genomDE Berlin 2025

Genome and Epigenome Diagnostics with Nanopore Sequencing

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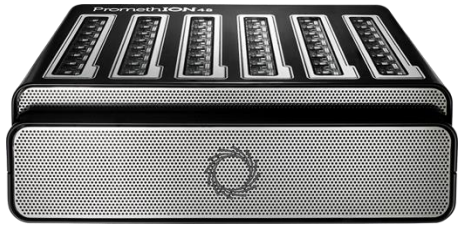
Stephan Ossowski
Institute of Medical Genetics and Applied Genomics

Universitätsklinikum
Tübingen

Nanopore Diagnostics Platform (Accredited)

Lab technicians

Sample Preparation

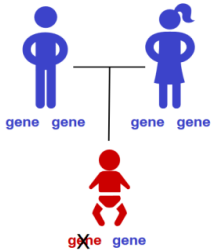


Sequencing

Clinicians

Genetic Analysts

Patients



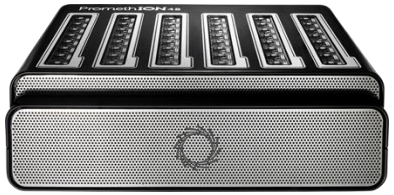
Sample collection

IT Infrastructure

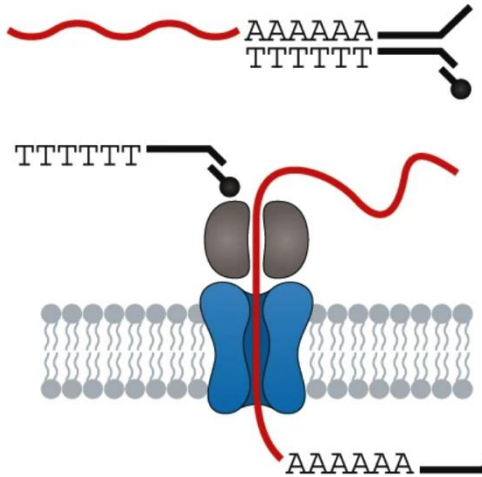
Genome Analysis Software

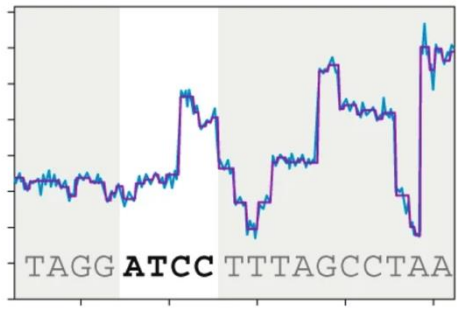
Databases

Genome & Methylome Diagnostics with Nanopore

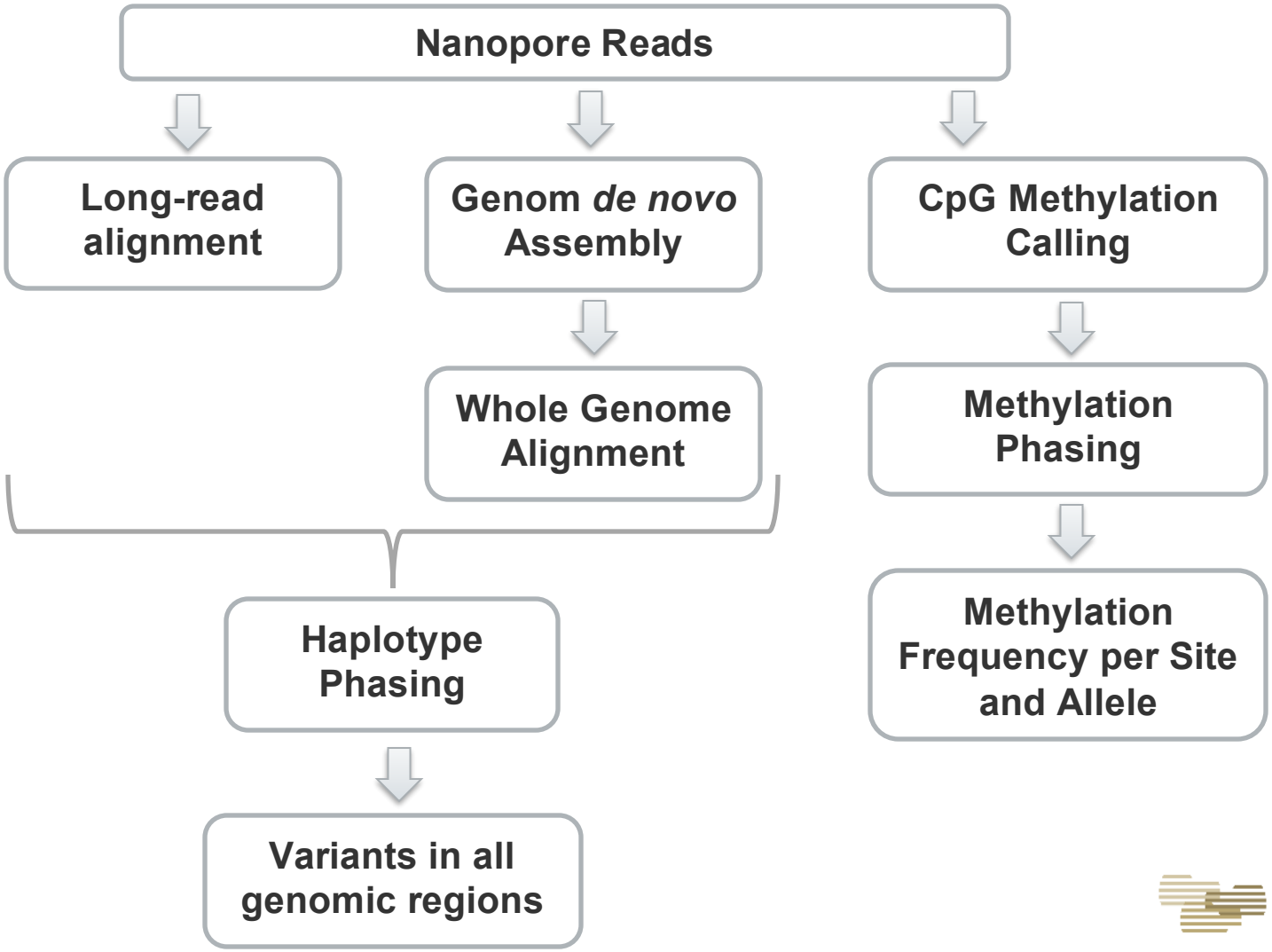


Oxford Nanopore





New analysis options:



Nanopore Analysis Pipelines

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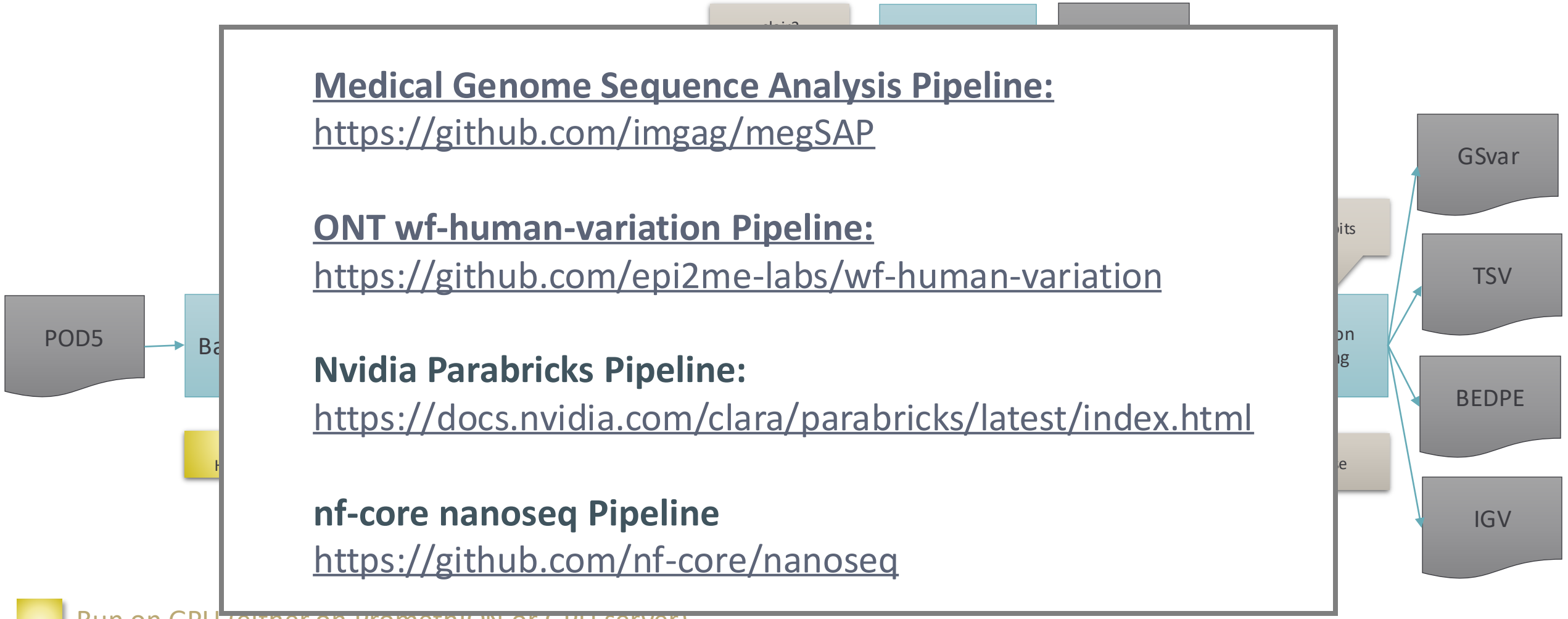

Medical Genome Sequence Analysis Pipeline:
https://github.com/imgag/megSAP

ONT wf-human-variation Pipeline:
https://github.com/epi2me-labs/wf-human-variation

Nvidia Parabricks Pipeline:
https://docs.nvidia.com/clara/parabricks/latest/index.html

nf-core nanoseq Pipeline
https://github.com/nf-core/nanoseq

Run on GPU (either on PromethION or GPU server)

Run on CPU (on normal CPU server)

megSAP Pipeline

Nanopore Analysis Pipelines

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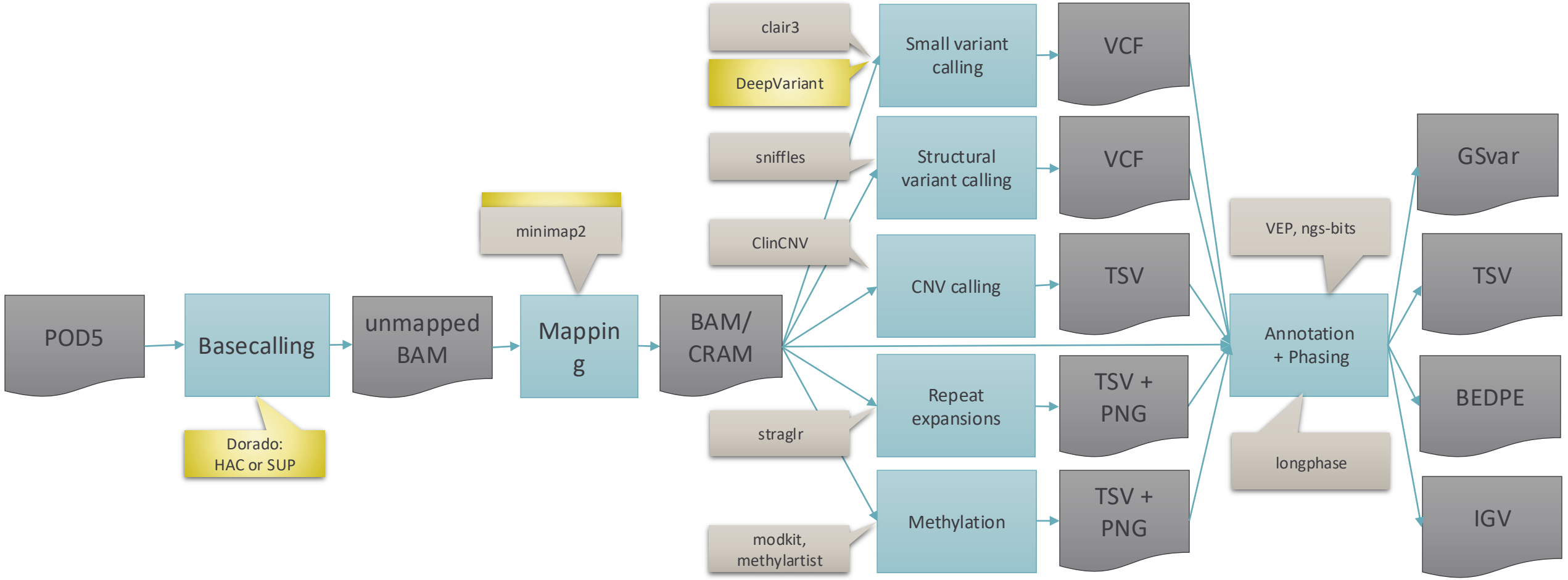

Run on GPU (either on PromethION or GPU server)

Run on CPU (on normal CPU server)

megSAP Pipeline

Clinical Decision Support for Nanopore Diagnostics

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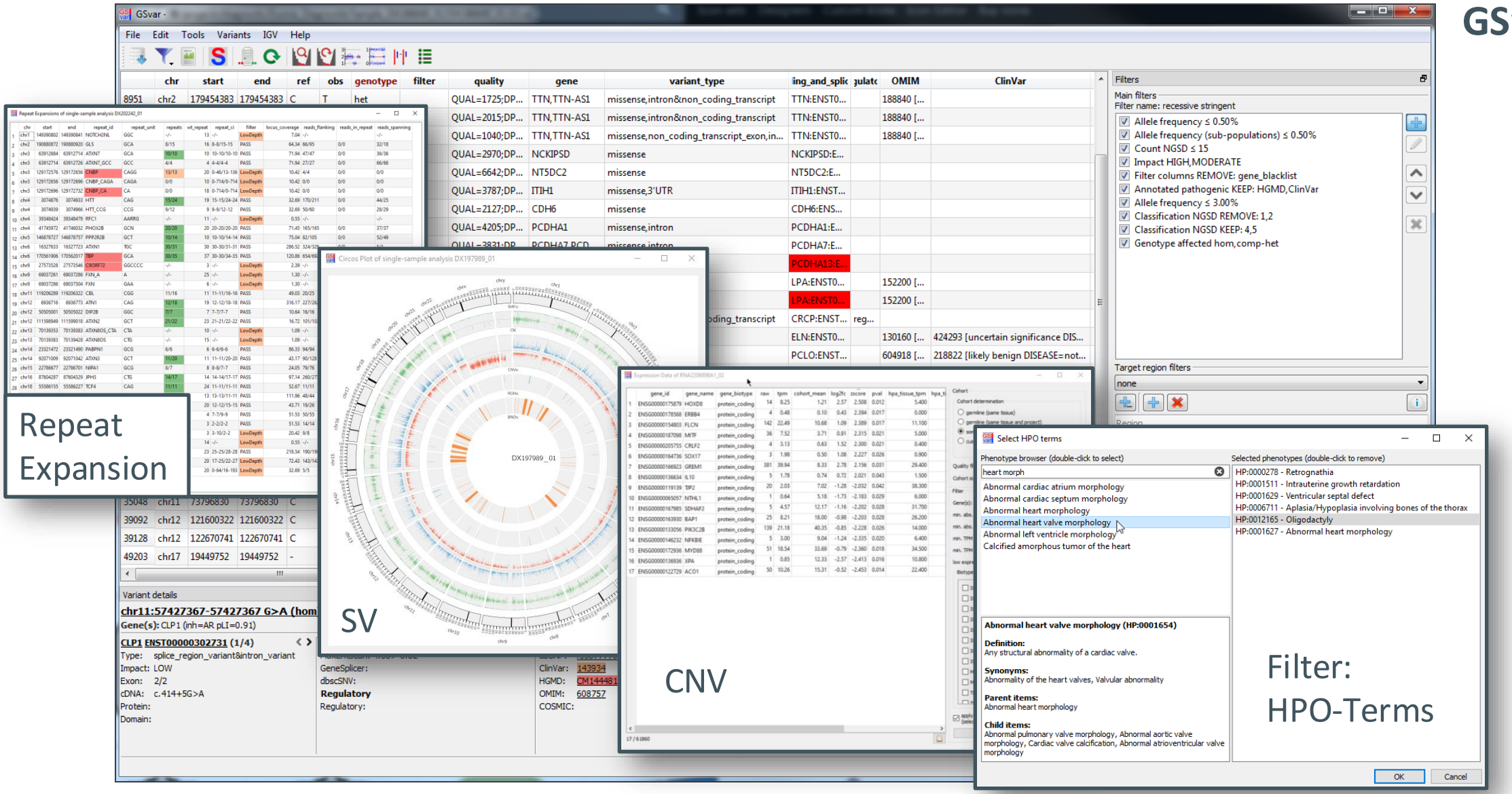

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Benchmarking the Quality of Reads and Variant Calls

Yield per Flowcell and reaching “Diagnostic Coverage”

