Welcome to the

limbus™ WORKSHOP

Clinical diagnostics with automated SNP and CNV analysis using varvis™

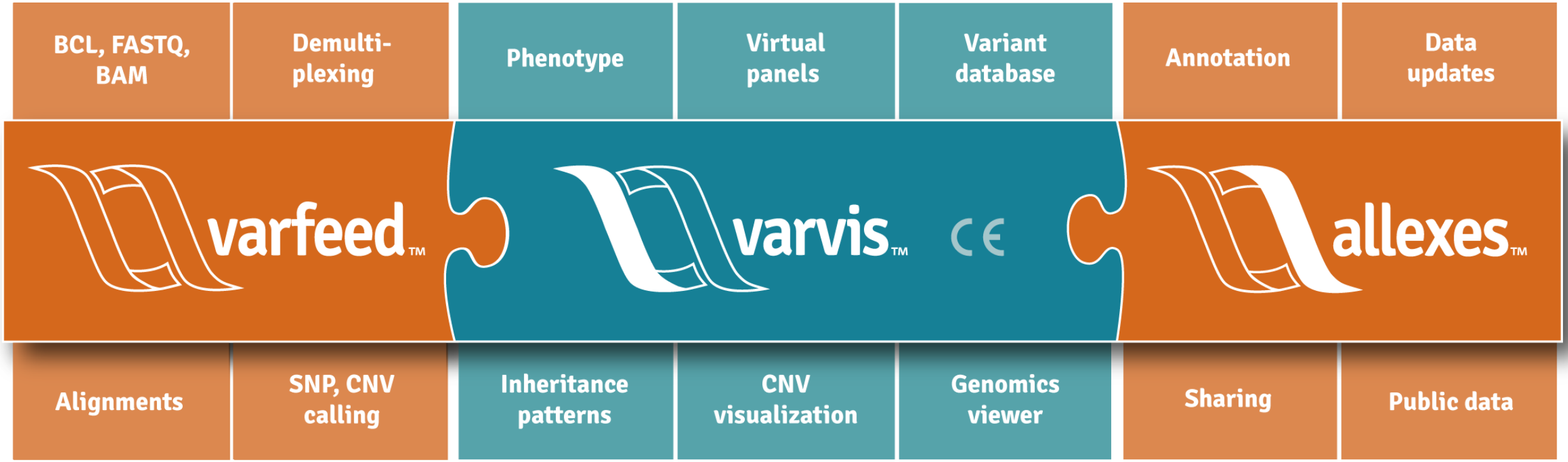
Dr. Yvonne Schmitz, Product Management

yvonne.schmitz@limbus-medtec.com

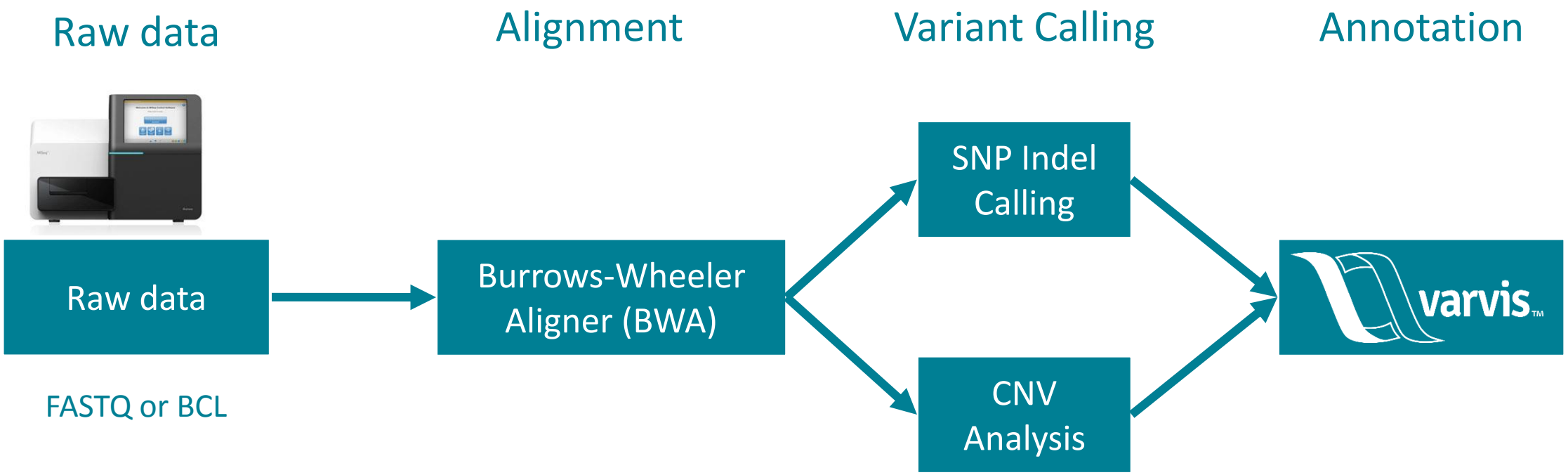
What to expect

- Brief introduction to the varvis genomics platform
  - Components and workflow
  - Data regulation, protection and availability
