybridisation (FISH) is the most widely used method for detecting unbalanced chromosome rearrangements in preimplantation embryos but it is known to have several technical limitations. We describe the clinical application of a molecular-based assay, array comparative genomic hybridisation (array-CGH), to simultaneously screen for unbalanced translocation, deletions and aneuploidy of all 24 chromosomes.

**METHODS:** Cell biopsy was carried out on cleavage-stage embryos (Day 3). Single cells were first tested and DNA amplified by whole-genome amplification (WGA). WGA products were then processed by array-CGH using 24 Sure-4 arrays, BlueGnome. Balanced/normal euploid embryos were then selected for transfer on Day 5 of the same cycle.

**RESULTS:** Twenty-eight consecutive cycles of preimplantation genetic diagnosis were carried out for 24 couples carrying 18 different balanced translocations. Overall, 187/200 (93.5%) embryos were successfully diagnosed. Embryos suitable for transfer were identified in 17 cycles (60.7%), with transfer of 20 embryos (mean 1.1 ± 0.5). Twelve couples achieved a clinical pregnancy (70.6% per embryo transfer), with a total of 14 embryos implanted (80.6% per transferred embryo). These patients delivered three healthy babies, starting within the other pregnancies (two babies and seven abortions) are ongoing beyond 30 weeks of gestation.

16 NEWS

Professor Simon Fisher with baby Oliver, who was conceived using pioneering IVF treatment in 1980.

**genomic nosome analysis**

FA, B.S., Times Eyewitness, B.S.; \*Jays Award, FA, B.S.\*  
 Active Sciences, Oxford Business Park, ...

... of two array-CGH protocols (2).

... error rate compared with 0.3% and ... clinically. The aneuploidy rate for ... significantly increasing with maternal age. The chromosomes most ... 5, 21, 22, and 23. We report the first live births after array-CGH combined with ...

... proved to be highly robust (2.2% no results) and specific (1.5% error rate) when ... of single cells biopsied from cleavage-stage embryos. This comprehensive ... is the first to be validated by re-analyzing the same embryos with another ... some alternative techniques for comprehensive chromosome screening, uterine ... of parental DNA and thus advance planning and careful scheduling are ...

... (C) 2011 by American Society for Reproductive Medicine.

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# ダウン症の母体血を用いた生前前診断 (Sequenom社)

## Sequenom Preliminary Full-year 2012 Revenues Grow 59 Percent to \$89M

January 07, 2013



By a GenomeWeb staff reporter

NEW YORK (GenomeWeb News) – Sequenom on Sunday said that it expects its full-year 2012 revenues to grow 59 percent year-over-year to \$89 million.

Based on the company's preliminary financial results, diagnostic services grew more than five-fold during the year to about \$46 million, compared to \$8.3 million in 2011, while Genetic Analysis posted \$43 million in sales.

The number of prenatal and retinal diagnostic tests accessioned in 2012 totaled more than 92,000. Sequenom said that more than 60,000 MaterniT21 Plus non-invasive fetal aneuploidy tests were accessioned in 2012, and the annualized run rate of the tests exceeded 120,000 at the end of the year. That is up from a 90,000 annualized test volume run rate that Sequenom said the test had reached during its [third-quarter earnings conference call](#) in November.

aA Type size: + -

Email

Printer-friendly version

RSS Feed

<http://www.genomeweb.com/clinical-genomics/sequenom-preliminary-full-year-2012-revenues-grow-59-percent-89m>



# イルミナも同様な検査が可能になる技術を持つ会社を買収

## ILLUMINA TO ACQUIRE VERINATA FOR UP TO \$450M

January 07, 2013

いいね! 3 Tweet 1 +1 2 Share 31

By a GenomeWeb staff reporter

NEW YORK (GenomeWeb News) - Illumina today said that it signed a definitive agreement to acquire Verinata Health for \$350 million with additional potential milestone payments of up to \$100 million through 2015.

aA Type size: + -

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The deal provides Illumina with Verinata's Verifi non-invasive prenatal test for detecting trisomy 21, 18, and 13. The test analyzes cell-free fetal DNA using next-generation sequencing.

"Building on the recent acquisition of BlueGnome Ltd. and our expertise in next-generation sequencing, this announcement further establishes Illumina as a leader in reproductive health," Illumina President and CEO Jay Flatley said in a statement.

<http://www.genomeweb.com/clinical-genomics/illumina-acquire-verinata-450m>



# Verinata社の技術

“This agreement with Verinata demonstrates Illumina’s commitment to developing innovative diagnostic solutions and providing our partners with the most advanced technologies for improved patient care,” said Jay Flatley, President and CEO of Illumina. “Building on the recent acquisition of BlueGnome Ltd. and our expertise in next-generation sequencing, this announcement further establishes Illumina as a leader in reproductive health.”