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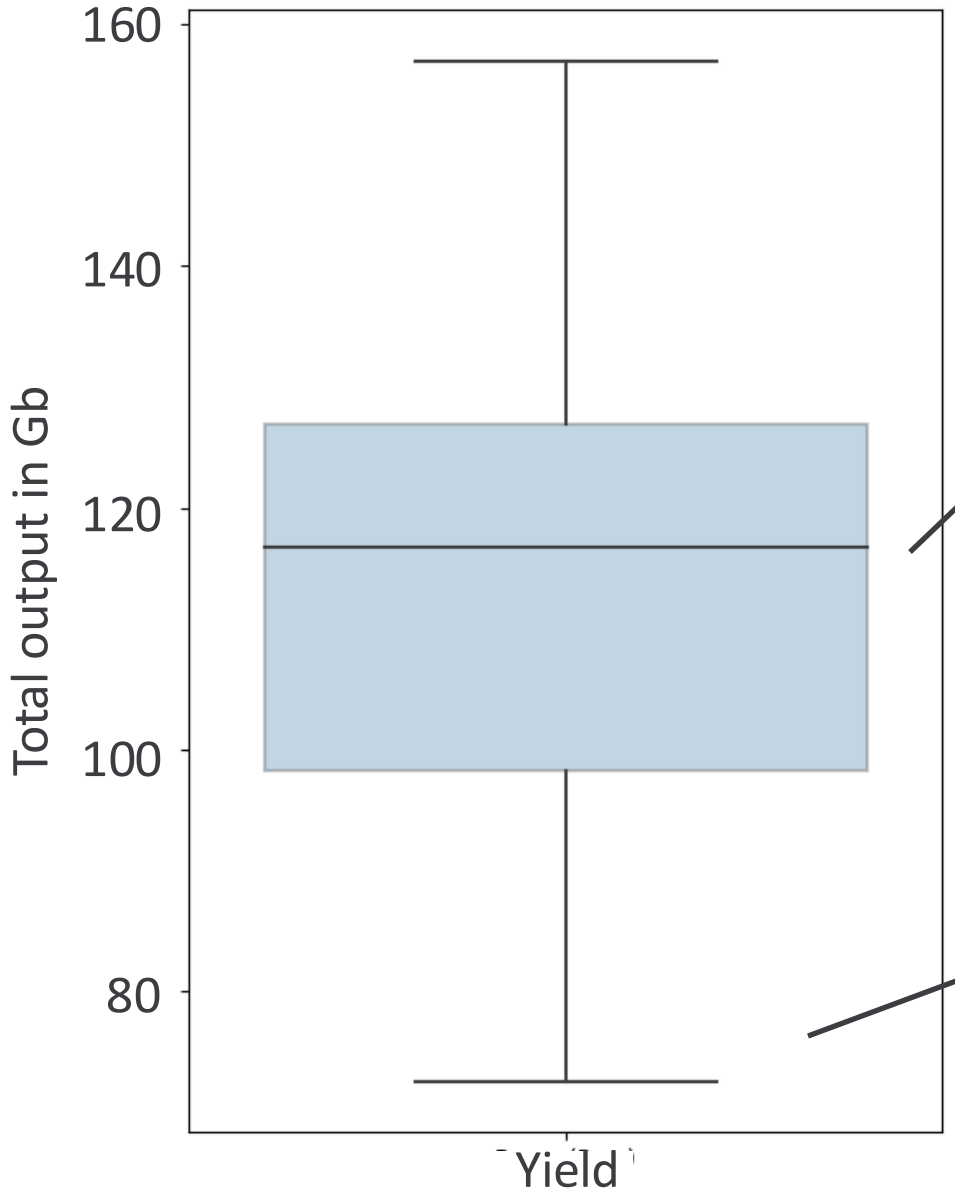
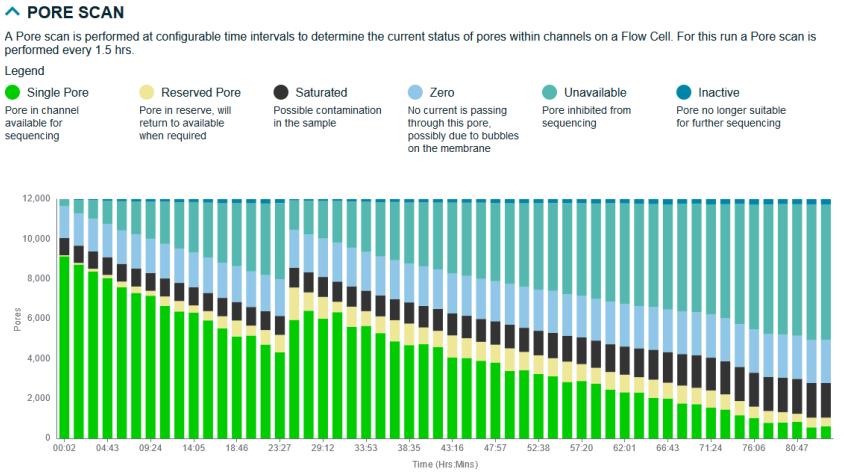
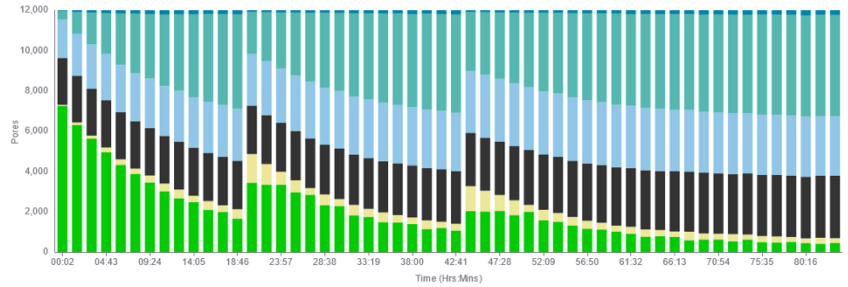
Diagnostic Genome:
35-40x Coverage

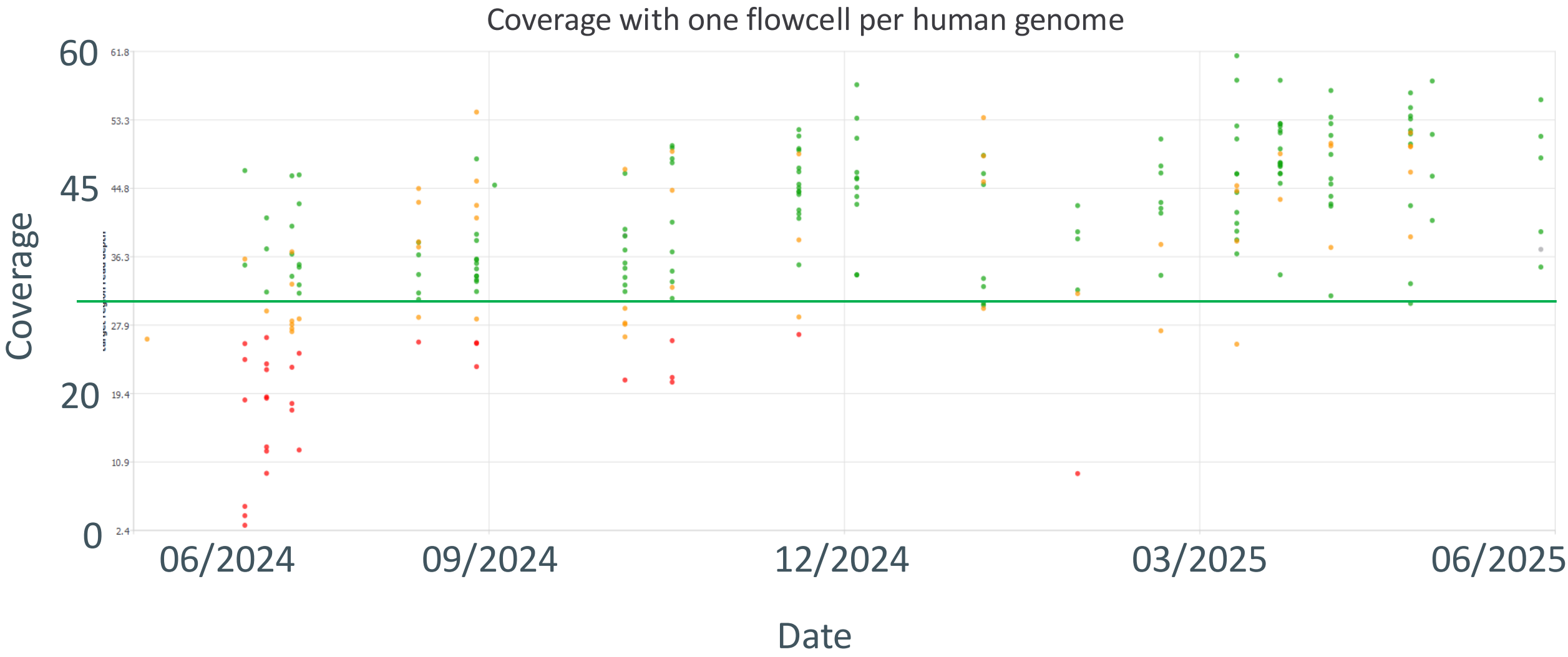
30x Coverage







One Human Genome per Flowcell: Coverage Yield

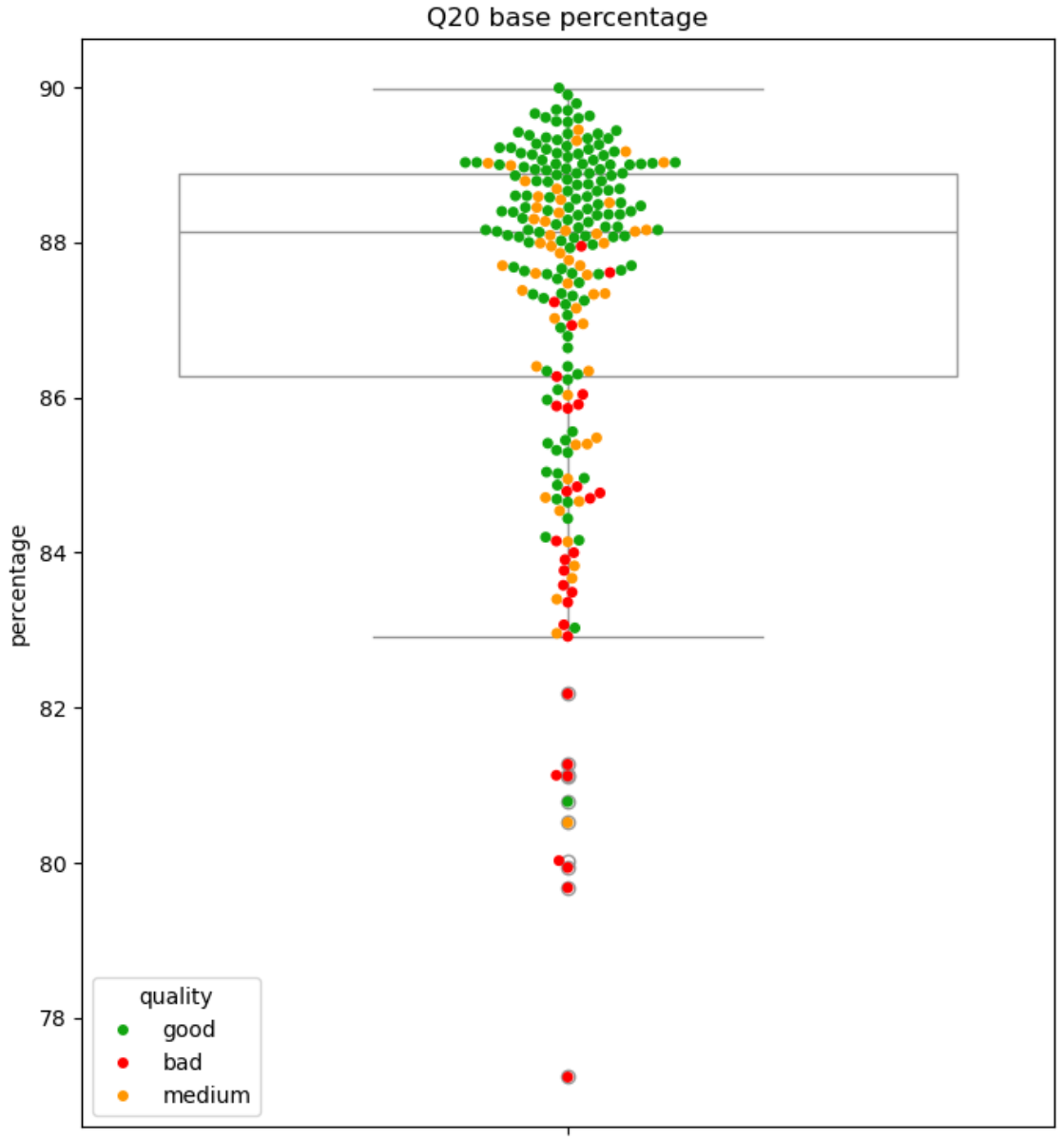


Base Quality

QC-Analysis on 233 diagnostic Nanopore-GS:

Good quality: 80% of bases > Q20

Note: we remove reads with Read-Q < 9 before base quality analysis!



Summary: Essential QC Parameter

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1. Coverage: >= 30x
2. Saturation: >= 20x coverage in > 95% of the genome
3. N50 read length: >10kb (optimal: 20-25kb)
4. Base quality: 80% of all bases have Q > 20
5. Remove reads with Read-Quality < 9

Benchmarking with Genome in a Bottle Reference Data

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NIST

PROJECTS/PROGRAMS

Genome in a Bottle

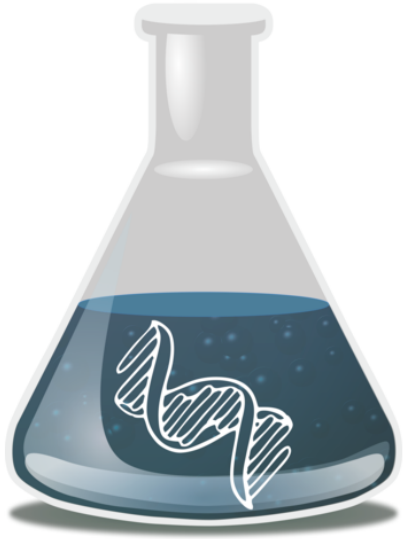
Summary

Consortium hosted by NIST dedicated to authoritative characterization of benchmark human genomes. Sign up for General GIAB and Analysis Team email lists. Public workshops held annually - next workshop will be rescheduled after COVID-19. **Interested in job opportunities with us? Contact Justin Zook at the email in the right panel.**
Click here for the GIAB FAQ

DESCRIPTION

Consortium goals:
The Genome in a Bottle Consortium is a public-private-academic consortium hosted by NIST to develop the technical infrastructure (reference standards, reference methods, and reference data) to enable translation of whole human genome sequencing to clinical practice and innovations in technologies. The priority of GIAB is authoritative characterization of human genomes for use in benchmarking, including analytical validation and technology development, optimization, and demonstration.

Reference samples:
GIAB has currently characterized a pilot genome (NA12878/HG001) from the HapMap project, and two son/father/mother trios of Ashkenazi Jewish and Han Chinese ancestry from the Personal Genome Project (selected because, unlike the pilot genome, they are consented for commercial redistribution). These samples and their IDs from NIST, Coriell, and PGP are in this table.



Reference samples:

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GiaB Benchmark for Coding Regions
March 2025 – Nanopore long-read GS

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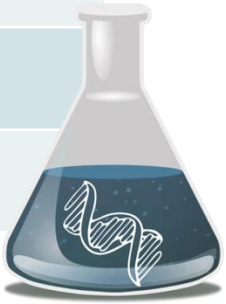
	recall/sensitivity	precision	genotyping accuracy
Exome SNVs	97.25	99.91	99.91
Exome INDELs	91.7	98.18	99.54
Nanopore SNVs	1.000	0.999	1.000
Nanopore INDELs	0.966	0.983	0.998



https://github.com/imgag/megSAP/blob/master/doc/performance.md

Structural Variant Calling srGS vs lrGS

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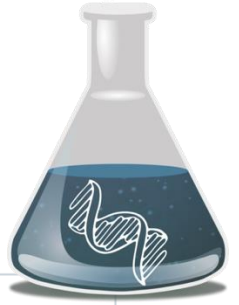
All structural variant benchmarks are done on the GIAB reference sample NA24385/HG002 using the draft SV benchmark v1.1. The analyses were performed with the short-read and long-read single sample pipelines.

Sensitivity and positive predictive value (PPV) were measured using Hap-Eval.



Test	coverage	sensitivity	PPV
short-read WGS - Manta 1.6.0	39.5x	36.30%	96.53%
short-read WGS - DRAGEN 4.2.4	39.5x	50.26%	97.64%
long-read WGS (high accuracy) - Sniffles 2.4	40.5x	90.59%	98.03%
long-read WGS (super accuracy) - Sniffles 2.4	40.5x	91.02%	98.05%

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The Advantage of Long Reads for Complex Clinical Use Cases

ELRIN* - European Long-Read Initiative for RD

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- Form a European network of experts developing Nanopore standards for clinical testing
- Explore the potential of Nanopore sequencing in Rare Diseases and Familial Cancer
- Gather deep control data Structural Variants and Methylation analyses
- Standardize reporting of repeat expansions, duplicate genes, mobile elements, SVs and methylation disorders
- Provide long-read next-generation sequencing data to the European +1Million-Genomes initiative.

