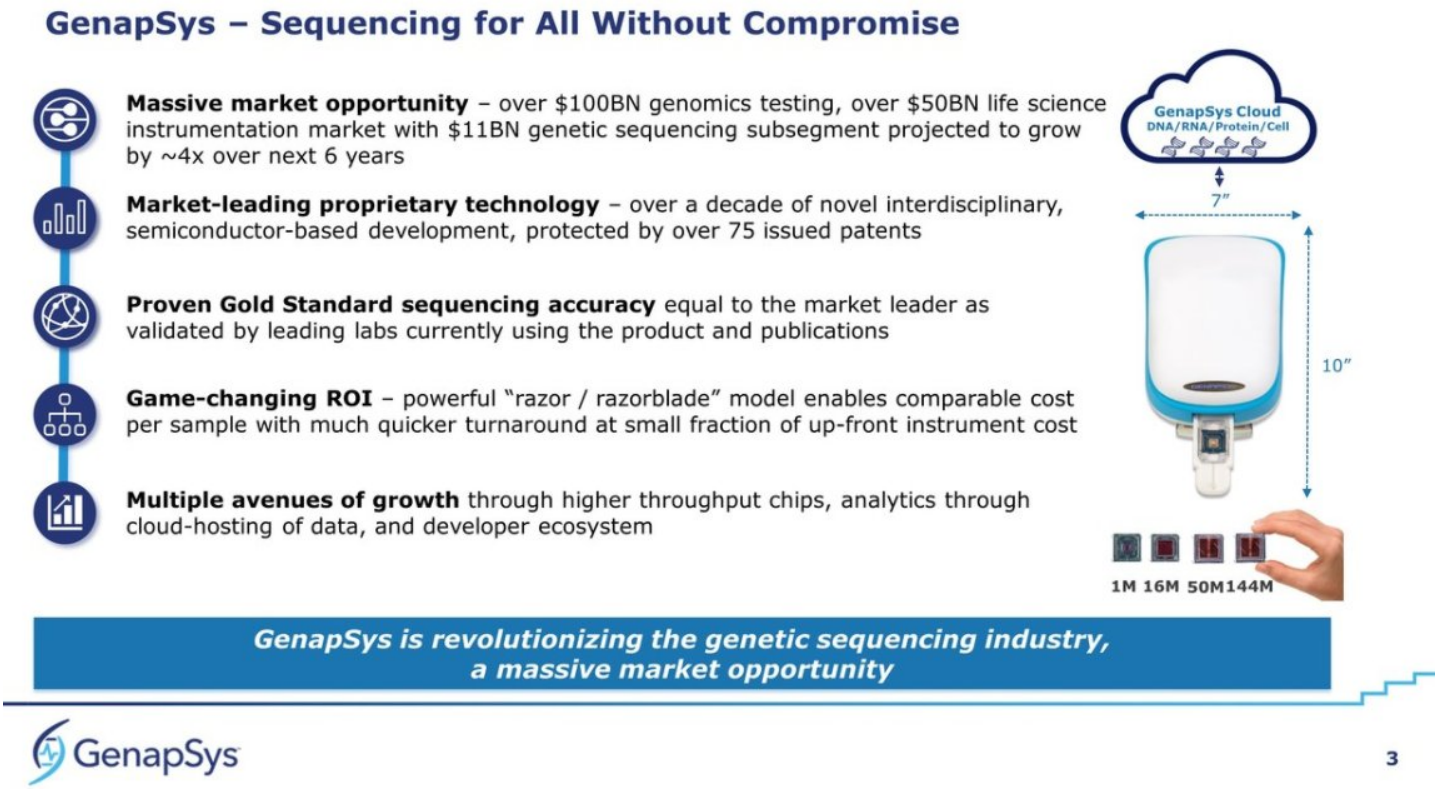