- Examples: real case studies from routine diagnostics
  - Presented by Dr. Konrad Platzer, Human Genetics, University Hospital Leipzig
- Hands-on practical session
  - Puzzle over some real case and play with the software yourself
- Solutions and discussion
- Final remarks

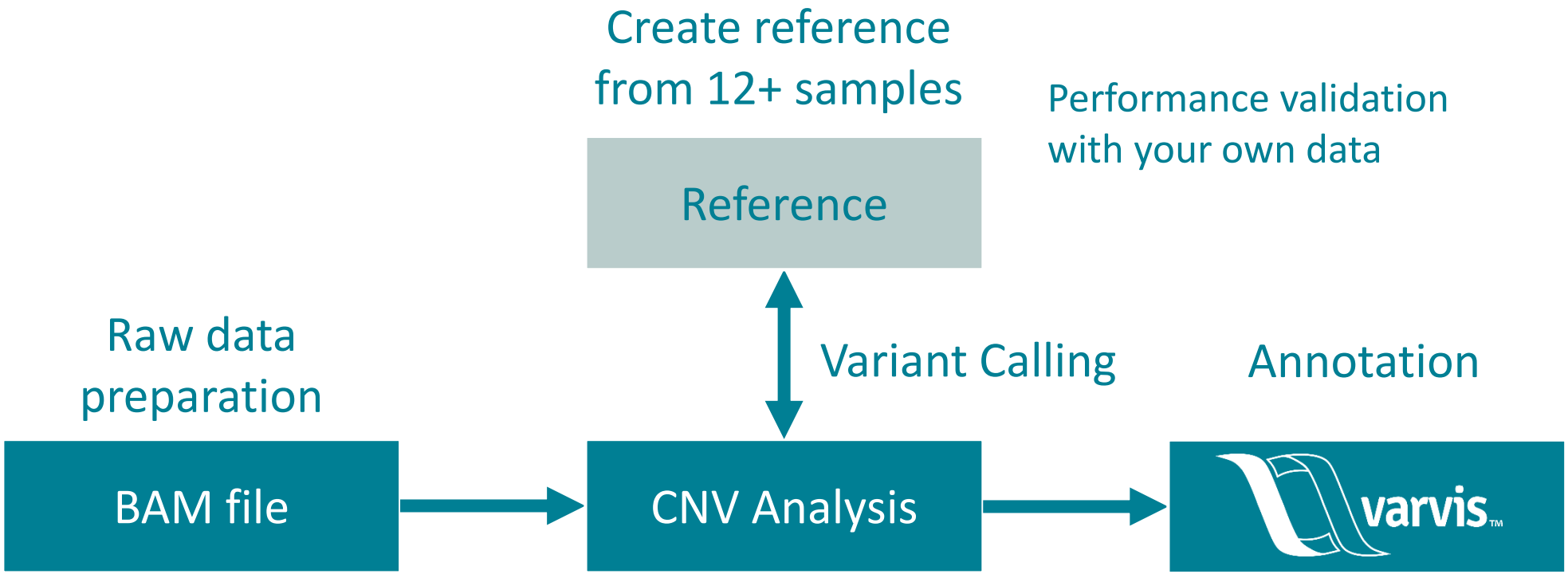
The **varvis™** genomics platform is a complete solution for clinical diagnostics