Coordinators: Olaf Rieß, Beate Kristmann, Tübingen

**ELRIN Study is supported by ONT*

lonGER Consortium Pilot Study

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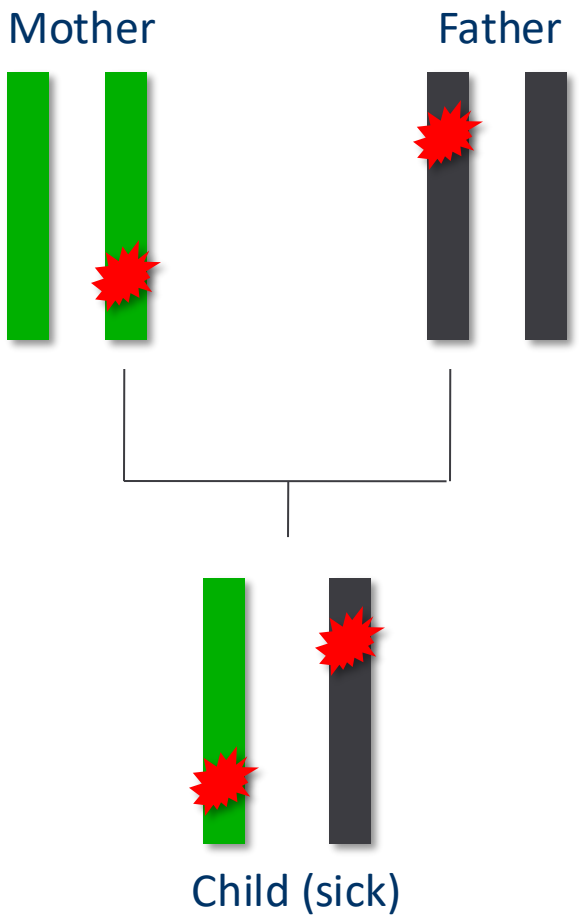
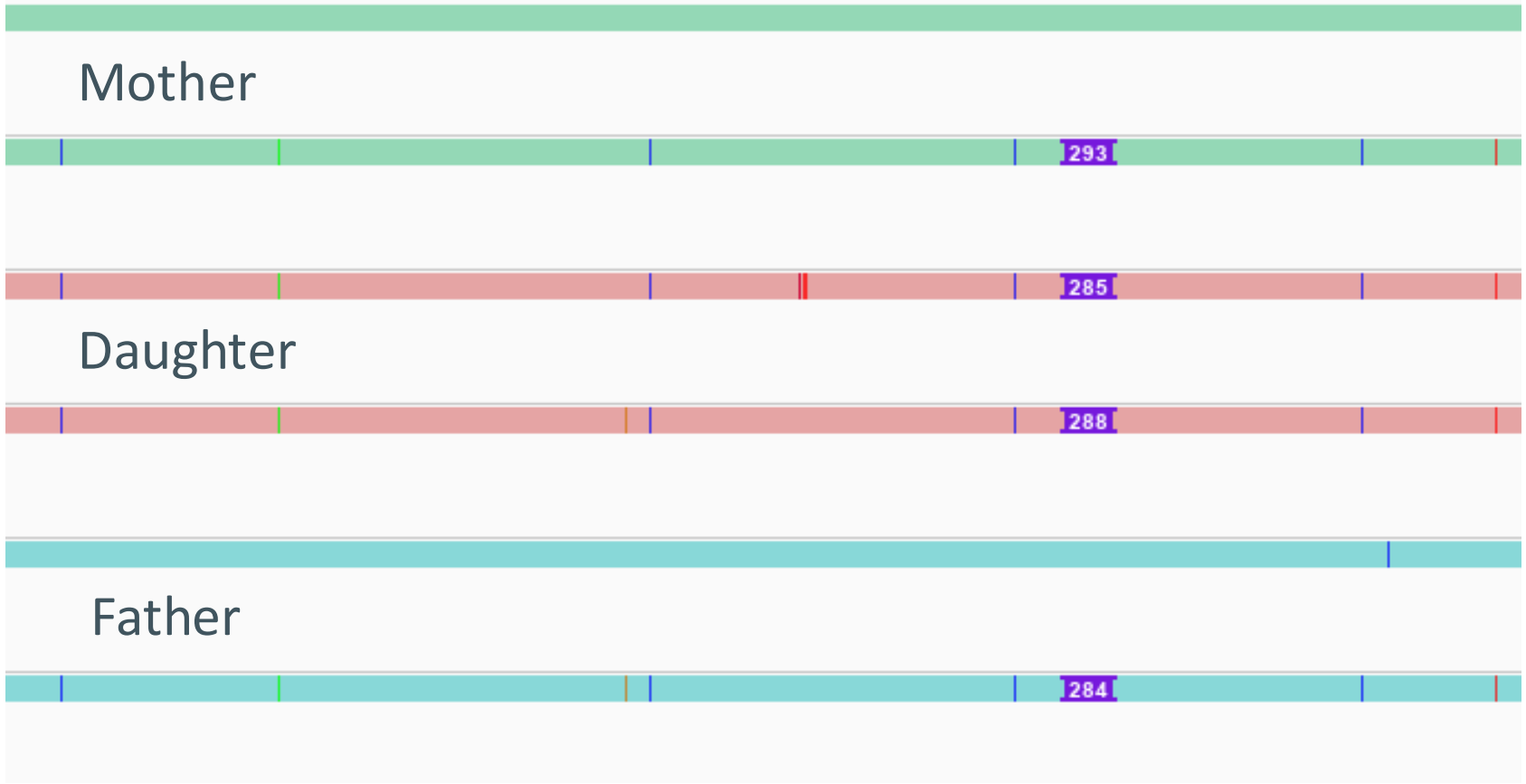
- Benchmarking **2 Genome in a Bottle** cell lines sequenced at each site
- **1000 RD patient** samples (1 FC per genome)
- **Clinical use cases**
  - Compound heterozygotes
  - Complex structural variants and mobile elements
  - Repeat expansions
  - Duplicated genes (genes with pseudogene copy)
  - Pathogenic haplotypes (e.g. OPN1-Cluster)
  - Methylation (imprinting disorders)

**lonGER – Clinical Long-read Genome Initiative is supported by ONT*

Haplotype-Phased Genomes – Compound Hets



Compound Heterozygotes can be resolved sequencing only the index case



Much Higher Sensitivity for SV Detection

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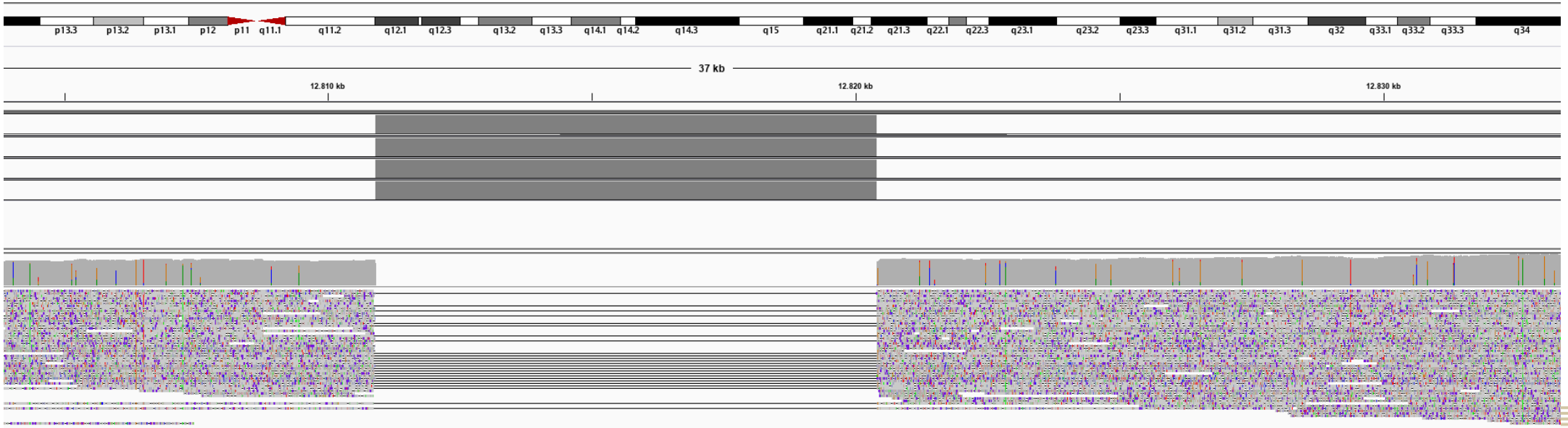
- Much higher sensitivity
- Accurate breakpoints
- True positive SVs per case:
  - Short reads: 10,000
  - Long reads: **23,000**

BioNano*

Nanopore



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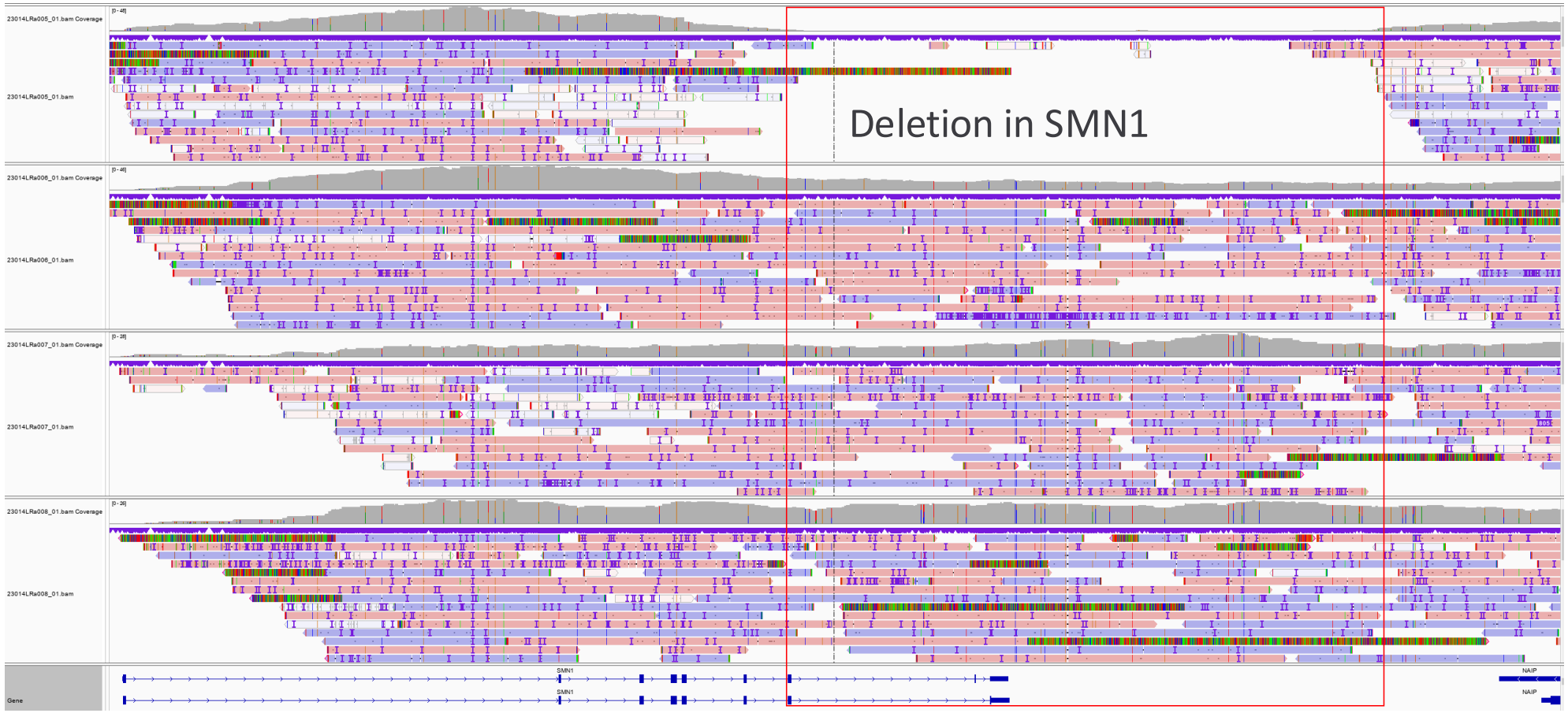
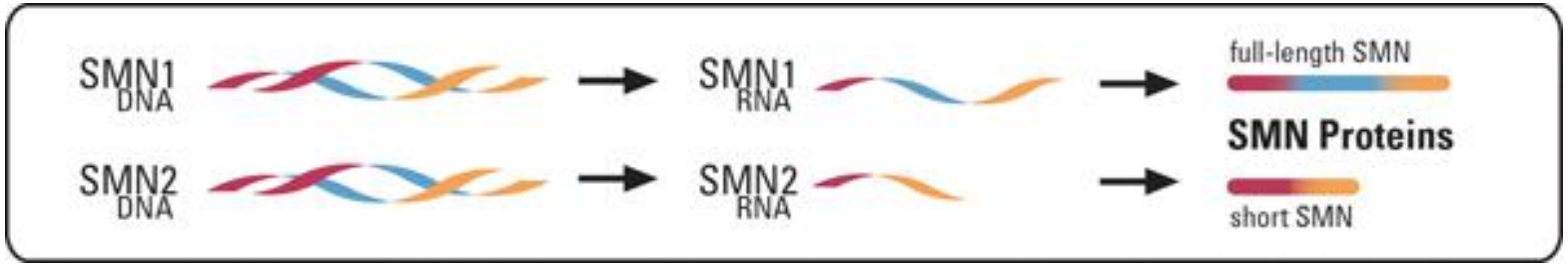
Duplicate Genes and Pathogenic Haplotypes

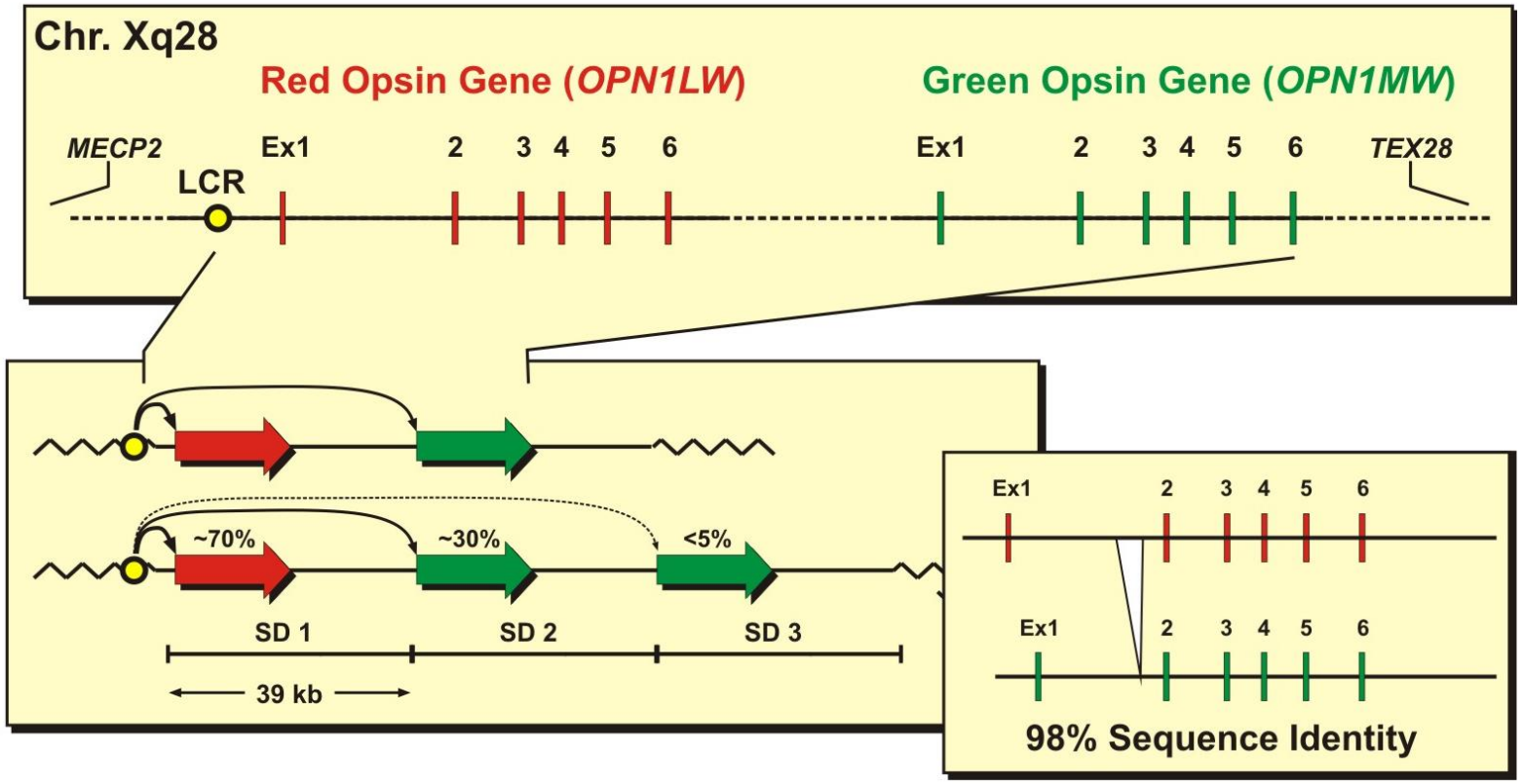
Duplicate Genes: SMN1 and SMN2 in Spinal Muscular Atrophy