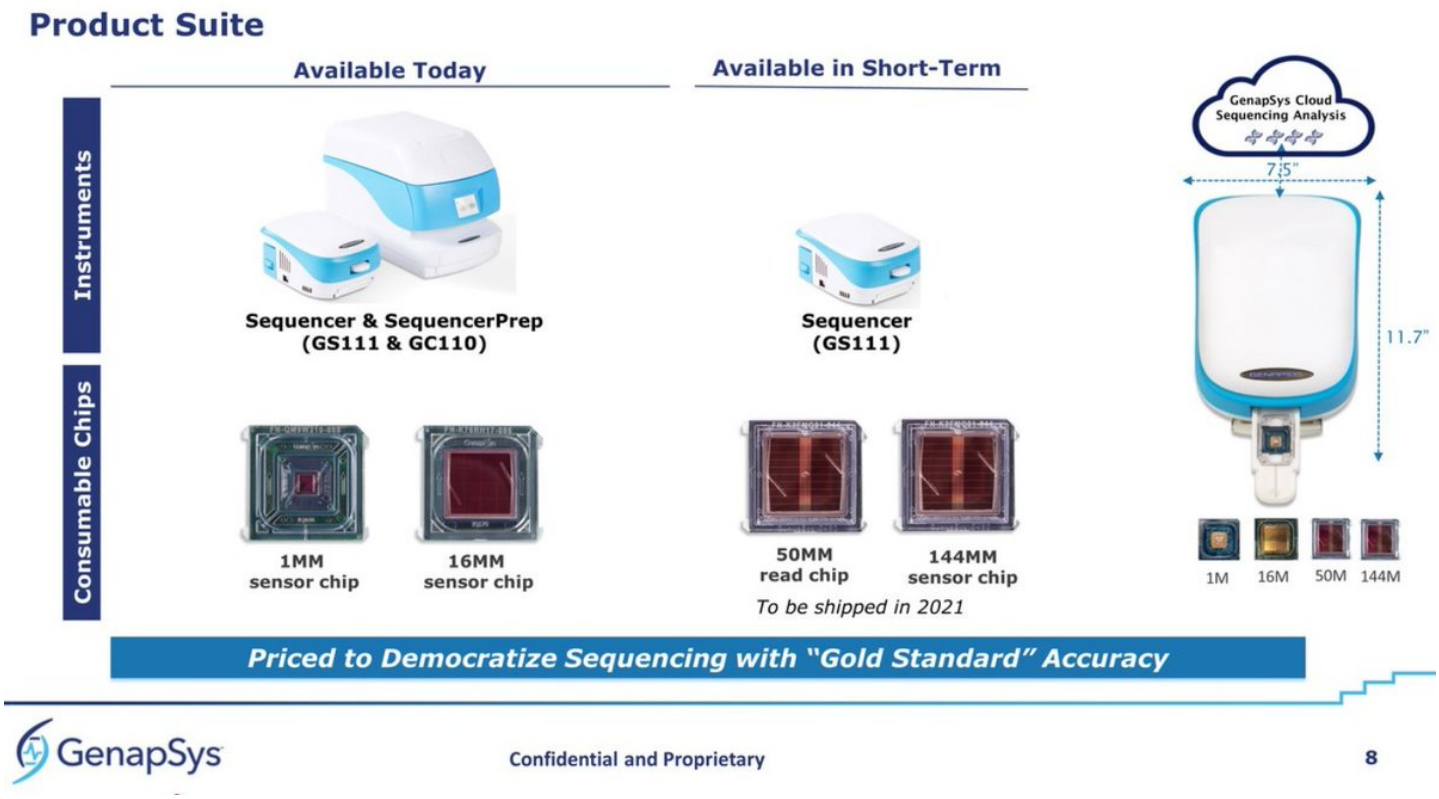
Twitter Thread by Albert Vilella

