HUMAN WHOLE GENOME SEQUENCING

KINGHORN CENTRE FOR CLINICAL GENOMICS

KINGHORN CENTRE FOR CLINICAL GENOMICS | GARVAN INSTITUTE OF MEDICAL RESEARCH

The Garvan Institute's Kinghorn Centre for Clinical Genomics (KCCG) is offering large scale human whole genome sequencing using the Illumina HiSeq X™ Ten system.

Research sequencing

The Kinghorn Centre for Clinical Genomics (KCCG) is providing access to whole genome sequencing on the Illumina HiSeq X Ten platform for research purposes [1]. The Illumina HiSeq X Ten system is exclusively for use in human whole genome sequencing.

Specifications

Sample	1 µg high quality genomic DNA (TruSeq Nano); 3 µg high quality genomic DNA (TruSeq PCR-Free). Research sequencing is conditional on appropriate ethical approvals.
Turn-around Time	4 – 6 weeks for 90% of samples from date of acceptance to date of data delivery. Remaining samples may take up to 4 additional weeks to complete *dependant on project size.
Data specifications	HiSeq X Reagent Kit v2.5 • >100 Gb raw data • >75% bases >= Q30 at 2x 150bp • Genome coverage >= 30x as defined by Illumina [2,3]
Data Format	Raw data (FASTQ). Alignments (BAM) and variants (VCF) available through bioinformatics service (see below)
Bioinformatics services	BAM and VCF files are generated using KCCG's implementation of the GATK best practices pipeline on DNAnexus.
Delivery	Cloud-based data delivery via DNAnexus [4]. Hard drive delivery available on request.

Test data - NA12878

High quality data from our HiSeq X Ten system is freely available at http://garvan.org.au/kccg-sample-data.

Contacts

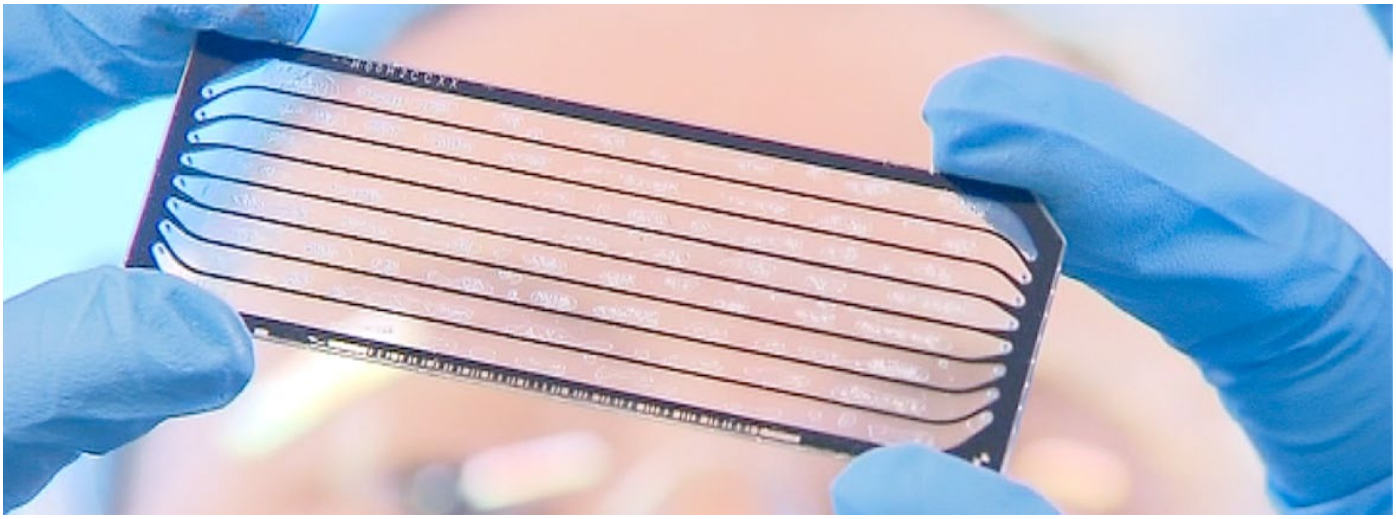
For sequencing services:

AGRF (Australian Genome Research Facility)
W: http://www.agrf.org.au/campaigns/whgs
T 03 9321 3716 **E** humanWGS@agrf.org.au

Ramaciotti Centre for Genomics
W: http://www.ramaciotti.unsw.edu.au/
T 02 9385 1658 **E** nextgenseq@unsw.edu.au

Further information

Visit: http://www.garvan.org.au/kccg.



WHY WHOLE GENOME SEQUENCING?

Uniform coverage

Whole genome sequencing (WGS) provides consistent coverage. Using the latest version of Illumina's SBS chemistry, we routinely achieve 35.5X coverage per lane of sequencing [3] (see graphic below).

Exhaustive representation of the genome

The consistent and genome-wide coverage provided by whole genome sequencing (WGS) makes it possible to call copy number variants, chromosomal alterations, such as translocations and inversions, and much more.

Extensive data analysis applications

Whole genome sequencing (WGS) covers protein coding genes, non-coding RNAs, mitochondria and regulatory regions, regardless of annotation. Whole genome data can be re-queried as new functional elements are identified and as answers to new questions are sought

Our Technology

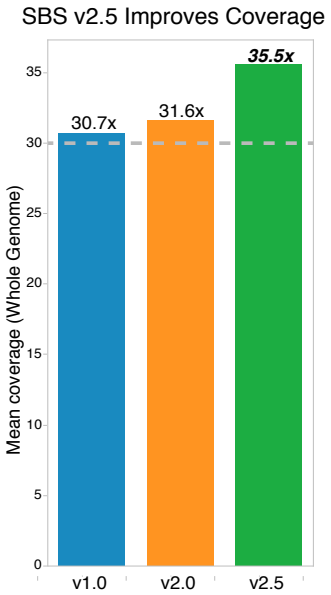
Following the Illumina SeqLab [5] principles, we:

- use the latest version of Illumina's chemistry (Version 2.5) which makes our sequencing coverage deeper and more consistent.
- use automated liquid handling and library preparation systems to streamline laboratory workflows for maximum efficiency.
- offer seamless integration from sample accession to data analysis.

HiSeq X Performance

The statistics below describe the HiSeq X System performance at the Kinghorn Centre for Clinical Genomics since adopting v2.5 technology.

	v2.5 (n = 1,646)
Mean Coverage	35.5X
Clusters PF	431M
Yield	129.54 Gb
Mean Quality Score R1/R2	38.36/33.38
Av % Bases >=Q30	82.55
Median Fragment Size	416 bp
% Unique Read Pairs	90.51



Notes

1 Pricing is subject to capacity and agreed terms and conditions from AGRF or Ramaciotti Centre (for orders from within Australia).

2 Higher coverage also available (e.g. equivalent to 60X, 90X, etc.).

3 See the Illumina Product Sheet (http://tinyurl.com/pj4kmvt) for details.

4 DNAnexus delivery includes 90 days free storage.

5 See the Illumina SeqLab webpage (http://tinyurl.com/q3yf44k).

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