Human *Opsin* Gene Cluster (Color Blindness)

- Genes OPN1LW and OPN1MW
- Copy no. variability (n=2-8)
- Expression gradient (only first two copies are relevant)
- High sequence conservation
- 5 pathogenic variants
- Very difficult for diagnostics



Collaborators: Caspar Gross, Elena Buena-Atienza, Bernd Wissinger

Human *Opsin* Gene Cluster (Color Blindness)

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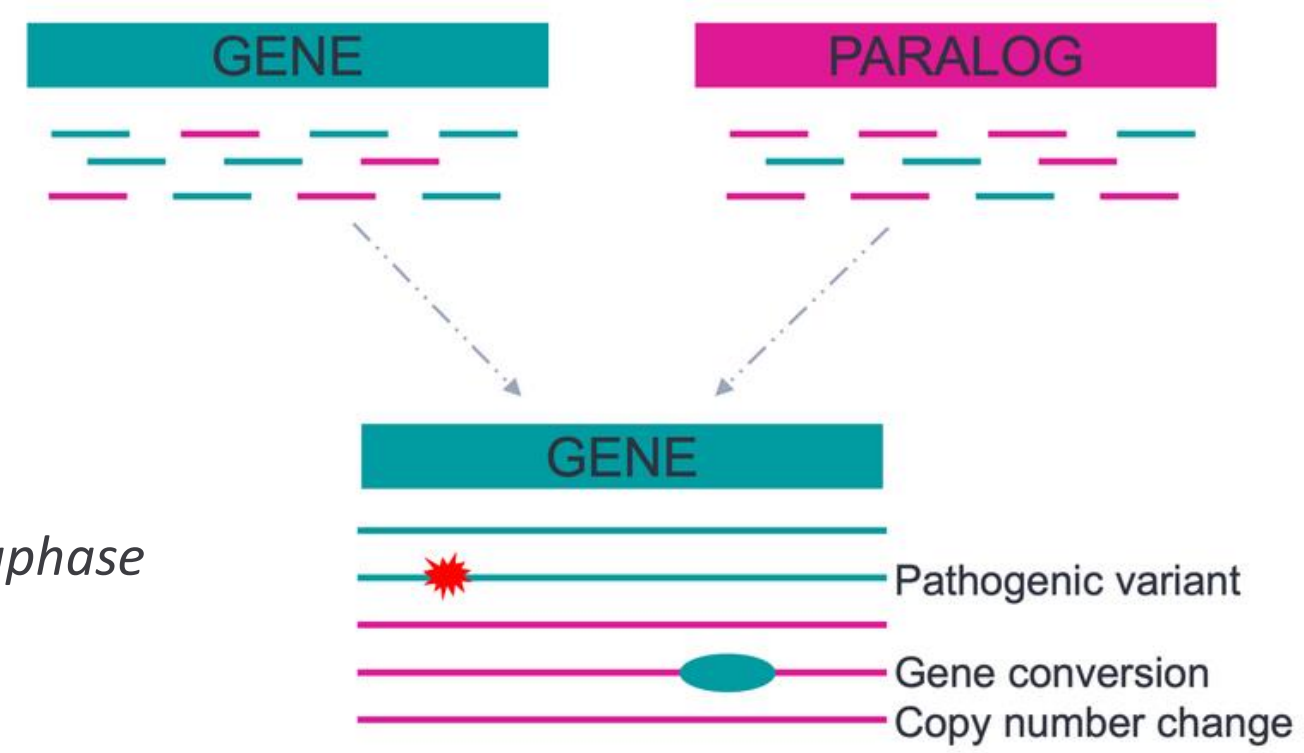
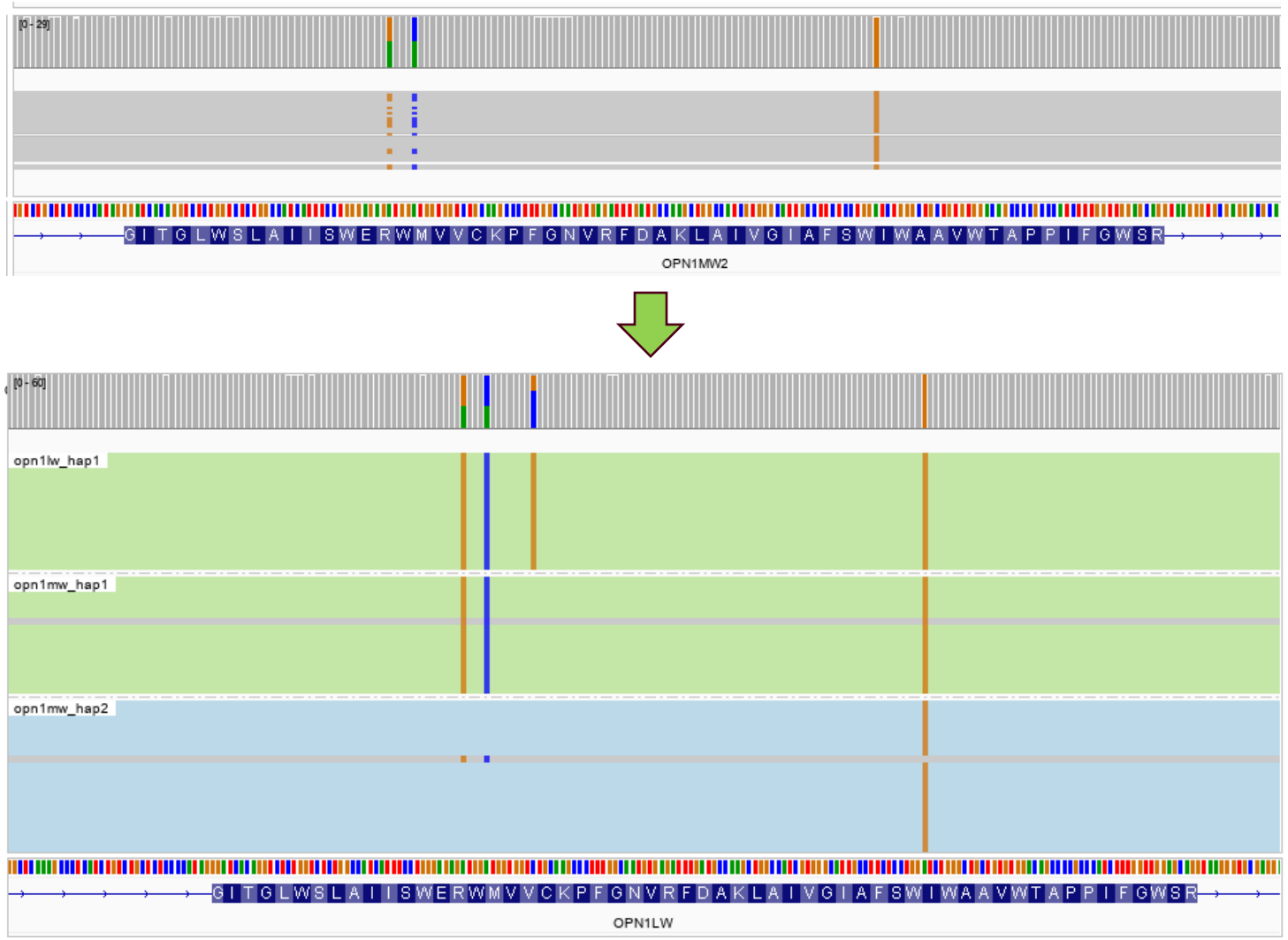
- Characterization of LW/MW gene variants not possible with srWGS
- Nanopore: evaluate copy number, order of copies, hetero-/hemizygous variants, haplotypes
- Diseases: e.g.
  - Blue Cone Monochromacy,
  - X-linked cone dysfunction disorders
  - TODO
- Paraphase tool for long reads:

  https://github.com/PacificBiosciences/paraphase



Haplotype Phasing: Determine Variants in 3 Gene Copies



Un-phased haplotypes

Phased haplotypes

*OPN1LW*_hap1:LVAVA$^{c.465G}$

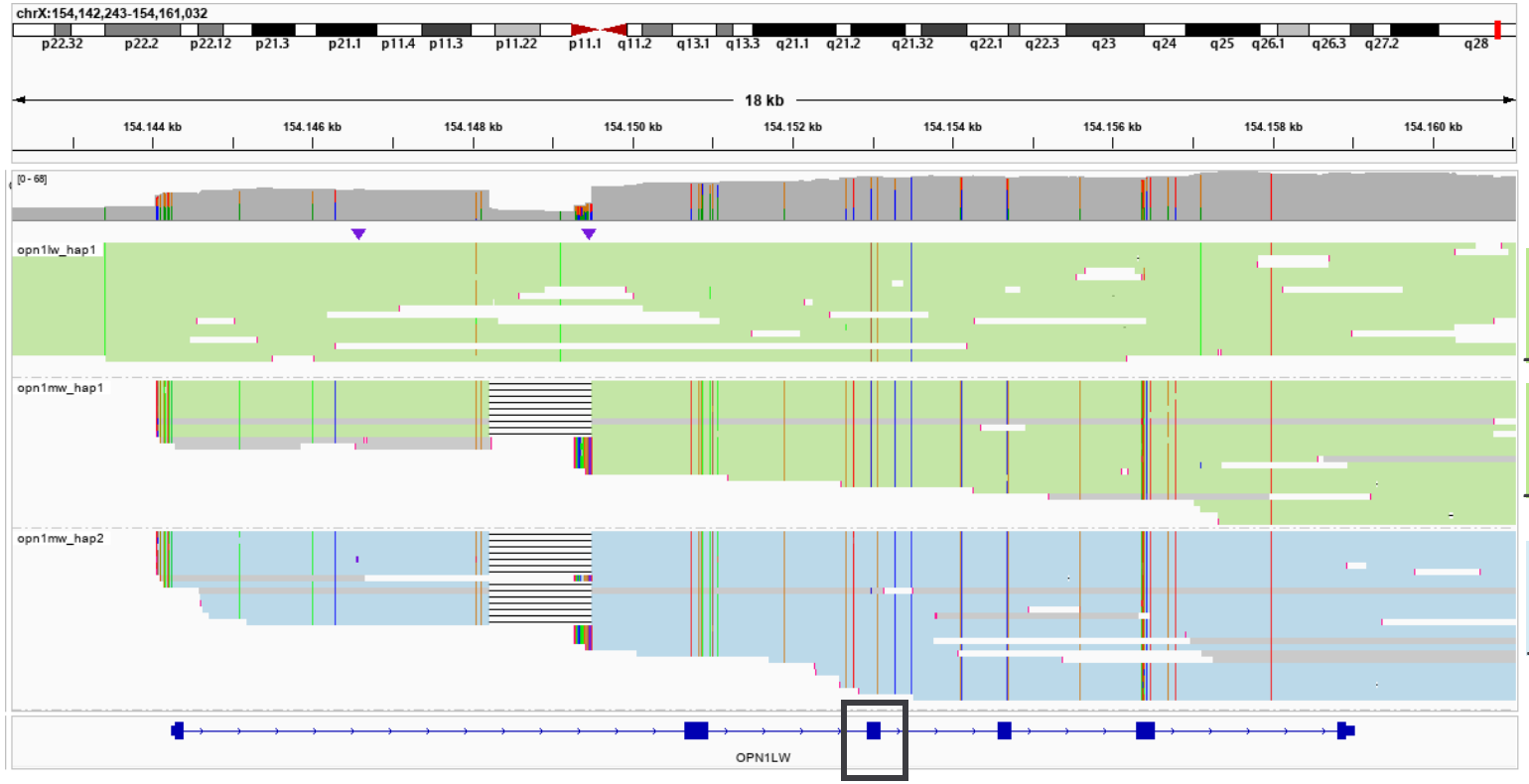
*OPN1MW*_hap1:LVAVA$^{c.465C}$

*OPN1MW*_hap2:MVAVA$^{c.465C}$

Collaborators: Caspar Gross, Elena Buena-Atienza, Bernd Wissinger

Diagnosis for Patient

Gender	N50 read length (kb)	Coverage WGS	Coverage *OPN1*	Copy Number	Haplotype 1	Haplotype 2	Haplotype 3	Phenotype	Mutation type
male	19,7	43	80	3	*OPN1**LW**_hap1* LVAVA	*OPN1**MW**_hap1* LVAVA	*OPN1**MW**_hap2* MVAVA	Blue Cone Monochromacy (BCM)	Exon 3 splicing-deficient Haplotypes



*OPN1**LW**_hap1* LVAVA

*OPN1**MW**_hap1* LVAVA

*OPN1**MW**_hap2* MVAVA

Exon 3

Why is important?

- 2 first copies expressed
- LVAVA and MVAVA have different pathogenicity (5% vs 50% correctly spliced transcripts)

(Buena-Atienza et al., 2016; Neitz et al., 2021)

Collaborators: Caspar Gross, Elena Buena-Atienza, Bernd Wissinger

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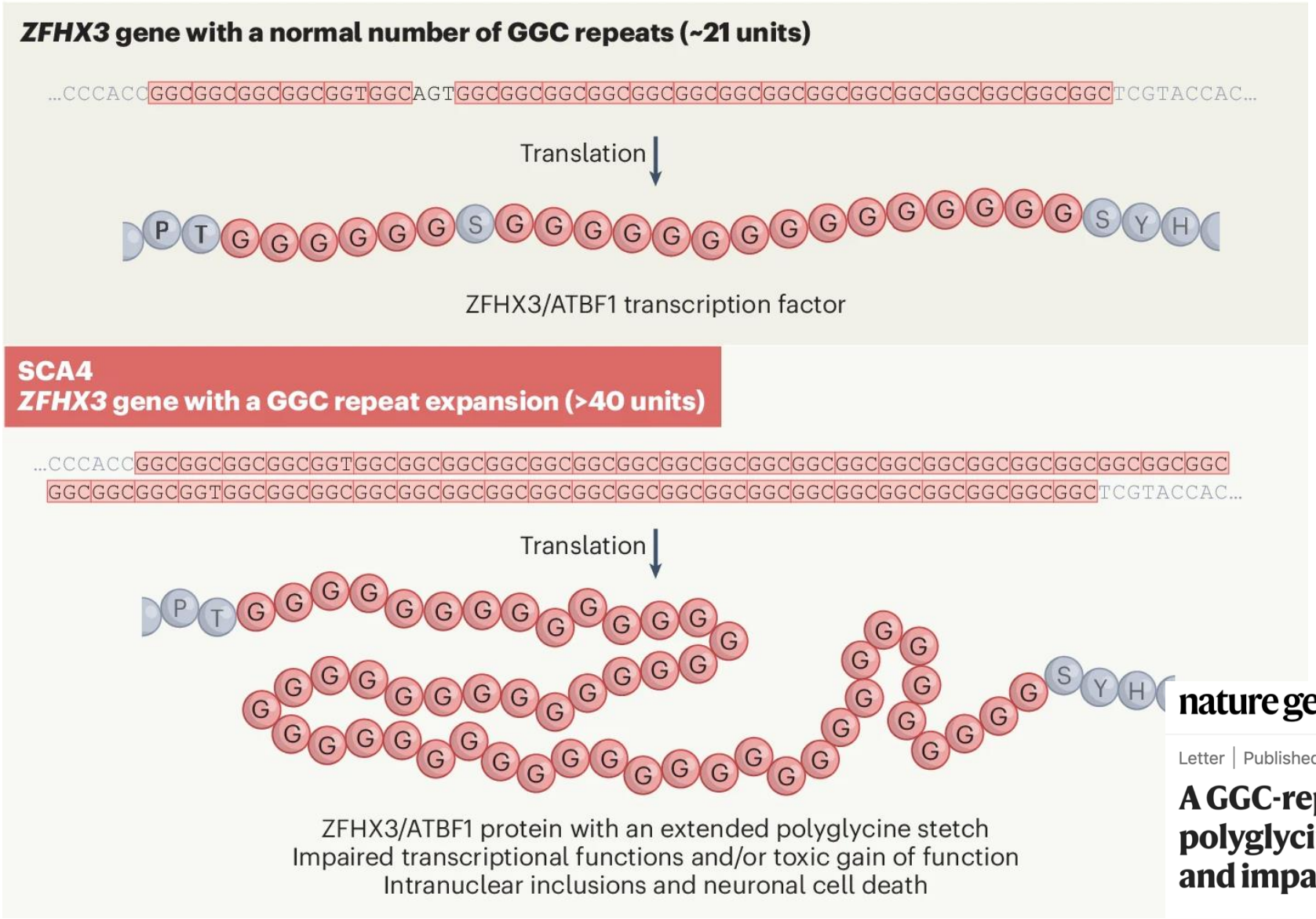
Repeat Expansion Diseases