Albert Vilella
@AlbertVilella

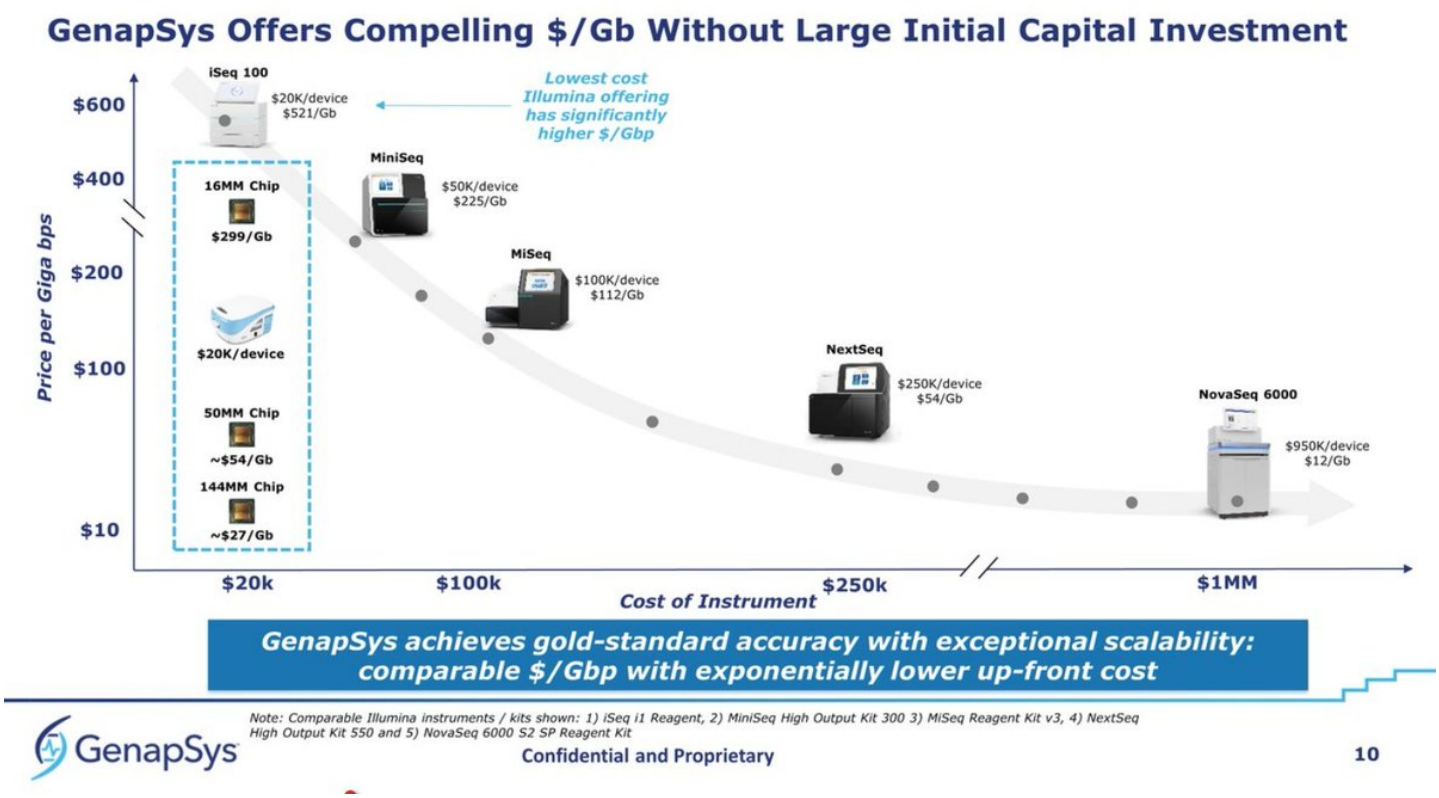
#JPM2021 @GenapSys My Highlights: I think it's fair to say I am more excited than most about this #NGS company, as I see them as an example of how to enter the market while keeping a small profile ($249M raised so far).



They now are aiming at 2021 to ship two new chips: 50MM read chip and 144MM sensor chip (not sure what the difference is between read/sensor).

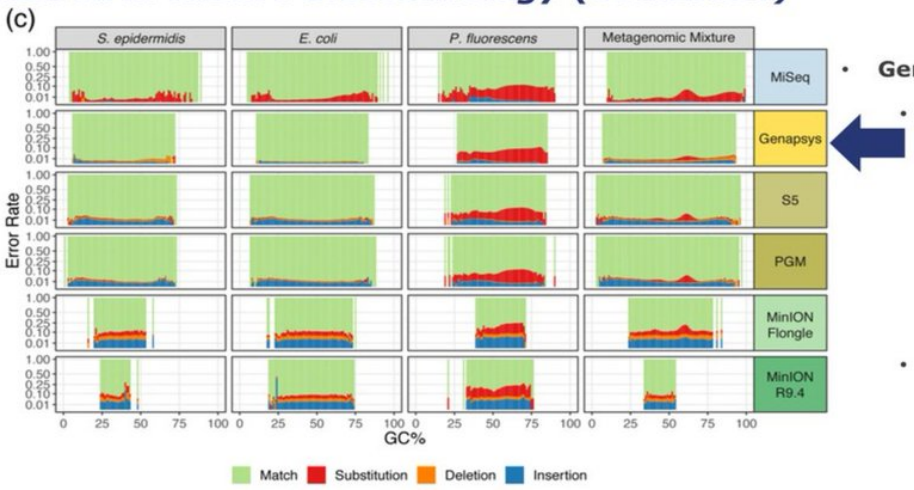


They show a slide of price per Gb with #Illumina products as a reference, and their products now lined up, I think, for the first time with price per Gb info. Lowest will be the 144MM chip at ~$27/Gb.