Available through a physician, the verifi test analyzes cell-free fetal DNA naturally found in a pregnant woman’s blood to look for missing or extra copies of chromosomes (referred to as aneuploidies). Specifically, the test detects Down syndrome (trisomy 21 or T21), Edwards syndrome (trisomy 18 or T18) and Patau syndrome (trisomy 13 or T13). It is the first non-invasive prenatal test that offers the option to include evaluation of sex chromosome aneuploidies, such as Turner syndrome (Monosomy X), Triple X (XXX), Klinefelter syndrome (XXY) and Jacobs syndrome (XYY) – the most common fetal sex chromosome abnormalities.

[http://investor.illumina.com/phoenix.zhtml?  
c=121127&p=irol-  
newsArticle&ID=1771460&highlight=](http://investor.illumina.com/phoenix.zhtml?c=121127&p=irol-newsArticle&ID=1771460&highlight=)



# MiSeq関連の出来事

# オリンピックにおける MiSeq (Genome Webの記事より)

## オリンピック期間中、感染症が発生時に病原菌をMiSeqでシーケンス解析



### UK's HPA and University of Oxford Adopt MiSeq for Public Health Protocol During Olympics

August 08, 2012

#### UK's HPA and University of Oxford Adopt MiSeq for Public Health Protocol During Olympics

By Monica Heger

**Next-generation sequencing for public health surveillance could be put to the test these Olympic games. The UK's Health Protection Agency has implemented the technology as part of its public health protocol for monitoring outbreaks during the London Olympics.**

In collaboration with the University of Oxford, the HPA will use the university's Illumina MiSeq to sequence pathogens responsible for any disease outbreaks that may arise during the Olympics. The agency is also looking into acquiring its own NGS system for use in public health surveillance but has not yet decided on a specific technology.

# 米国FDAの選択

- ▶ 2012年9月18日、米国FDAとイルミナは契約を結び、MiSeqシーケンシングシステムと試薬を提供すると発表した
- ▶ FDAは、これらを用いて食品媒介病原菌検出のための基礎となるデータを収集する
- ▶ 契約は5年間で最大1700万ドルがイルミナに支払われる
- ▶ FDAは既にMiSeqシステムを使用しており、概念実証イニシアチブの一環として、全ゲノム解析の能力を増強してきた。この契約によりFDAは将来のアウトブレイクに備え、更にリソースを収集する計画だ
- ▶ 具体的には、様々な腸内病原菌の検出、分離、同定をFDA傘下のラボで行なう。
- ▶ 全米の農業物由来と農産物を取り巻く環境由来のサルモネラ菌や志賀毒素産生性大腸菌にMiSeqを用いて全ゲノム解析を行い、高品質のデータを収集、NCBIのデータベースに即時アップロードする。これによりNCBIデータベースの感染ルート追跡における価値が高まると期待される。



# FDAからの認可

## MiSeqDx™ FDA Submissions

In December, Illumina became the first company to submit an NGS system to the FDA with submissions for the **MiSeqDx platform** and **MiSeqDx Cystic Fibrosis system**. The MiSeqDx platform will enable customers to develop their own diagnostic assays, while the Cystic Fibrosis system will provide customers the ability to perform CF carrier screen testing as well as full CFTR gene sequencing.

## MiSeqDx Cystic Fibrosis System

One platform. One solution.