Repeat Expansions: Known and Novel Pathogenic Expansions



Genome-wide:
82 known pathogenic repeat expansions

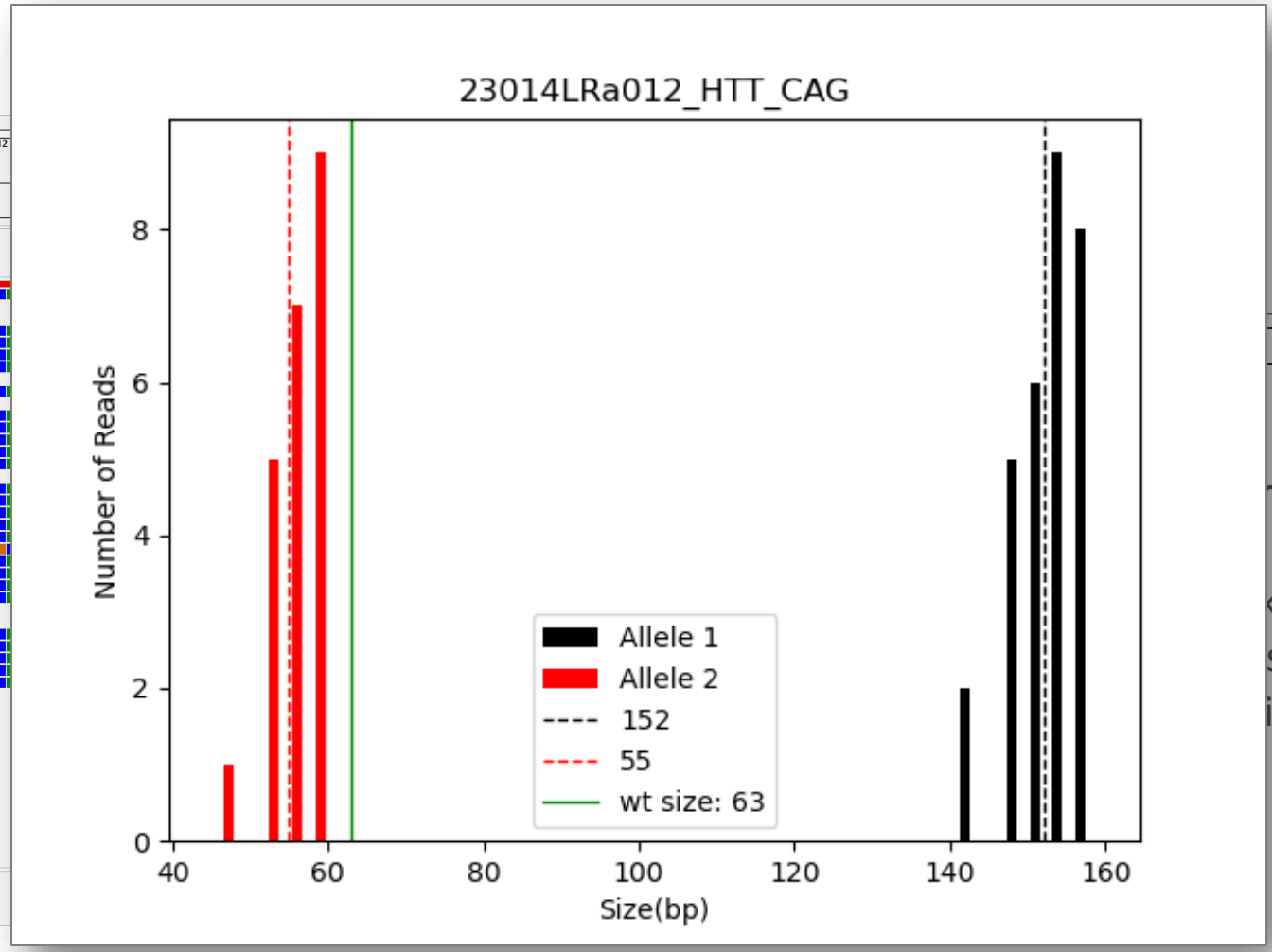
nature genetics

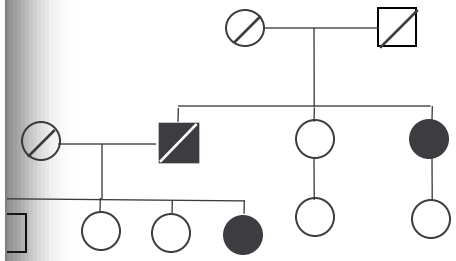
Letter | Published: 29 April 2024

A GGC-repeat expansion in *ZFHX3* encoding polyglycine causes spinocerebellar ataxia type 4 and impairs autophagy

Karla P. Figueroa, Caspar Gross, Elena Buena-Atienza, Sharan Paul, Mandi Gandelman,

Example: Huntington Disease (Tübingen Case 15)





n: Huntington disease

kinetic movement disorder
sturbance
ine motor coordination

Extended allele: 53 CAG Repeats in HTT

Example: Ataxia (SCA3, Tübingen Case 16)

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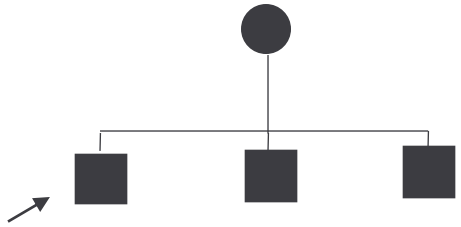
Indication: SCA 3

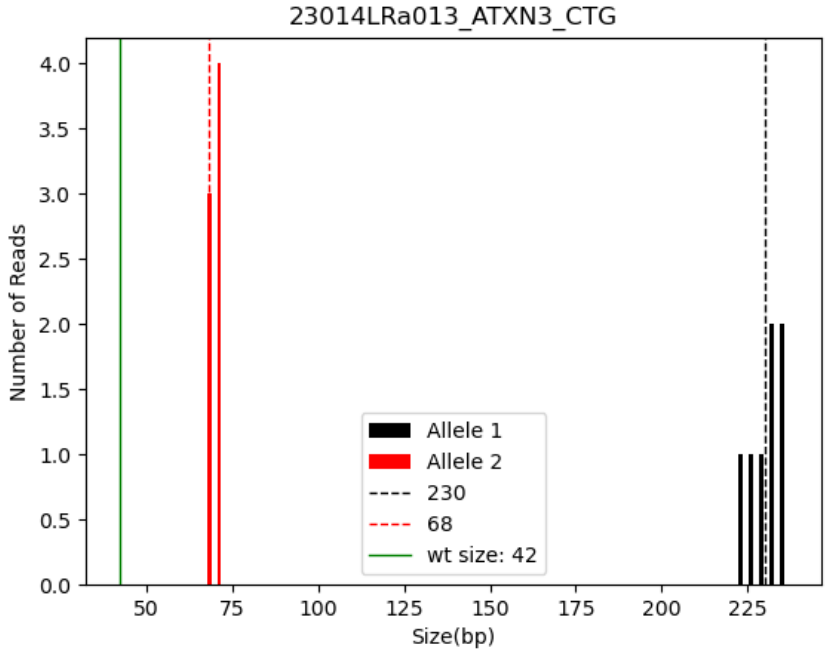
- ataxia
- dysarthria
- dysphagia
- cognitive impairment
- ophthalmoparesis
- parkinsonism



Extended allele: 77 CAG Repeats in ATXN3

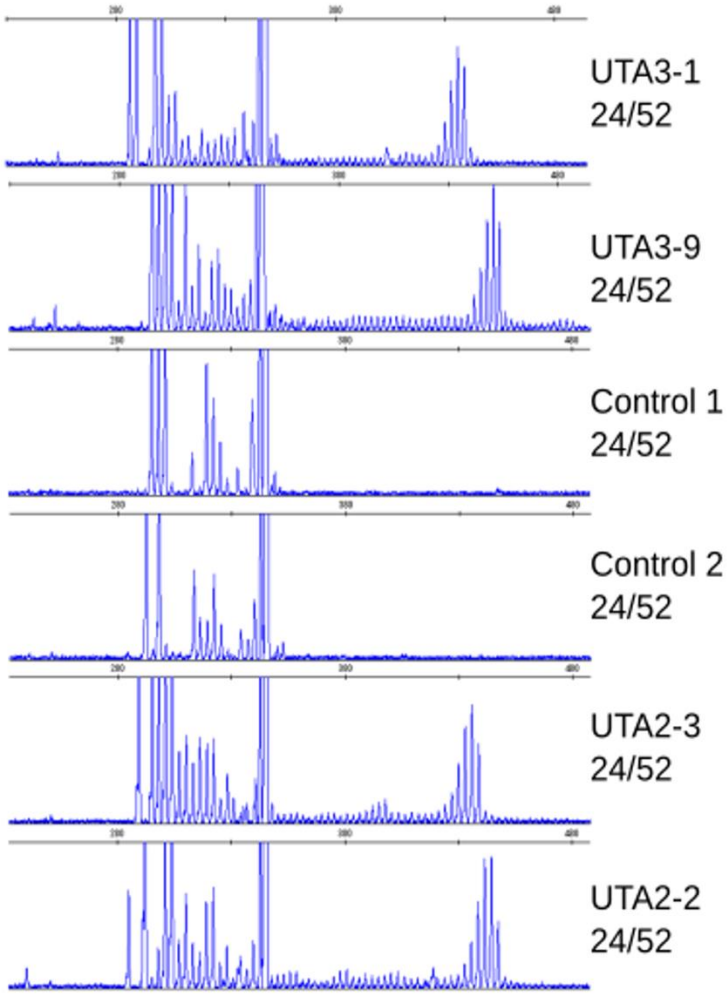
ONT Long Read Analysis

- ATNX3: $(CAG)_{23}$ / $(CAG)_{77}$

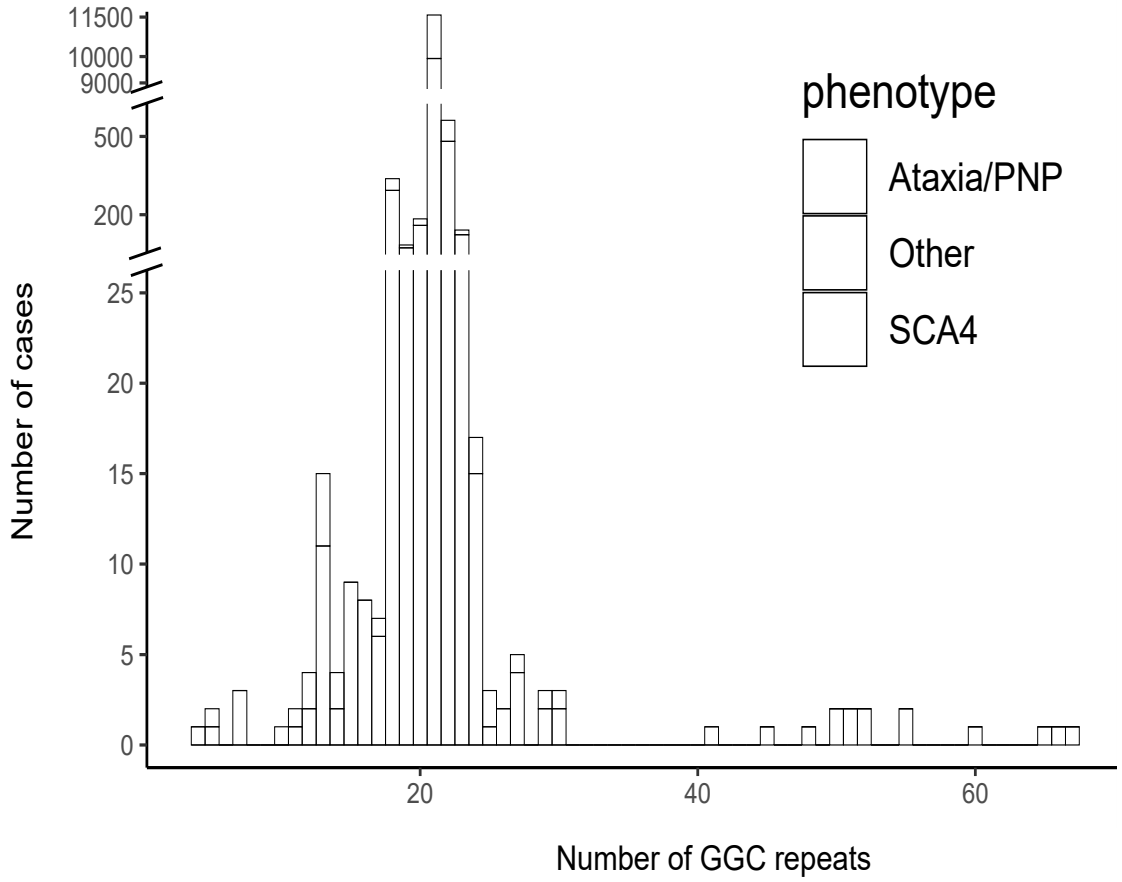


SCA4: Novel Pathogenic Repeat Expansion in ZFHX3 Discovered

Validation (PCR amplification)



Pathogenic repeat length



Collaboration with K. Figueroa, S. Pulst, U. Utha; M. Spielmann, U. Kiel

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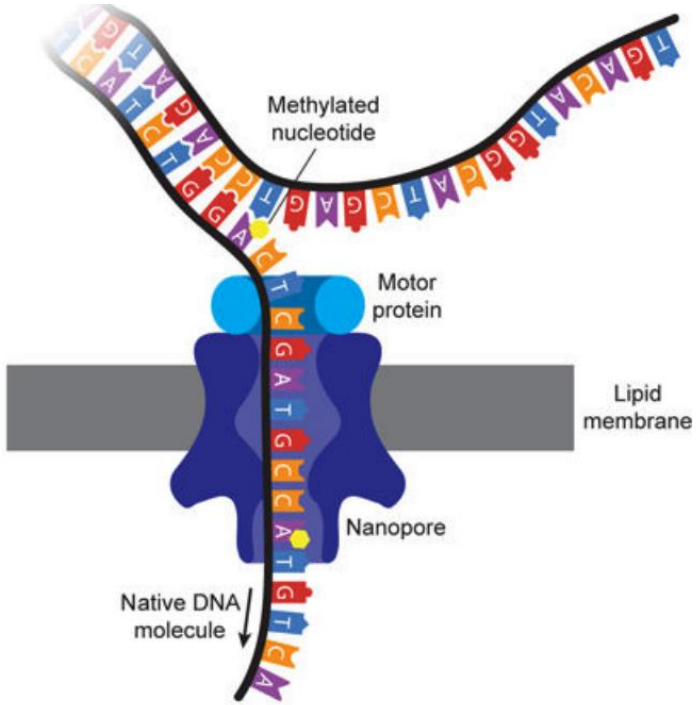
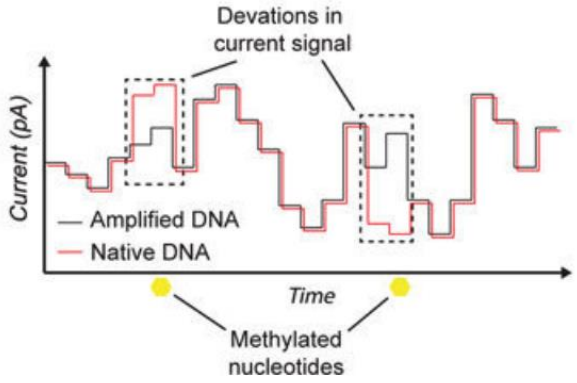
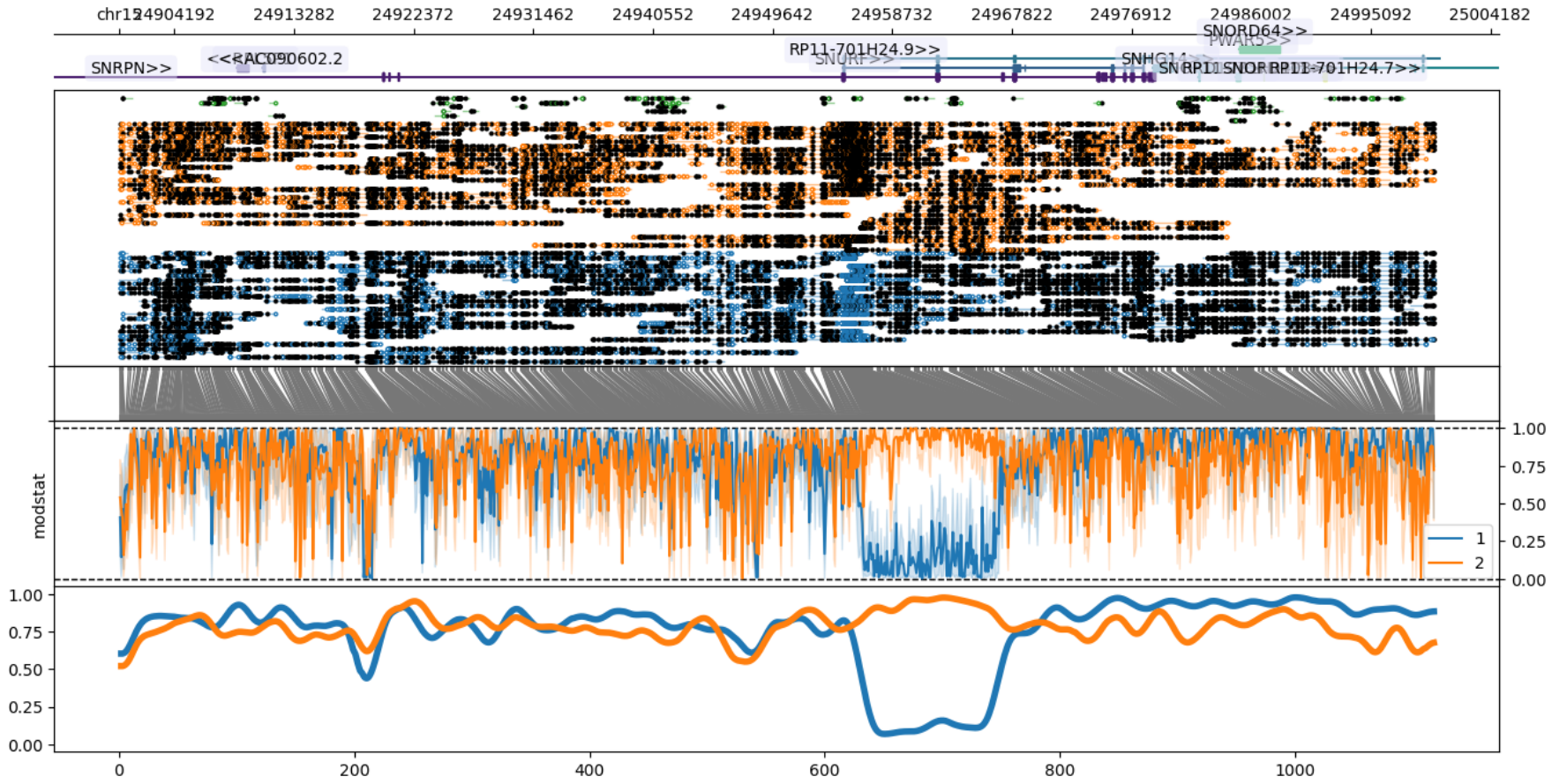
Methylation: Imprinting Diseases

