Genomic Data Lakehouse Architecture

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Not an Agenda per se...

- 1. Does *Data Lakehouse* = Data Warehouse + Data Lake?
- 2. Why does Genomic Data + Data Lakehouse ≠ *Genomic Data Lakehouse* ?
- 3. Is the SeQuiLa project to the rescue?

About me

- ► Chief Data Architect @GetInData | Part of Xebia
- ► Research Assistant^a at Warsaw University of Technology
- ▶ putting the finishing touches to his Phd dissertation...
- ▶ keen long distance runner, gravel bikes enthusiast and absolutely in love with the Italian Lakes!



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Data platforms architectures

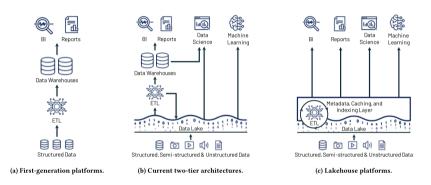


Figure: Evolution of data platform architectures to today's two-tier model (a-b) and the new Lakehouse model (c)[1]

Data Lakehouse design principles

- low-cost and efficient storage for very large-scale heterogeneous data backed by cloud object storage systems
- first-class support for machine learning and data science workloads with distributed
 DataFrame-like APIs
- ► state-of-the-art SQL analytical queries performance
- open direct-access file formats such as Parquet, ORC or DeltaLake

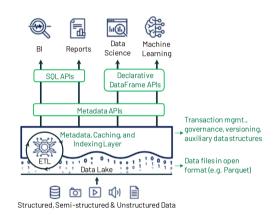


Figure: Data Lakehouse design[1]

Challenges of Cloud Genomic Data Lakehouse

- genomic analyses primitives like e.g. range joins, depth of coverage or pileup summary
- ▶ genomic-specific file **formats** like BAM/CRAM or VCF
- secondary analyses pipelines efficient reusing of the existing tools or native implementations
- ► GWAS-specific statistical methods
- ▶ set unified of unified APIs (**SQL** and **DataFrame**) shift from traditional shell scripting/CLIs
- ▶ **cloud** challenges: legal, skills
- ► *ephemeral* computing vs long-lived infrastructure the need of **IaC**^a approach

ahttps://en.wikipedia.org/wiki/Infrastructure_as_code

The SeQuiLa project (1/2)

- ▶ originally started as a Phd project in 2018^a
- meant for the elements of secondary and tertiary analysis
- extensions to the Apache Spark engine[8, 7, 6]
- ▶ **SQL** and **DataFrame** APIs
- support for bioinformatics file formats
- \blacktriangleright a set of **cloud** recipes^b



a://biodatageeks.github.io/sequila/

^bhttps://github.com/biodatageeks/sequila-cloud-recipes

The SeQuiLa project (2/2)

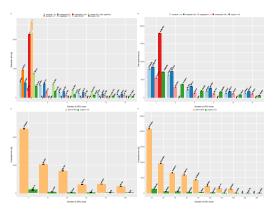


Figure: Depth of coverage[6]

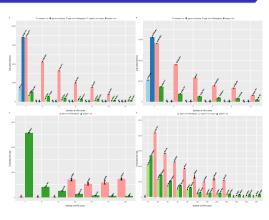
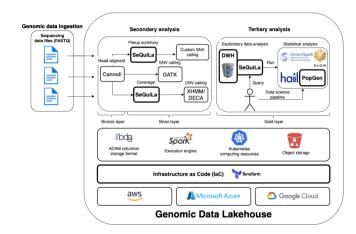


Figure: Pileup summary[6]

Cloud Genomic Data Lakehouse

- ► cloud-agnostic
- secondary and tertiary analysis
- ► medallion architecture[3]
- ► Apache Spark-based projects like ADAM[5], DECA[4], VariantSpark[2]



The 3 take-home messages

- ▶ Does Data Lakehouse =
 Data Warehouse + Data Lake ? No^a
- Why does Genomic Data + Data Lakehouse ≠ Genomic Data Lakehouse ? – It's complicated^b
- ► Is the SeQuiLa project to the rescue? Absolutely yes!^c

^aa novel data platform architecture

^bbioinformatics file formats and distributed genomic operations

^cwell, still some room for improvement

Thank you! Q&A marek.wiewiorka@gmail.com

Bibliography

- Michael Armbrust andothers. "Lakehouse: a new generation of open platforms that unify data warehousing and advanced analytics". in Proceedings of CIDR: 2021.
- [2] Arash Bayat andothers. "VariantSpark: Cloud-based machine learning for association study of complex phenotype and large-scale genomic data". in GigaScience: 9.8 (august 2020). ISSN: 2047-217X. DOI: 10.1093/gigascience/giaa077.
- [3] Ron L'Esteve. "Databricks". en. in The Azure Data Lakehouse Toolkit: Berkeley, CA: Apress, 2022, pages 83–139. ISBN: 978-1-4842-8232-8 978-1-4842-8233-5. DOI: 10.1007/978-1-4842-8233-5_3. URL: https://link.springer.com/10.1007/978-1-4842-8233-5_3 (urlseen 11/02/2023).
- [4] Michael D. Linderman andothers. "DECA: Scalable XHMM exome copy-number variant calling with ADAM and Apache Spark". inBMC Bioinformatics. 20.1 (october 2019). Publisher: BioMed Central Ltd., pages 1–8. ISSN: 14712105. DOI: 10.1186/S12859-019-3108-7/TABLES/2. URL: https://bmcbioinformatics.biomedcentral.com/articles/10.1186/S12859-019-3108-7 (urlseen 03/01/2023).
- [5] Matt Massie andothers. "Adam: Genomics formats and processing patterns for cloud scale computing". in University of California, Berkeley Technical Report, No. UCB/EECS-2013: 207 (2013), page 2013.
- [6] Marek Wiewiórka andothers. "Cloud-native distributed genomic pileup operations". inBioinformatics: (december 2022). byeditorPeter Robinson. ISSN: 1367-4803. DOI: 10.1093/bioinformatics/btac804. URL: https://academic.oup.com/bioinformatics/advance-article/doi/10.1093/bioinformatics/btac804/6900922.
- [7] Marek Wiewiórka andothers. "SeQuiLa-cov: A fast and scalable library for depth of coverage calculations". in GigaScience: 8.8 (august 2019). ISSN: 2047-217X. DOI: 10.1093/gigascience/giz094. URL: https://academic.oup.com/gigascience/article/doi/10.1093/gigascience/giz094/5543653.
- [8] Marek Wiewiórka andothers. "SeQuiLa: an elastic, fast and scalable SQL-oriented solution for processing and querying genomic intervals". inBioinformatics: 35.12 (june 2019). byeditorJohn Hancock, pages 2156–2158. ISSN: 1367-4803. DOI: 10.1093/bioinformatics/bty940. URL: https://academic.oup.com/bioinformatics/article/35/12/2156/5182295.