varfeed: Variant calling pipeline for NGS



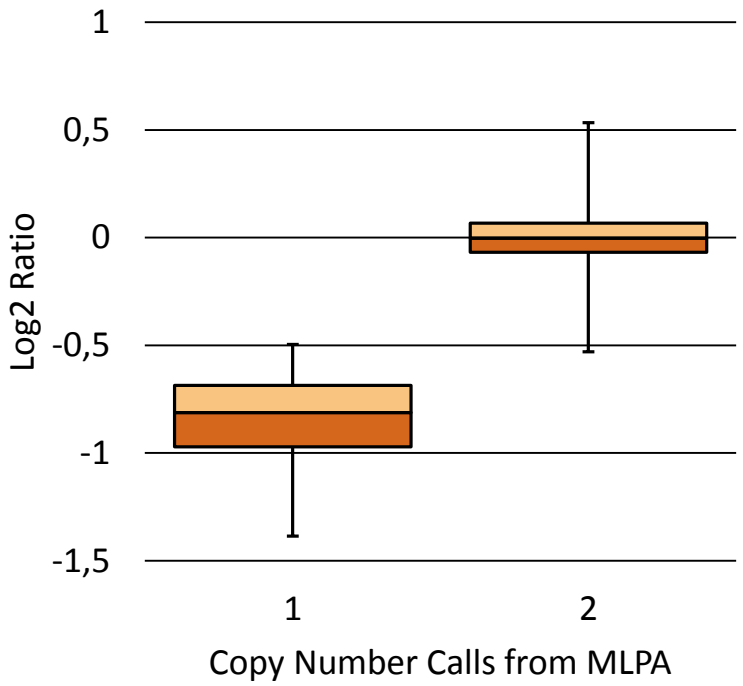
CNV analysis from targeted panels



Validated for clinical application

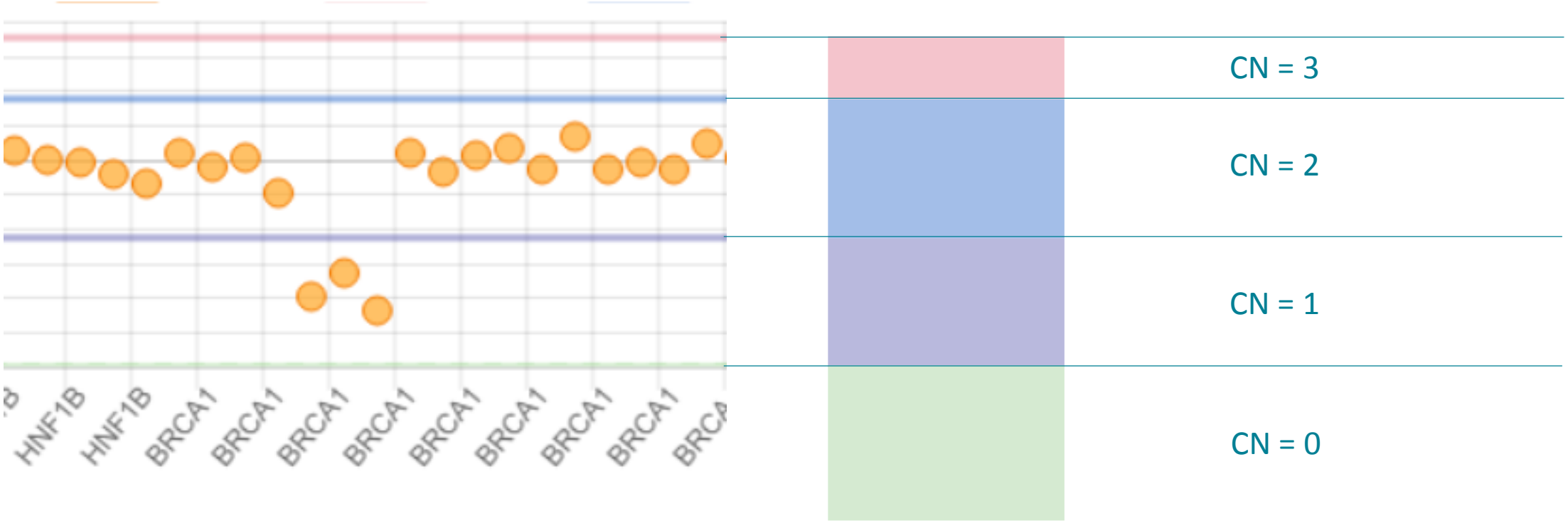
Performed on 430 TruSight Cancer Panels on MiSeq