Haplotype-Phased DNA Methylation Calling





Plot: MethylArtist

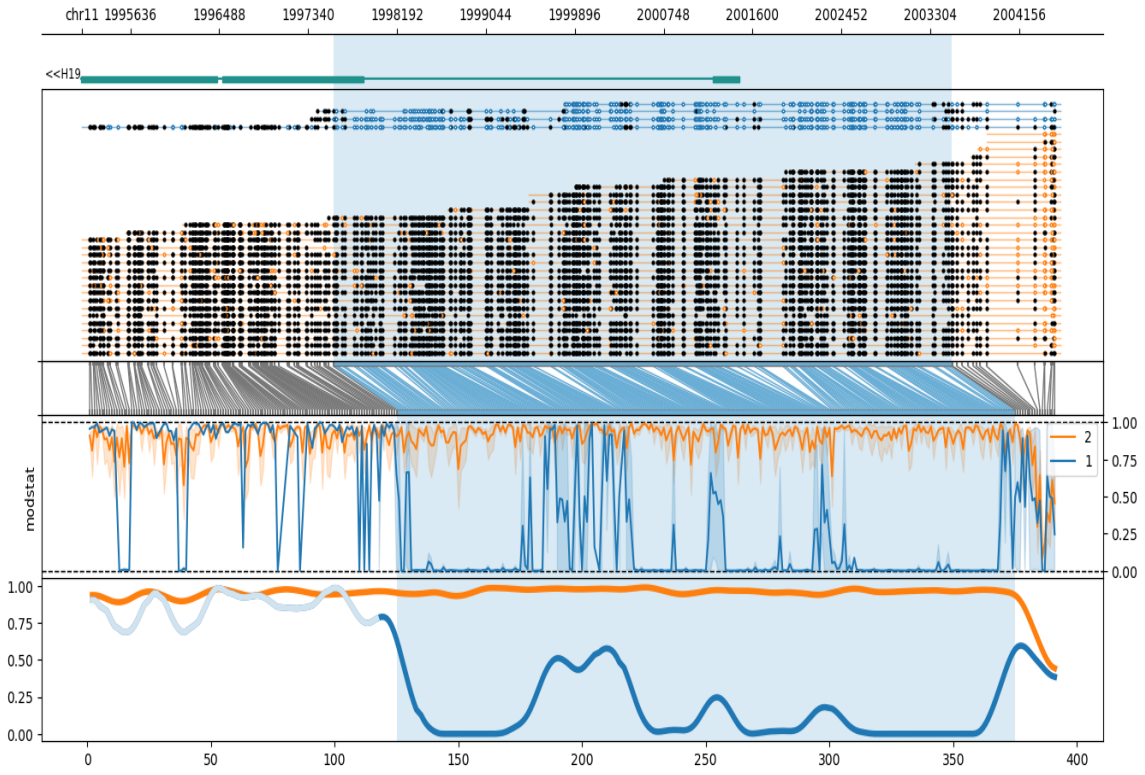


Imprinting Locus

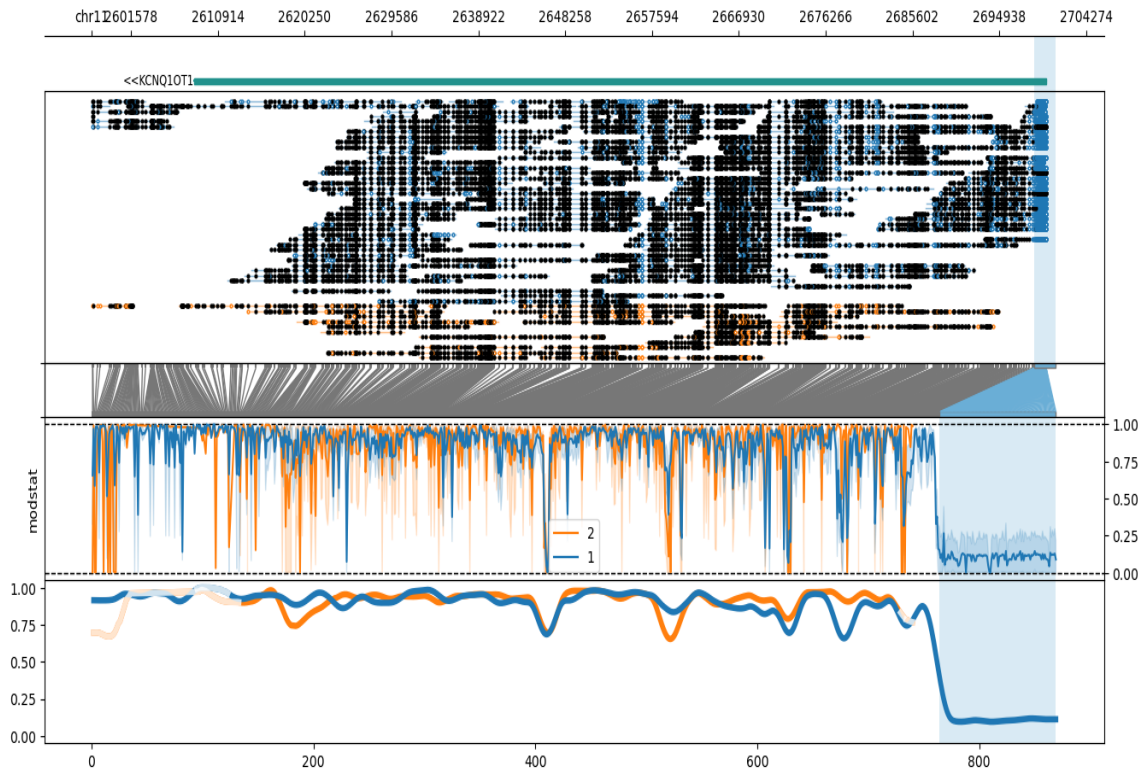
Imprinting Defects: Detecting Anomalies in Haplotype-Phased Methylation

- Beckwith-Wiedemann syndrome (BWS) caused by Uniparental Disomy 11 (UPD11)
- Genomic imprinting at genes H19 and KCNQ1OT1 – only one parental allele should be expressed

H19 – Gain of methylation



KCNQ1OT1 – Loss of methylation

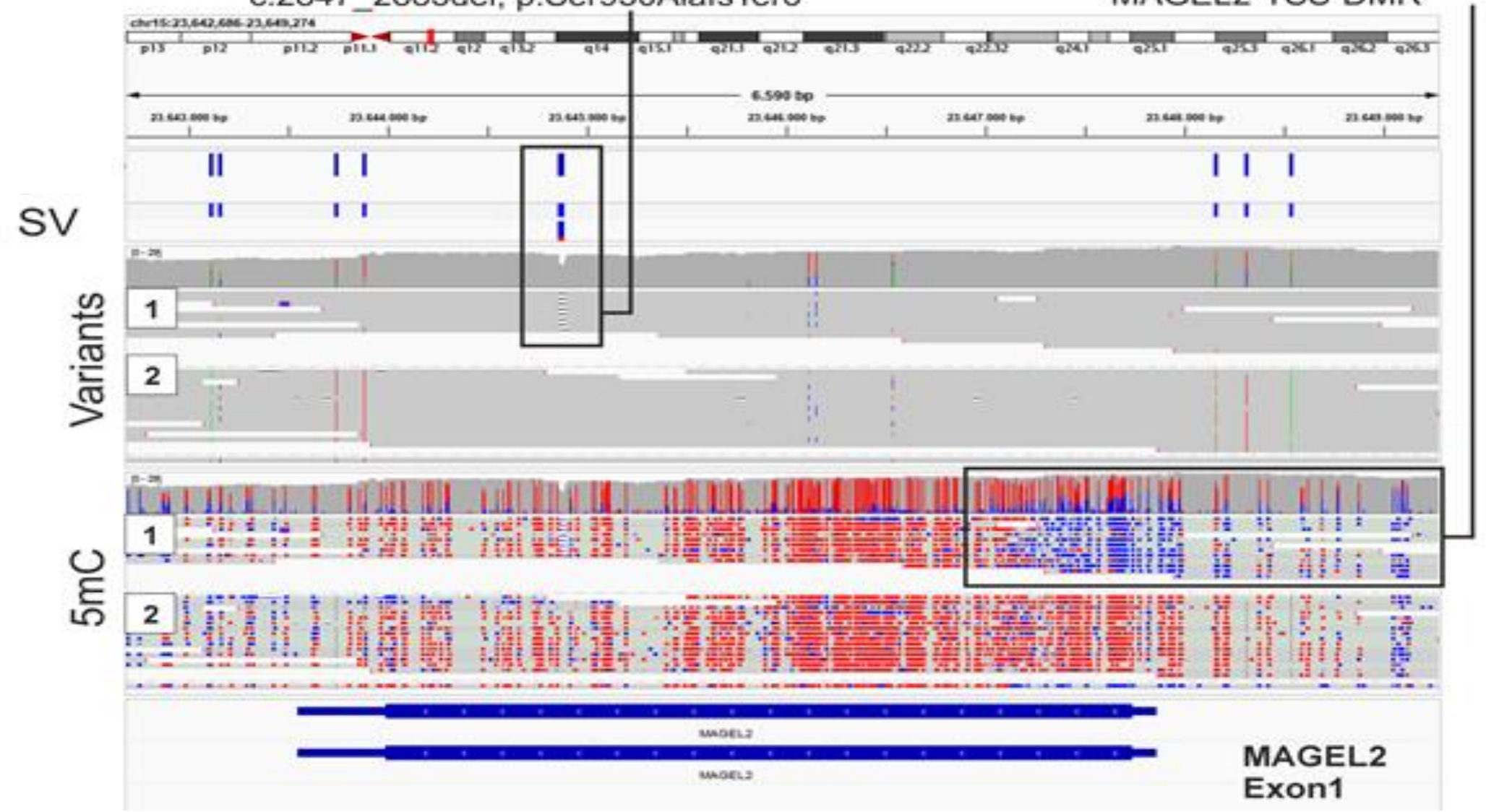


Collaboration with F. Kraft, I. Kurth, T. Eggermann, UK Aachen

Compound Het: Methylation + Frameshift Indel

Schaaf-Yang Syndrome: **MAGEL2 Exon 1**
c.2847_2883del; p.Ser950AlafsTer6

Hypomethylated allele
MAGEL2-TSS-DMR



Fragile X Syndrome: FMR1 Inactivation by Methylation

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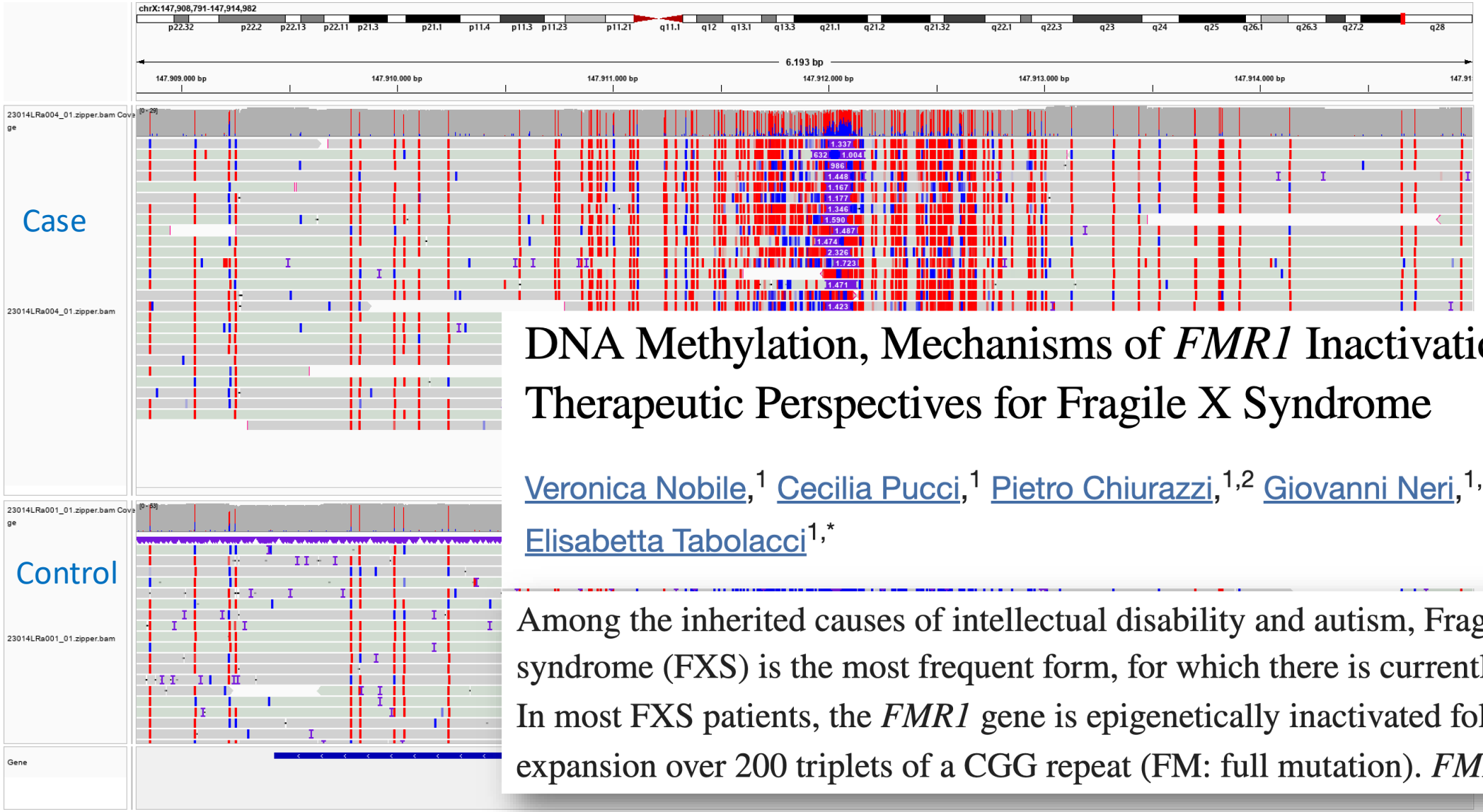
CGG expansion & Hypermethylation in FMR1 (X chromosome, male patient),