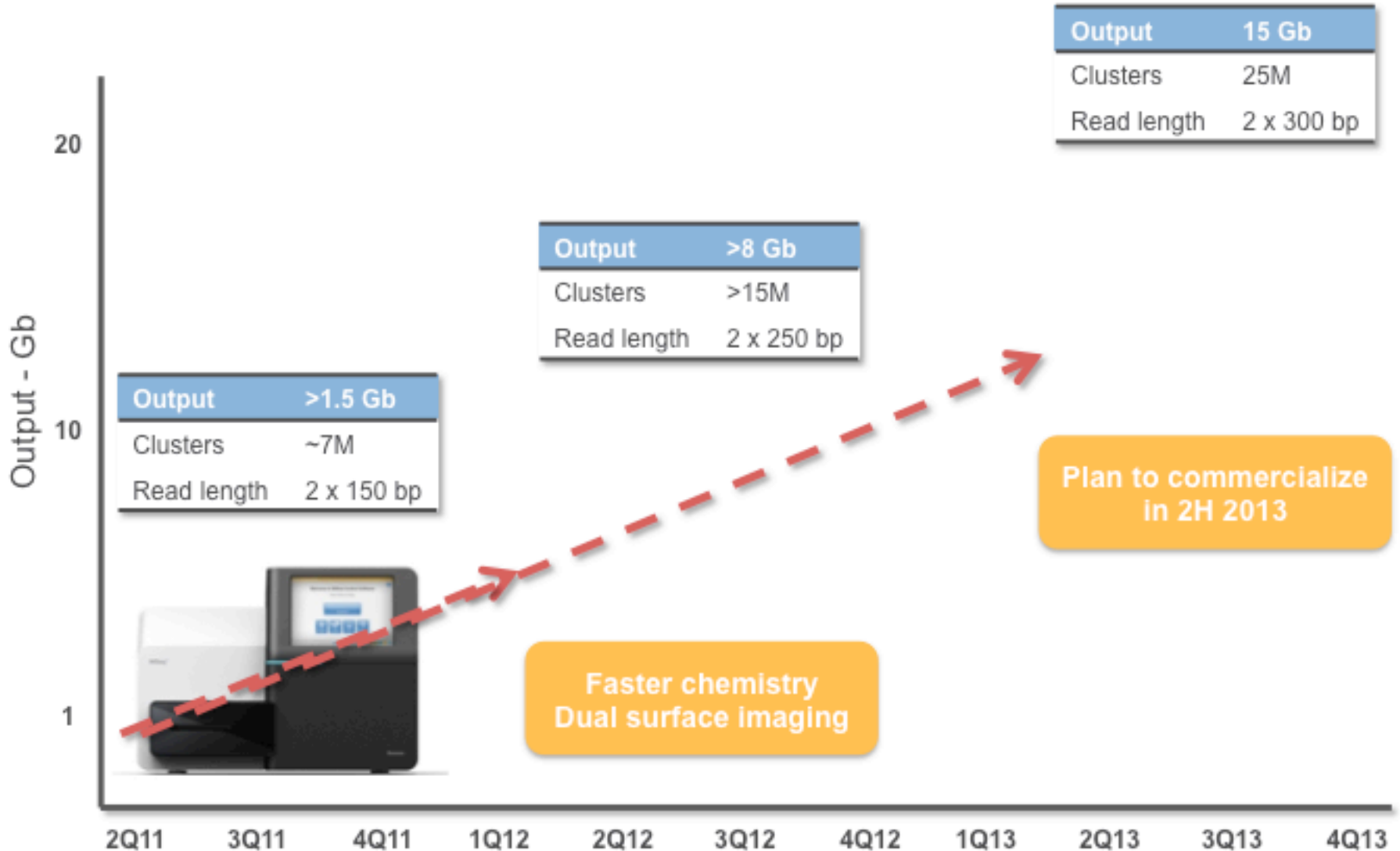
For investigational use only.  
Performance characteristics have not been established.





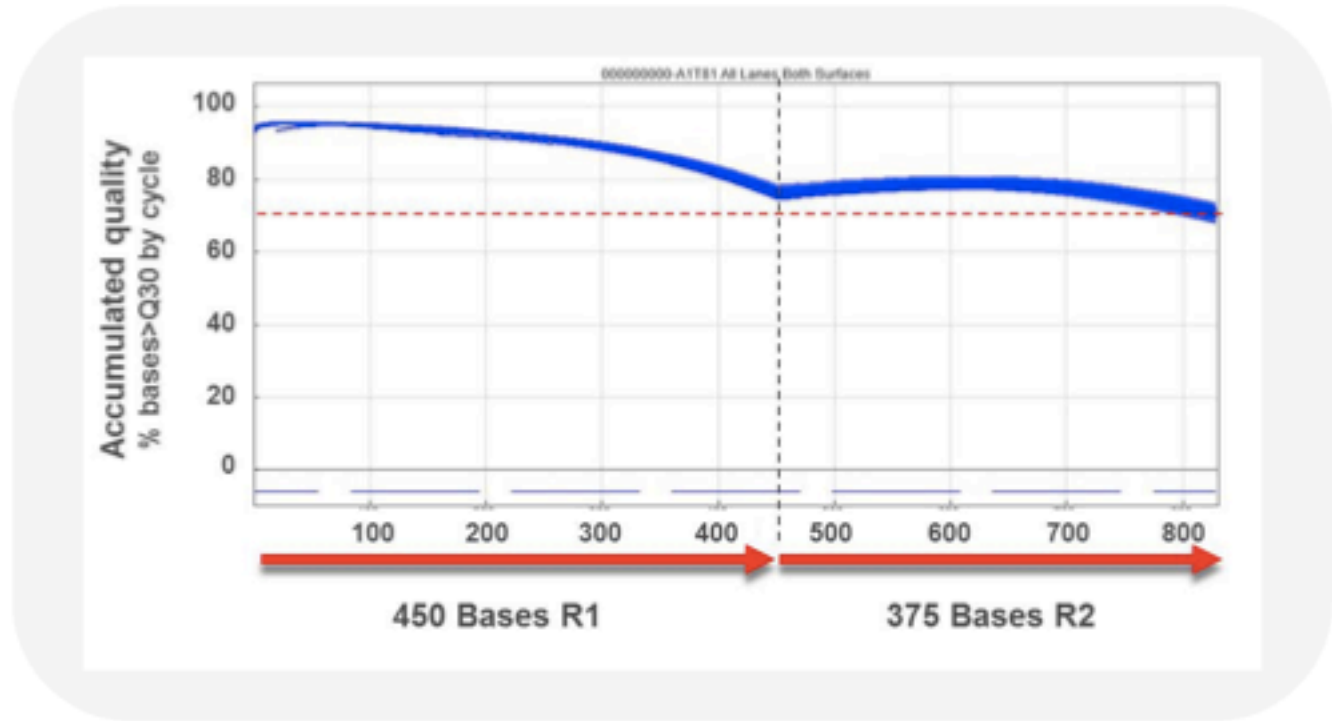
# MiSeq – Continuous Performance Improvements