Comparison to MLPA

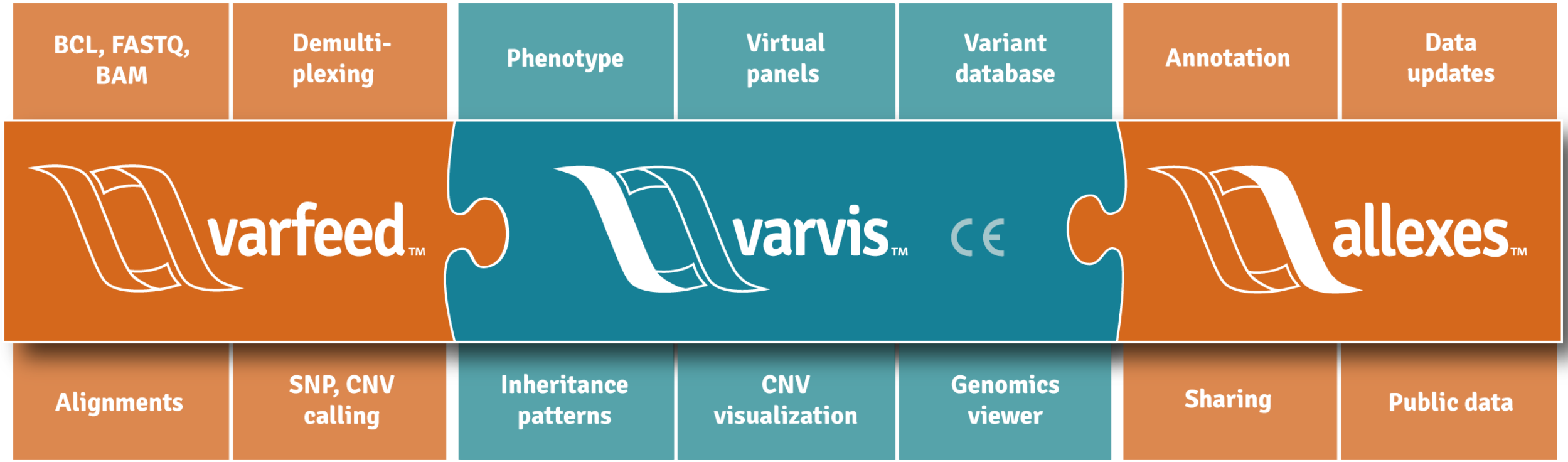


Parameter	Symbol	Value
True positive	TP	117
True negative	TN	48236
False positive	FP	10
False negative	FN	0
Sensitivity/Recall	TPR=TP/(TP+FN)	100.00%
Specificity	SPC=TN/(TN+FP)	99.98%