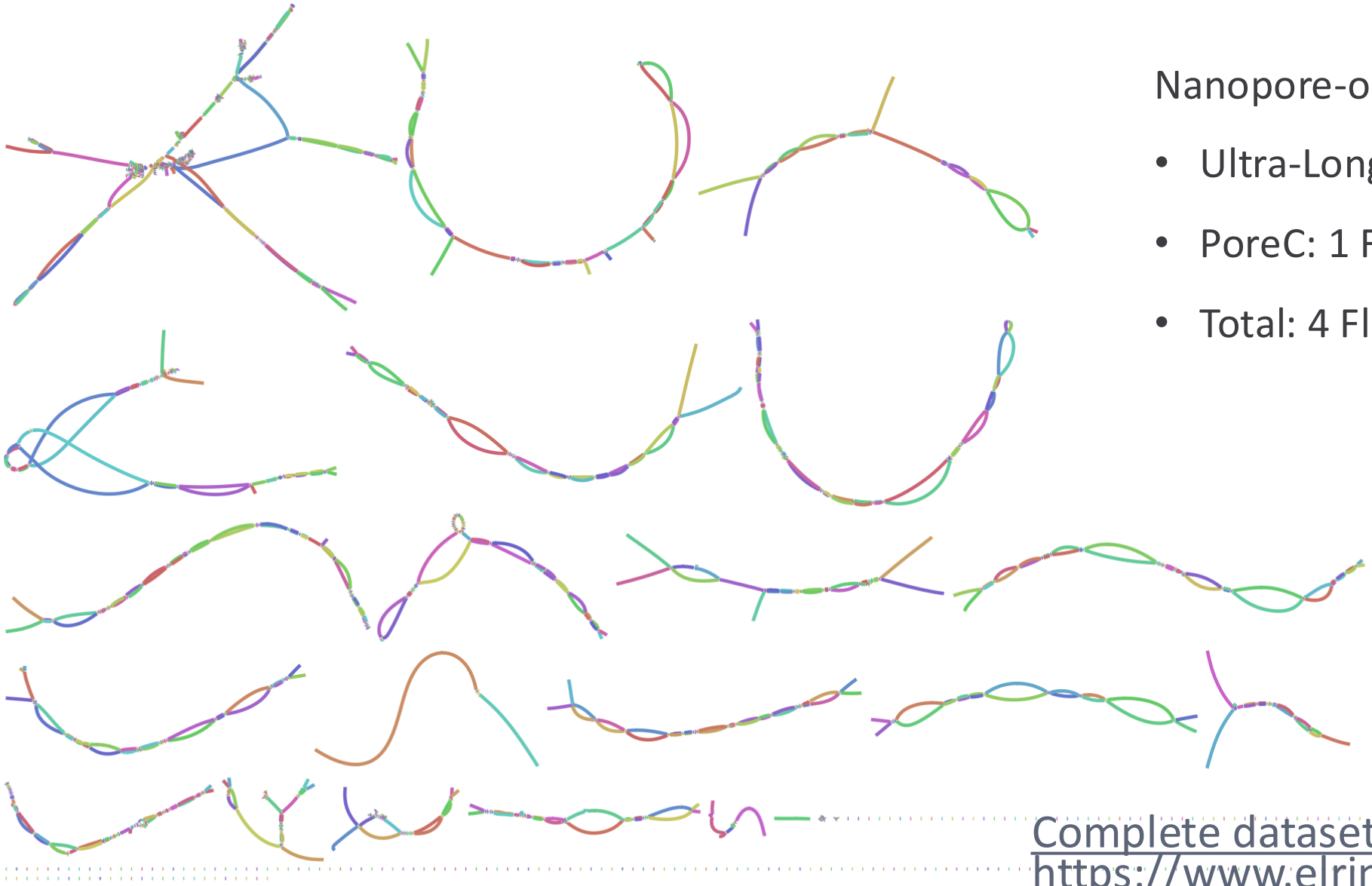


DNA Methylation, Mechanisms of *FMR1* Inactivation and Therapeutic Perspectives for Fragile X Syndrome

Veronica Nobile,[1] Cecilia Pucci,[1] Pietro Chiurazzi,[1,2] Giovanni Neri,[1,3] and Elisabetta Tabolacci[1,*]

Among the inherited causes of intellectual disability and autism, Fragile X syndrome (FXS) is the most frequent form, for which there is currently no cure. In most FXS patients, the *FMR1* gene is epigenetically inactivated following the expansion over 200 triplets of a CGG repeat (FM: full mutation). *FMR1* encodes

Outlook: Telomere-Telomere Genomes



Nanopore-only T2T with:

- Ultra-Long Reads of 100kb: 3 FC
- PoreC: 1 FC
- Total: 4 Flowcells

Complete dataset:
https://www.elrin-network.eu/

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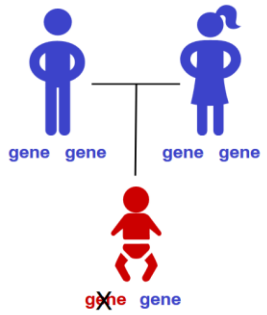
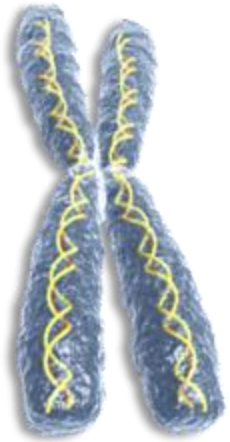
AI-assisted Diagnostics

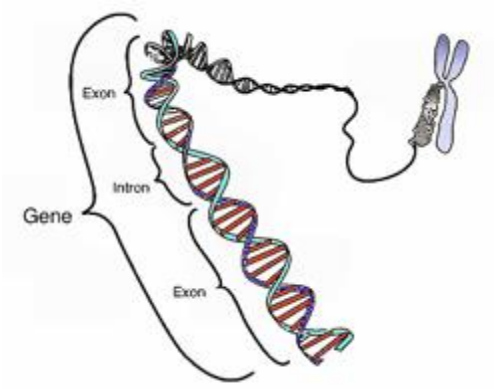
The Problem: Finding the Causal Variant in Genetic Disease

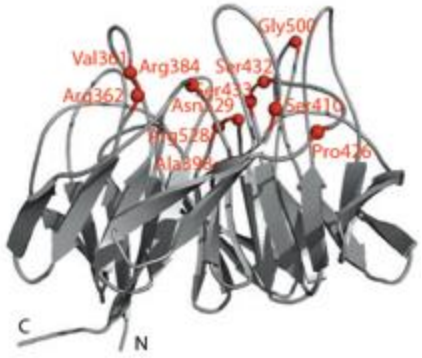


Pathogenicity Classification → Causal Variant Prioritization

Genome: ~4M SNVs → **Exome:** ~80k SNVs → **Alters Protein:** ~15k SNVs → **Causal variant:** 1 SNV



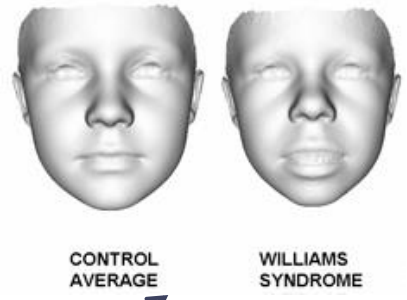




Annotation Features for AI Diagnostics

Population AF

Phenotype (e.g. HPO, Images, EHR)



Evolutionary Conservation

```
sample  EPPLSQETFS DLWNLLPENN
spec 1  DLPESQGSFQ ELWETVSYPP
spec 2  ELPLSQETFS GLWKLLPPED
spec 3  ELPLSQEFTS CLWKLLPPDD
spec 4  EPPDSQE-FA ELWNLIVRDN
```

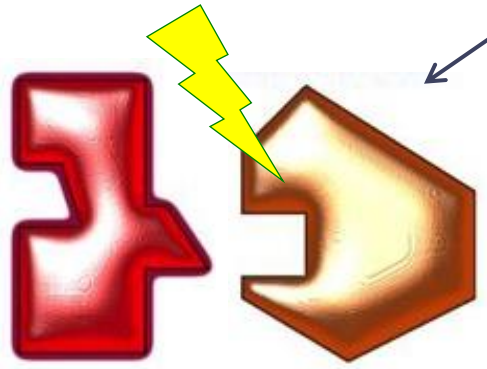
SNV

SNV

InDel

SNV

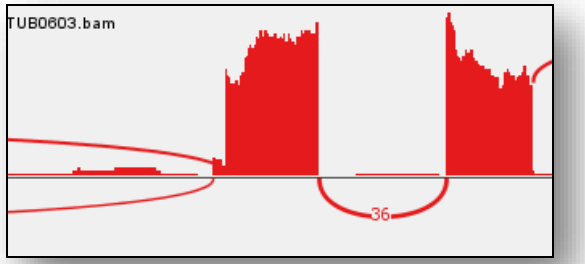
Structural Variant



Damage Scores

Disease Variants DBs

Splicing defects & alternative splicing



RNA-seq

Automated Diagnostics by AI: aiDIVA ensemble-AI

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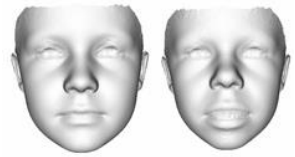
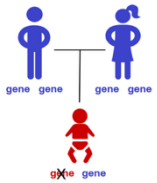
Trained on > 100,000 clinical cases
Evaluated on > 3,000 clinical cases

Variants

Annotation



Patient Information



Random-forest model

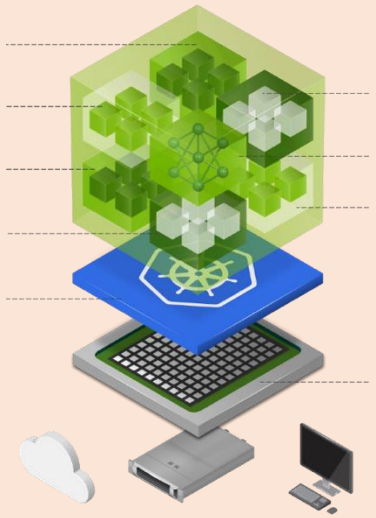
Pathogenicity Classification

Evidence-based model

dominant

recessive

Large Language Models



On NVIDIA NIMs

Meta-AI

Clinical Report

Improving Causal Variant Prioritization with Large Language Models (LLMs)

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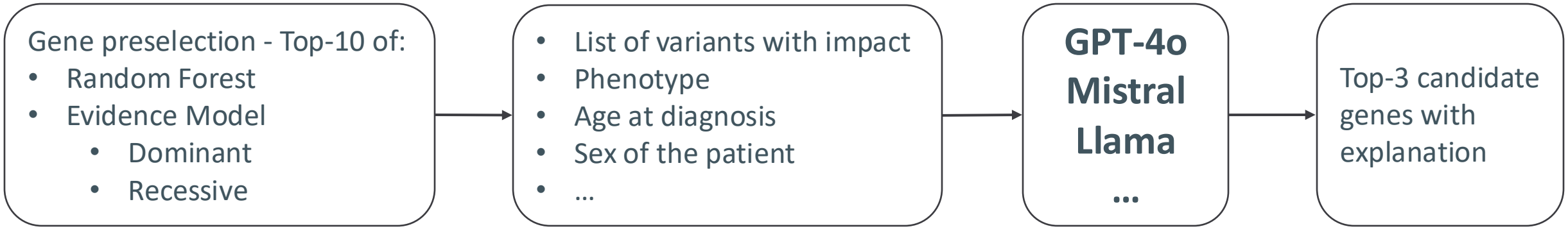

Example Prompt:

"A female rare disease patient of age 0 has the following symptoms: Facial hemangioma,Status epilepticus. A causal variant in which of the following candidate genes would best explain these symptoms? Candidate genes: PIEZO1,SEC23B,EFTUD2,CDH11,AASS,KIAA0753,SCN1A,ABCC8,VPS51, NDUFA9". Please provide sources.

Example LLM “ Diagnosis”

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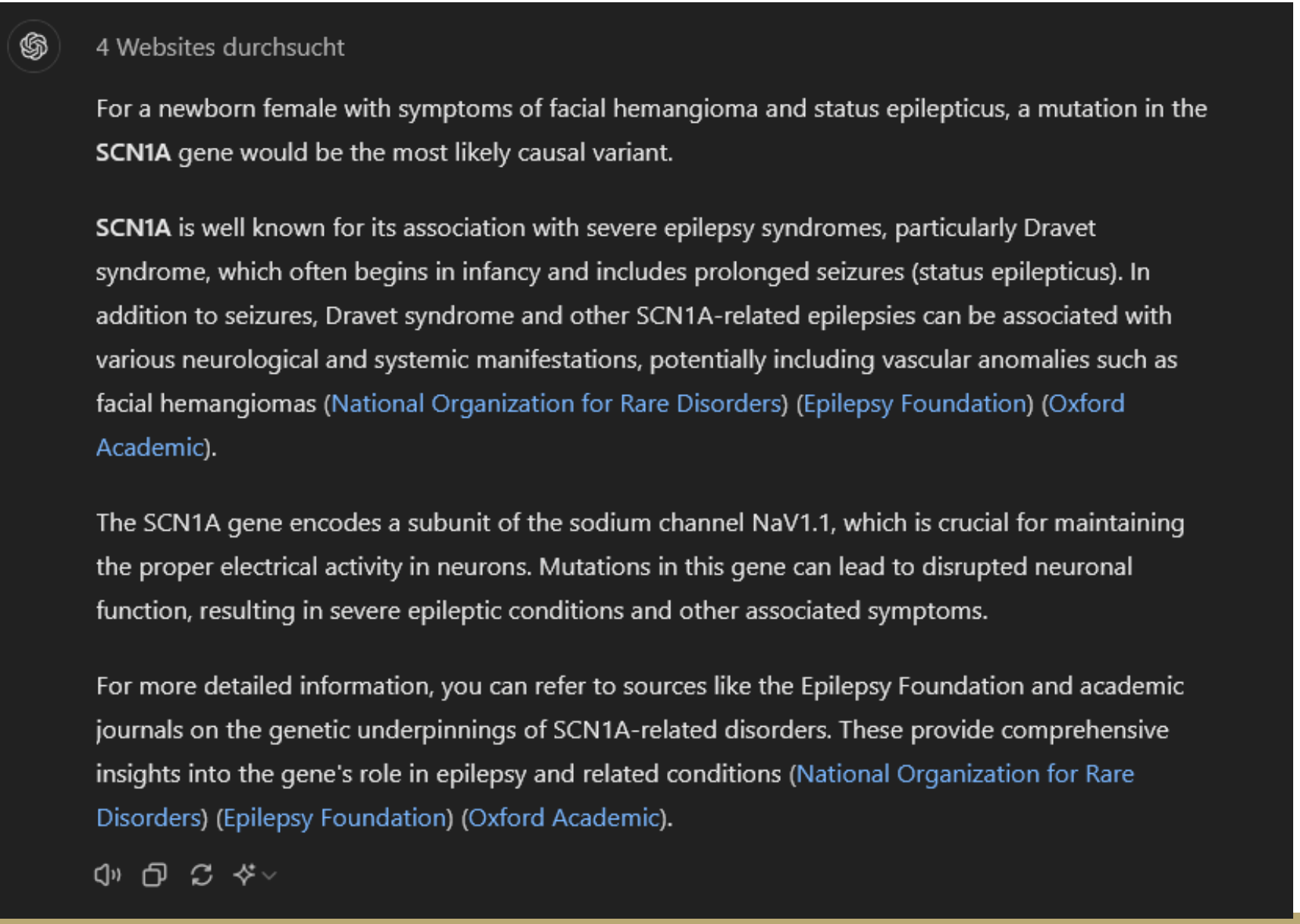


4 Websites durchsucht

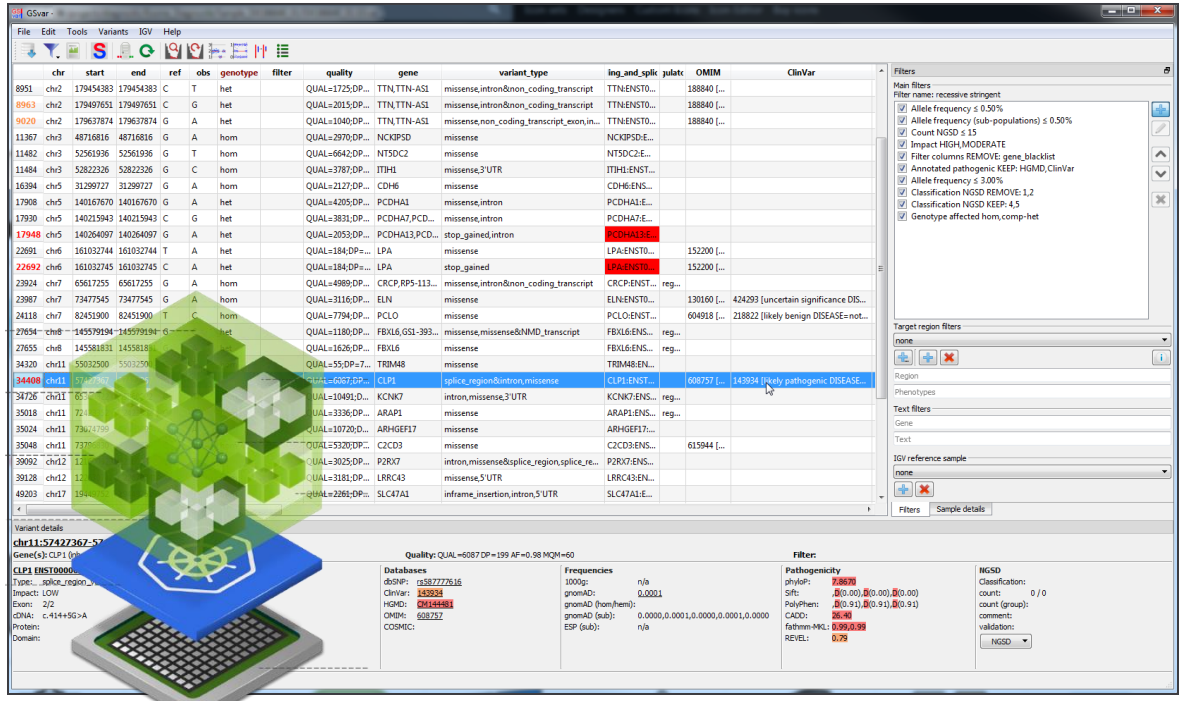
For a newborn female with symptoms of facial hemangioma and status epilepticus, a mutation in the **SCN1A** gene would be the most likely causal variant.