As their technology is closer to the #IonTorrent than to the cycle-by-cycle SBS-based sequencing methods, they should have microindel errors in their profile. These look much better, almost null, than the S5 data they show in comparison.

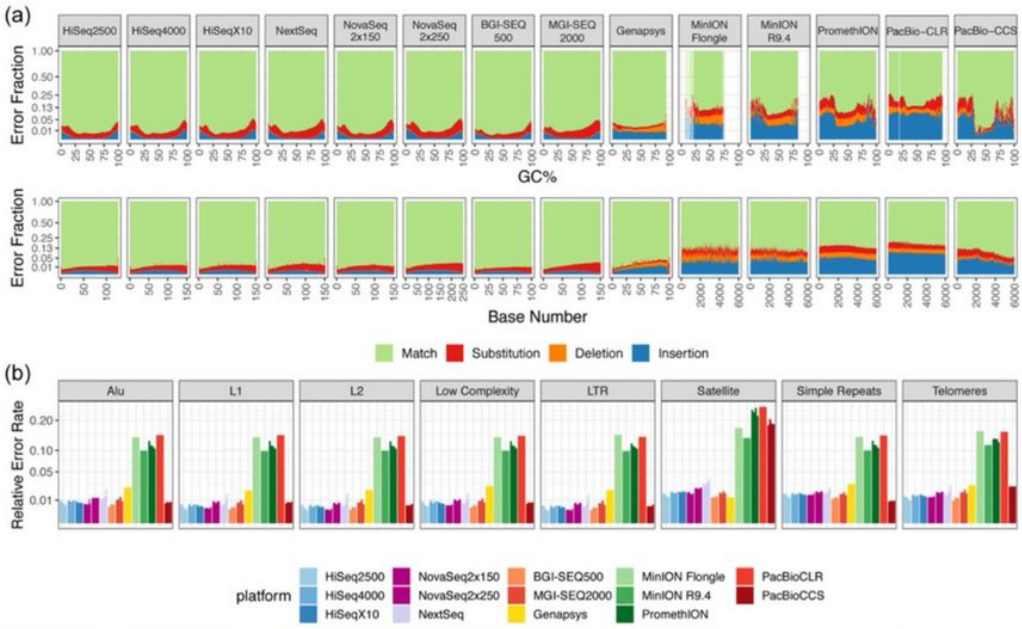
Multi-Platform Assessment of DNA Sequencing Performance, pre-print to be Published in *Nature Biotechnology (Continued)*



- **GenapSys data demonstrated:**
  - Gold-standard accuracy across all error types (mismatch, insertion, deletion) compared to established platforms: Illumina, S5 Ion Torrent, PGM, and MinION, across various samples, high, low, medium GC% and Metagenomic mixture.
  - Excellent coverage from GC content of 30-70% and across all genomes in alignment with the established platforms

The next slide, also about error rates, either for each base as the run progresses or sliced in GC% windows, has a lot of info, but difficult to read. I think the summary is that the @Genapsys profile shows a bit of microindel error but otherwise comparable to 100bp ILMN data.

Multi-Platform Assessment of DNA Sequencing Performance, pre-print to-be Published in *Nature Biotechnology (Continued)*



The error profile starts low, very low actually, and then it hikes up after cycle 50-100+. In the slide below, they've been naughty and put a textbox on top of their error blue line, trying to hide it: don't think we won't notice these tricks! ■

ASHG 2019 Report and Presentation by David Harrison

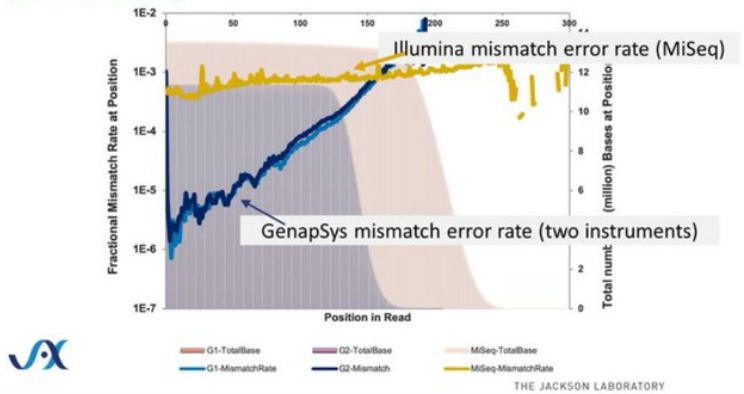
The Jackson Laboratory

"We performed ten runs using the GenapSys sequencer and observed minimal variability between GenapSys device runs... both machines [Illumina and GenapSys] performed almost identically" - David Harrison, Bioinformatics Analyst, The Jackson Laboratory