Path towards 15Gb per run; enabling broader range of applications



# MiSeq – R&D Demonstrated Scalability

Long read runs in excess of 20Gb



Output	22 Gb
Clusters	26.5M
Read length	450 (R1), 375 (R2)
Quality	71% ≥Q30
MR	1.7% (R1), 2.4% (R2)

Demonstrated by Illumina R&D



# 遺伝子診断に向けたシーケンスパネル製品

# TruSight 疾患パネル

- ▶ 専門家が選んだコンテンツにより、次世代シーケンサーによる特定疾患におけるテストの開発を可能に
  - **TruSight Autism:** 自閉症に関連する特徴の評価をアシスト
  - **TruSight Cancer:** 癌の予測マーカーに関連する遺伝子をターゲット
  - **TruSight Cardiomyopathy:** 心筋症の遺伝的要因の同定にフォーカス
  - **TruSight Inherited Disease:** 重篤で小児発症の劣性遺伝子にフォーカス
  - **TruSight Exome:** 希少疾患に関連すると思われるエクソームを選択

The graphic features five blue square icons arranged in a horizontal line, connected by a thin orange line. From left to right: 1. A DNA double helix icon labeled 'Rare Disease'. 2. An icon of a family (two adults and two children) labeled 'Inherited Disease'. 3. A puzzle piece icon labeled 'Autism'. 4. A white ribbon icon labeled 'Cancer'. 5. A heart icon labeled 'Cardiomyopathy'. The background is white with faint orange DNA sequence characters scattered around the icons.

## Introducing TruSight Portfolio

New Possibilities for Next-Generation Sequencing

[▶ LEARN MORE](#)

◀ ▶

For research use only and not intended for diagnostic use; however, labs can leverage this content to develop their own unique targeted tests in accordance with CLIA regulations

# TruSight コンテンツデザイン

## TruSight 自閉症

- Kennedy Krieger 研究所のDr. Jonathan Pevsnerグループと開発
- 発達遅延に關与する特徴から自閉症の同定および分類



## TruSight 癌

- Cancer Research 研究所のDr. Nazneen Rahmanグループと開発
- 癌との關連がある生殖細胞(胚細胞)の遺伝子変異および癌のGWASからのSNP



## TruSight 心筋症

- ハーバード大医学部のDr. Heidi Rehmグループとの開発
- よりよい治療と家族スクリーニングのための心筋症タイプの分類



## TruSight 遺伝疾患

- Children's Mercy HospitalのDr. Stephen Kingsmoreグループとの開発
- 遺伝医学者の検査ニーズに基づき、小児発症の重篤で劣性の疾患にフォーカス



## TruSight エクソーム

- Human Genome Mutation Databaseで同定された、疾患原因となる変異をもつ遺伝子にフォーカスしてイルミナがデザイン
- 診断が困難で稀な遺伝性疾患を含む



これらの製品は研究目的に限定されます



# TruSightパネル ターゲット領域

パネル	ターゲット領域	遺伝子数	100x (例)	Nano 75bp x2	Micro 75bp x2	標準 75bp x2
TruSight Autism 自閉症	328.3 Kb	100	34.8 Mb	150 Mb	600 Mb	2.25 Gb
TruSight Cancer 癌	454.1 Kb	94	45.4 Mb			
TruSight Cardiomyopathy 心筋症	243.8 Kb	46	24.3 Mb			
TruSight Exome エクソーム	7.76 Mb	2,761	776 Mb			
TruSight Inherited Disease 遺伝病	3.78 Mb	552	378 Mb			

ご清聴ありがとうございました。