SCN1A is well known for its association with severe epilepsy syndromes, particularly Dravet syndrome, which often begins in infancy and includes prolonged seizures (status epilepticus). In addition to seizures, Dravet syndrome and other SCN1A-related epilepsies can be associated with various neurological and systemic manifestations, potentially including vascular anomalies such as facial hemangiomas (National Organization for Rare Disorders) (Epilepsy Foundation) (Oxford Academic).

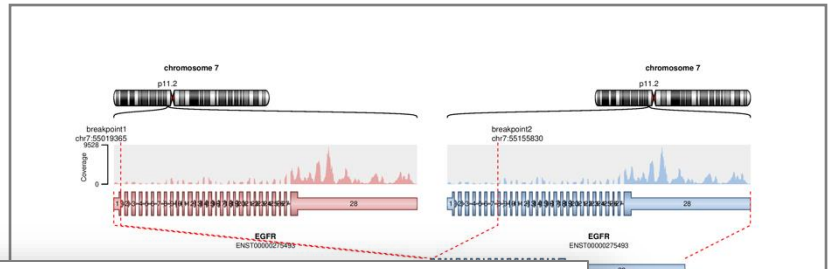
The SCN1A gene encodes a subunit of the sodium channel NaV1.1, which is crucial for maintaining the proper electrical activity in neurons. Mutations in this gene can lead to disrupted neuronal function, resulting in severe epileptic conditions and other associated symptoms.

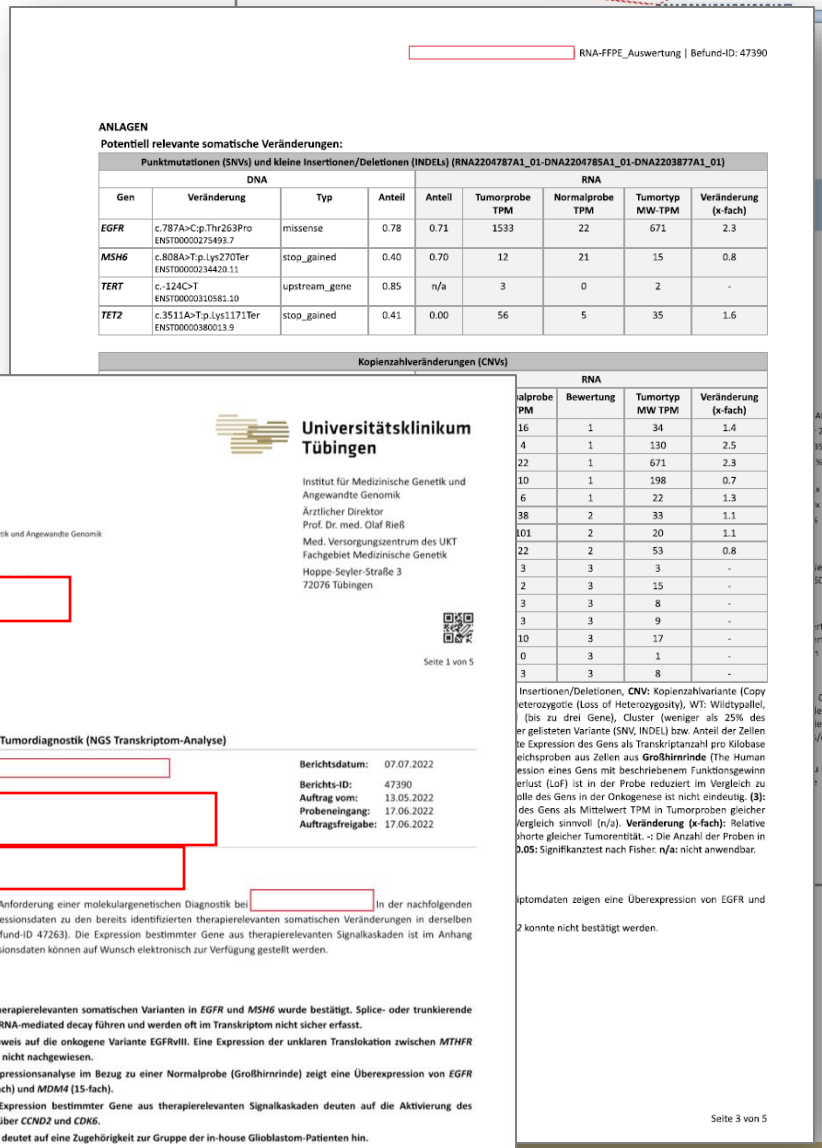
For more detailed information, you can refer to sources like the Epilepsy Foundation and academic journals on the genetic underpinnings of SCN1A-related disorders. These provide comprehensive insights into the gene's role in epilepsy and related conditions (National Organization for Rare Disorders) (Epilepsy Foundation) (Oxford Academic).

Automatically Generating Clinical Reports



AI-generated reports





RNA-FFPE_Auswertung | Befund-ID: 47390

ANLAGEN

Potentiell relevante somatische Veränderungen:

Punktmutationen (SNVs) und kleine Insertionen/Deletionen (INDELs) (RNA2204787A1_01-DNA2204785A1_01-DNA2203877A1_01)								
DNA				RNA				
Gen	Veränderung	Typ	Anteil	Anteil	Tumorprobe TPM	Normalprobe TPM	Tumortyp MW-TPM	Veränderung (x-fach)
EGFR	c.787A>C:p.Thr263Pro ENST00000275493.7	missense	0.78	0.71	1533	22	671	2.3
MSH6	c.808A>T:p.Lys270Ter ENST00000234420.11	stop_gained	0.40	0.70	12	21	15	0.8
TERT	c.-124C>T ENST00000310581.10	upstream_gene	0.85	n/a	3	0	2	-
TET2	c.3511A>T:p.Lys1171Ter ENST00000380013.9	stop_gained	0.41	0.00	56	5	35	1.6

Kopienzahlveränderungen (CNVs)								
					RNA			
					alprobe PM	Bewertung	Tumortyp MW TPM	Veränderung (x-fach)
					16	1	34	1.4
					4	1	130	2.5
					22	1	671	2.3
					10	1	198	0.7
					6	1	22	1.3
					38	2	33	1.1
					101	2	20	1.1
					22	2	53	0.8
					3	3	3	-
					2	3	15	-
					3	3	8	-
					3	3	9	-
					10	3	17	-
					0	3	1	-
					3	3	8	-

Insertionen/Deletionen, **CNV:** Kopienzahlvariante (Copy
leterozygotie (Loss of Heterozygosity), WT: Wildtypallel,
(bis zu drei Gene), Cluster (weniger als 25% des
er gelisteten Variante (SNV, INDEL) bzw. Anteil der Zellen
te Expression des Gens als Transkriptanzahl pro Kilobase
eichsproben aus Zellen aus **Großhirnrinde** (The Human
ession eines Gens mit beschriebenem Funktionsgewinn
erlust (LoF) ist in der Probe reduziert im Vergleich zu
olle des Gens in der Onkogenese ist nicht eindeutig. **(3):**
des Gens als Mittelwert TPM in Tumorproben gleicher
Vergleich sinnvoll (n/a). **Veränderung (x-fach):** Relative
horte gleicher Tumorentität. **-:** Die Anzahl der Proben in
0.05: Signifikanztest nach Fisher. **n/a:** nicht anwendbar.

iptomdaten zeigen eine Überexpression von EGFR und

2 konnte nicht bestätigt werden.

Seite 3 von 5

Universitätsklinikum Tübingen

Institut für Medizinische Genetik und Angewandte Genomik
Ärztlicher Direktor
Prof. Dr. med. Olaf Rieß
Med. Versorgungszentrum des UKT
Fachgebiet Medizinische Genetik
Hoppe-Seyler-Straße 3
72076 Tübingen

MVZ/Institut für Medizinische Genetik und Angewandte Genomik
Calwerstraße 7 • 72076 Tübingen

Frau

72076 Tübingen

Seite 1 von 5

Nachrichtlich an Verteiler ZPM

Bericht zur somatischen Tumordiagnostik (NGS Transkriptom-Analyse)

Patient:

Klinische Angaben:
Tumorprobe

Berichtsdatum: 07.07.2022
Berichts-ID: 47390
Auftrag vom: 13.05.2022
Probeneingang: 17.06.2022
Auftragsfreigabe: 17.06.2022

Sehr geehrte Frau

wir bedanken uns für die Anforderung einer molekulargenetischen Diagnostik bei In der nachfolgenden Übersicht finden sich Expressionsdaten zu den bereits identifizierten therapierelevanten somatischen Veränderungen in derselben Tumorprobe (s. unsere Befund-ID 47263). Die Expression bestimmter Gene aus therapierelevanten Signalkaskaden ist im Anhang dargestellt. Weitere Expressionsdaten können auf Wunsch elektronisch zur Verfügung gestellt werden.

ZUSAMMENFASSUNG

- **Die Expression der therapierelevanten somatischen Varianten in *EGFR* und *MSH6* wurde bestätigt. Splice- oder trunkierende Varianten können zu RNA-mediated decay führen und werden oft im Transkriptom nicht sicher erfasst.**
- **Es zeigte sich ein Hinweis auf die onkogene Variante EGFRvIII. Eine Expression der unklaren Translokation zwischen *MTHFR* und *ATP6V1E2* wurde nicht nachgewiesen.**
- **Eine differenzielle Expressionsanalyse im Bezug zu einer Normalprobe (Großhirnrinde) zeigt eine Überexpression von *EGFR* (70-fach), *CDK6* (83-fach) und *MDM4* (15-fach).**
- **Die Bewertung der Expression bestimmter Gene aus therapierelevanten Signalkaskaden deuten auf die Aktivierung des CDK4/6 Signalweges über *CCND2* und *CDK6*.**
- **Das Expressionsprofil deutet auf eine Zugehörigkeit zur Gruppe der in-house Glioblastom-Patienten hin.**

SUPPORTING READ COUNT

Split reads at breakpoint1 = 0
Split reads at breakpoint2 = 3
Discordant mates = 0

AP 2021_12-167-g25bd968c
2022_04-165-g8fc8be95
1570
%

ext Ultra II Directional RNA Library Prep
3000 als 2x75 bzw. 2x100 bp *paired-end*

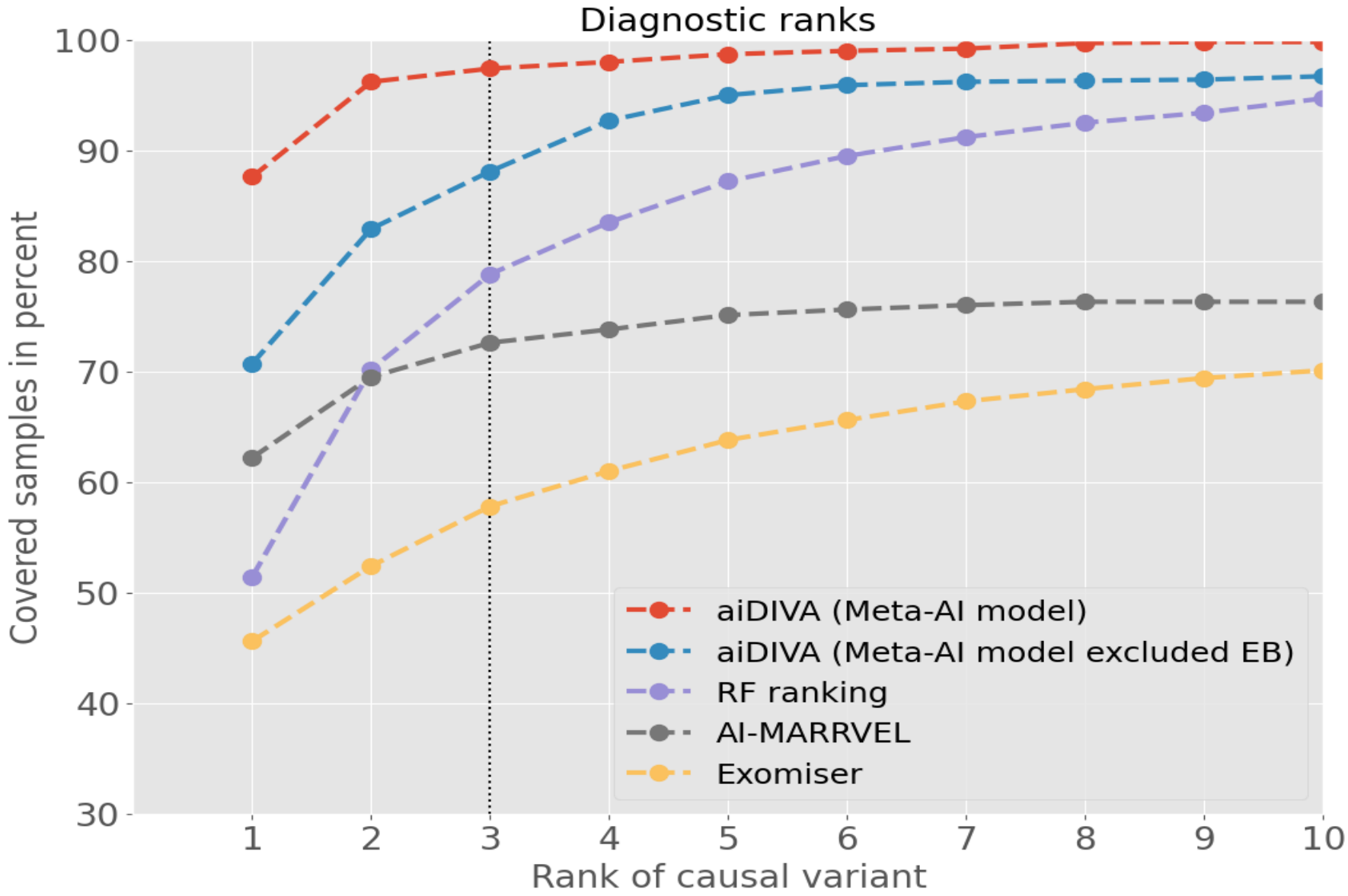
rt. Die Aufbereitung der Daten erfolgte
rte Genexpression, angegeben in TPM
wurden mit der Software STAR-Fusion

CNVs und Fusionen) wurden bereits in
lerungen wurden mit der hauseigenen
ler Varianten wurde nach den Variant
/onc_path_sop/).

kommt, wenn vorhanden, eine Angabe
Human Protein Atlas). Vergleichende

Seite 5 von 5

aiDIVA Finds Close to 100% Correct Diagnoses in Top-3 Ranks



Evaluated on 3,000 solved clinical cases of UKT

Method Development & Nanopore Implementation

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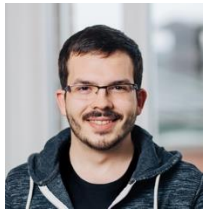
UNIVERSITÄT
TÜBINGEN

Elena Buena-Atienza

Nanopore Sequencing

Marc Sturm

Lead Diagnostic Bioinformatics



Dominic Boceck

AI Development

Caspar Gross

Long-read bioinformatics

Alexandr Chernov

Cloud Platform

Alexander Ott

Cancer Decision Support System

Jakob Admard

NGS Method development

German Demidov

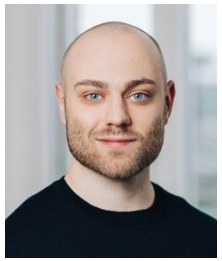
Structural Variants, Statistics

Victoria Paz Cepeda Espinoza

Pathogen Diagnostics

Leon Schütz

Decision Support Systems for RD and Nanopore



Vincent Hammer

Single Cell & Spatial Transcriptomics

Andreas Schmidt

Association Tests

Vladislav Lysenkov

Nanopore Analysis

Ramya Potabattula

T2T, Ultra-Long Reads

Fubo Cheng

T2T, PoreC

Chia Ying Ko

Repeat Expansions

Prithivi Jung Thapa

T2T Assembly

Thanks!

Olaf Riess

Tobias Haack

Medizinische Hochschule Hannover
Bernd Auber
Gunnar Schmidt

Charité – Universitätsmedizin Berlin
Nadja Ehmke

Berlin Institute of Health
Janine Altmüller
Manuel Holtgrewe
Claudia Quedenau

University Hospital Aachen
Ingo Kurth
Florian Kraft
Sebastian Gießelmann

University of Tübingen
Elena Buena-Atienza
Caspar Gross
Leon Schütz
Marc Sturm
Jakob Admard
Vladislav Lysenkov
German Demidov
Chia Ying Ko
Thomas Braun
Nicolas Casadei
Tobias Haack
Olaf Riess

Oxford Nanopore Technologies
Cerissa French
Tonya McSherry
Alexander Vogel
Daniel Mathow
Nicolai Stoye
Anthony Doran
Alexander Rotmann
Manuela Saathoff
Gerald Goh

NVIDIA
Uwe Samer
Harry Clifford
Lotfi Slim

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Karla P. Figueroa (U. of Utah)
Daniel R. Scoles (U. of Utah)
Stefan M. Pulst (U. of Utah)