Summary of Findings

- Performed 10 runs using the GenapSys sequencer with minimal variability
  - Run – Run / Machine - Machine
- Consistent high-quality data of >10MM reads at >140bp mean length with high mapping rates is obtainable
- Genome coverage distributions and GC content analysis show no bias when compared to Illumina MiSeq data
- The lower mismatch rate results in high sensitivity and specificity when performing SNP detection
- Preliminary analysis suggest this low error rate is consistently better than similar short-read data generated by Illumina MiSeq SBS
- The GC110 device greatly reduced the hands-on time for clonal amplification

Mismatch Rate vs. Read Position



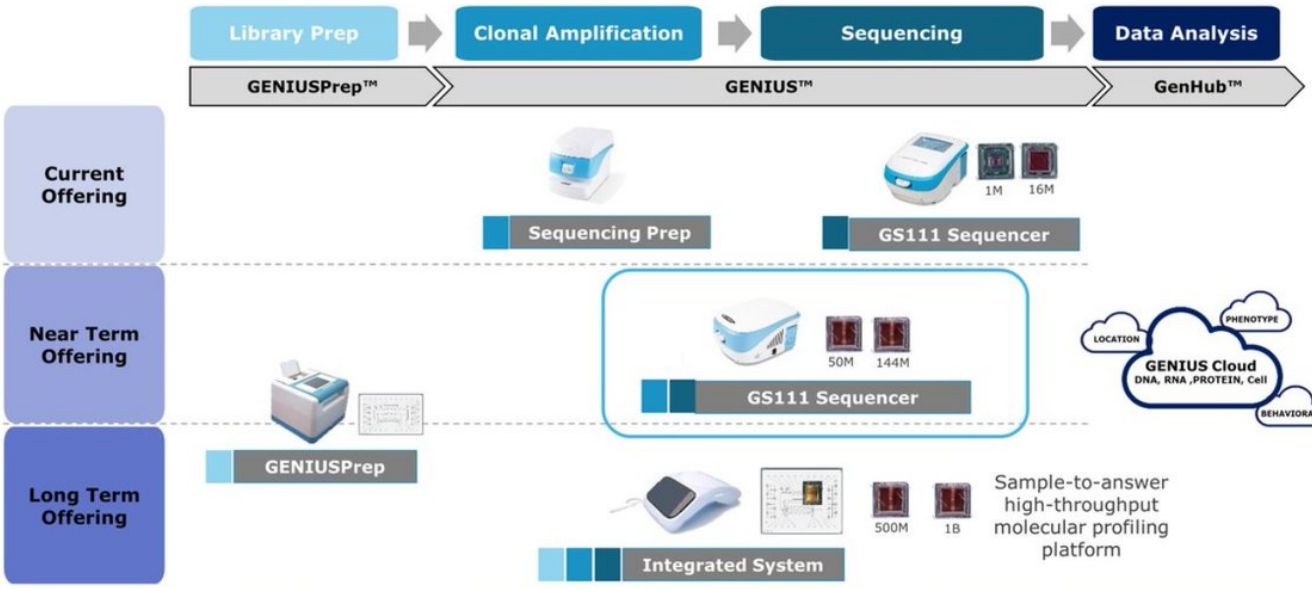
'GenapSys produced a ***lower mismatch error rate than Illumina*** *MiSeq,* ***across all low/medium/high GC*** *microbial samples.... GenapSys is able to reproducibly detect a similar amount of high confident variant calls as Illumina MiSeq, no real difference."*

Source: The Jackson Laboratory; *"Performance of a new electric based sequencer demonstrates lower mismatch error rates and higher sensitivity in SNV detection"*
The Jax's ASHG presentation is available: https://landing.genapsys.com/jax-lab-webinar-ashg-2019

They say they've placed or booked more than 110 instruments so far (does that count early access?). All this is based on a 10K GS111 Sequencer + 10K Sequencing Prep instrument. They show long term plans for a GENIUSPrep (high throughput sample prep), but also ...

... but also bigger chips (500MM / 1000MM chip), as well as an Integrated System (zoom in).

End-to-End Solution: Next Generation System Evolution



GenapSys

Details of the 50M Chip. They seem to have gone from 100bp to 150bp standard now, but raw accuracy still placed at 99.9% at position 125bp. This may be the actual 144M chip but with standard Poisson loading, rendering a bit over 50M reads, or

8Gb per run, which is impressive...

50M Chip Sequencing Results

# reads in FASTQ	>50M
# base in FASTQ	8.0Gb (first read)
Read Length	150
% bases >Q30 (e.g. >99.9% raw accuracy)	>80%
Raw Sequencing Accuracy (at Position 125)	99.9%

- Preliminary data shows gold standard performance and accuracy for 50M chip using bacterial and human genomes
- Initial demonstration of Paired-End Sequencing capability (ASHG 2020)
- Development is progress for a fully-automated end-to-end solution with integrated amplification and sequencing
- Project managed through a rigorous phase gate development process for launch in 2021

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... with a $20K capital investment, comparing the 50M @Genapsys at 8Gb ($54/Gb) and the #Illumina #iSeq100 at 1.2Gb ($542/Gb), that's roughly 8x better yield for Genapsys, and 10x better price per Gb. It makes the #iSeq100 look rather underwhelming in comparison.

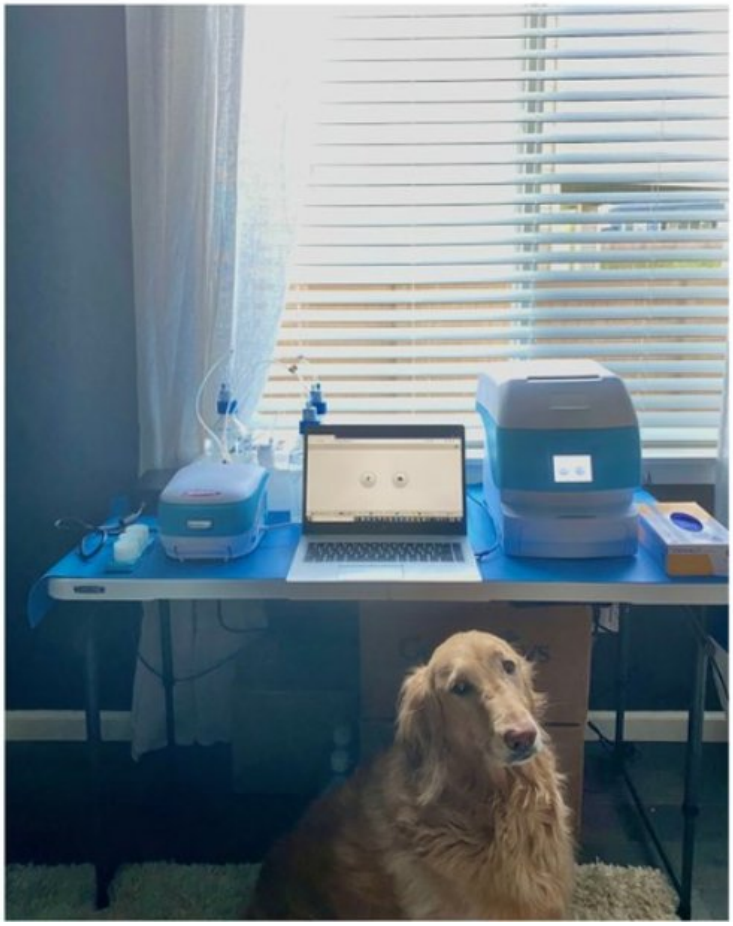
The slide about 2021 plans changes the naming from 144M chip to "G4-50M and G4-100M", so this could mean that the nomenclature changes to reads per run rather than the size of the chip. Maybe both based on the 144M chip, but with better loading giving the 50M->100M jump in reads?

No reference to paired-end reads, only indications that 150bp may not be the end of it, and they may be able to go longer than that. Good news about the G4-100M, as it seems they have scope of improvement in loading.

- Commercial Launch of G4-50M chip to broaden application utility
- Completing the development of G4-100M chip to broaden application utility

Bonus picture of a "Personalize Genomics!" lab, or lab-at-home.

Personalized Genomics!



@d2unroll unroll