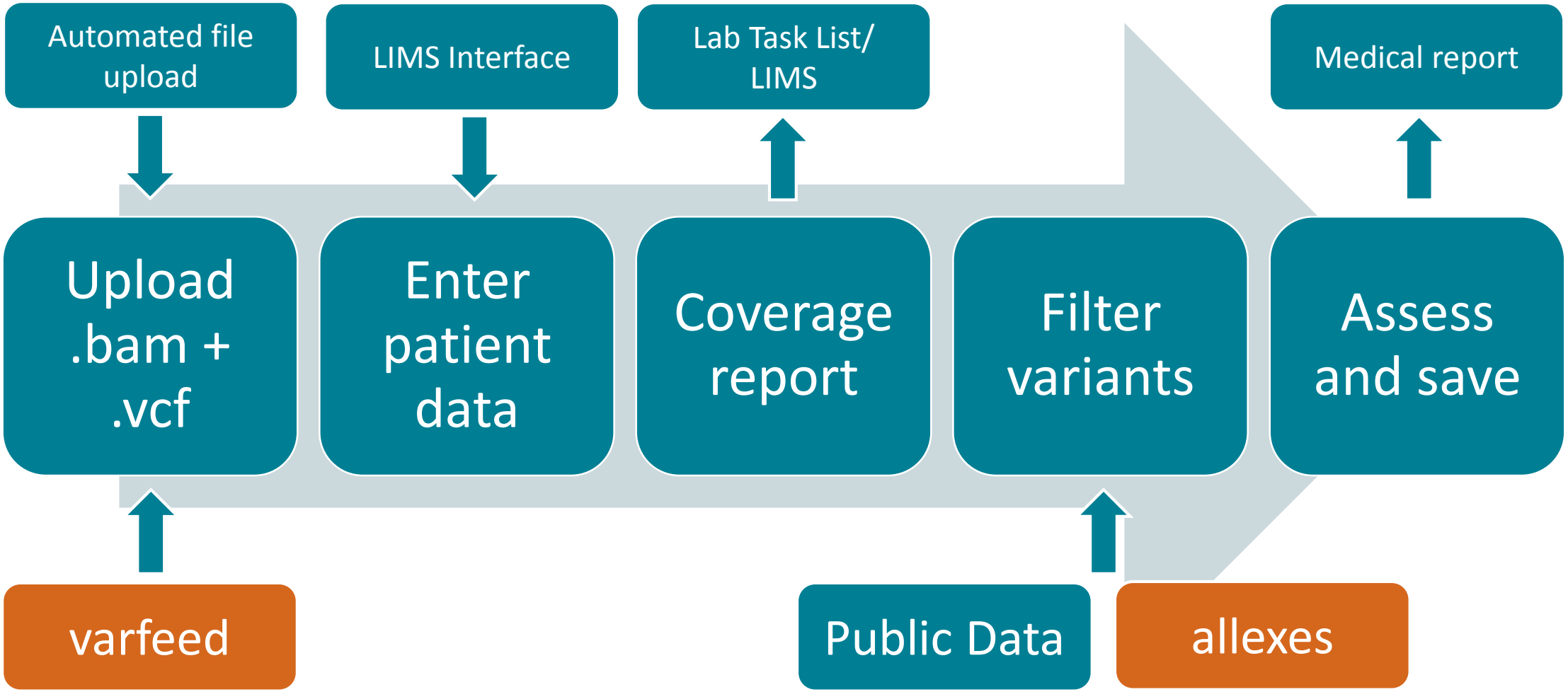
CNV visualization



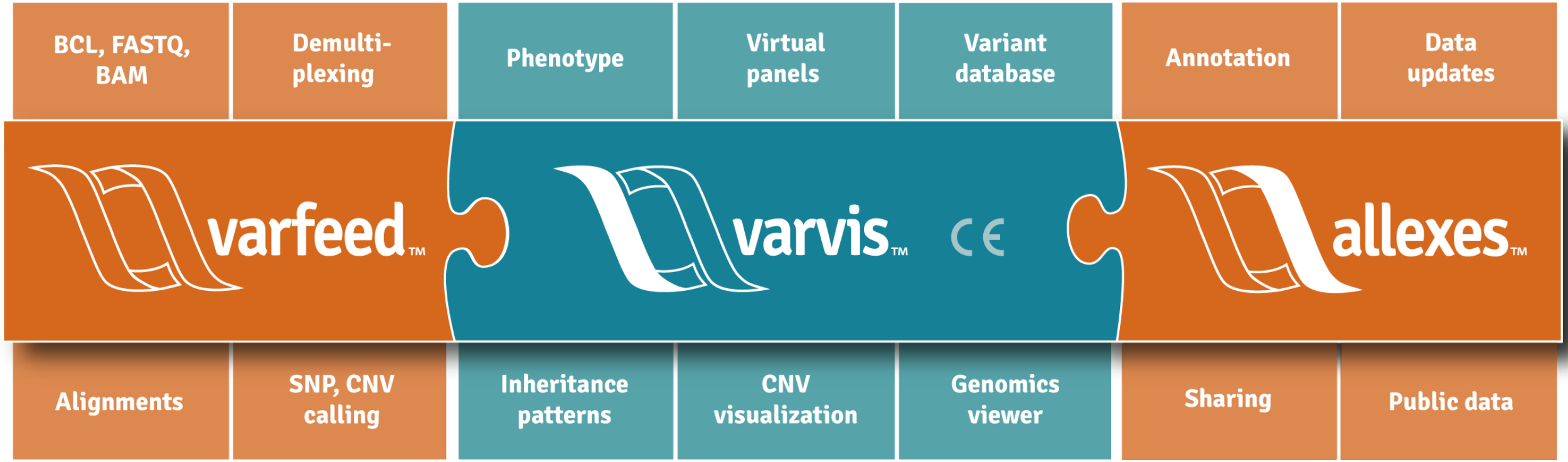
The **varvis™** genomics platform is a complete solution for clinical diagnostics



varvis Workflow



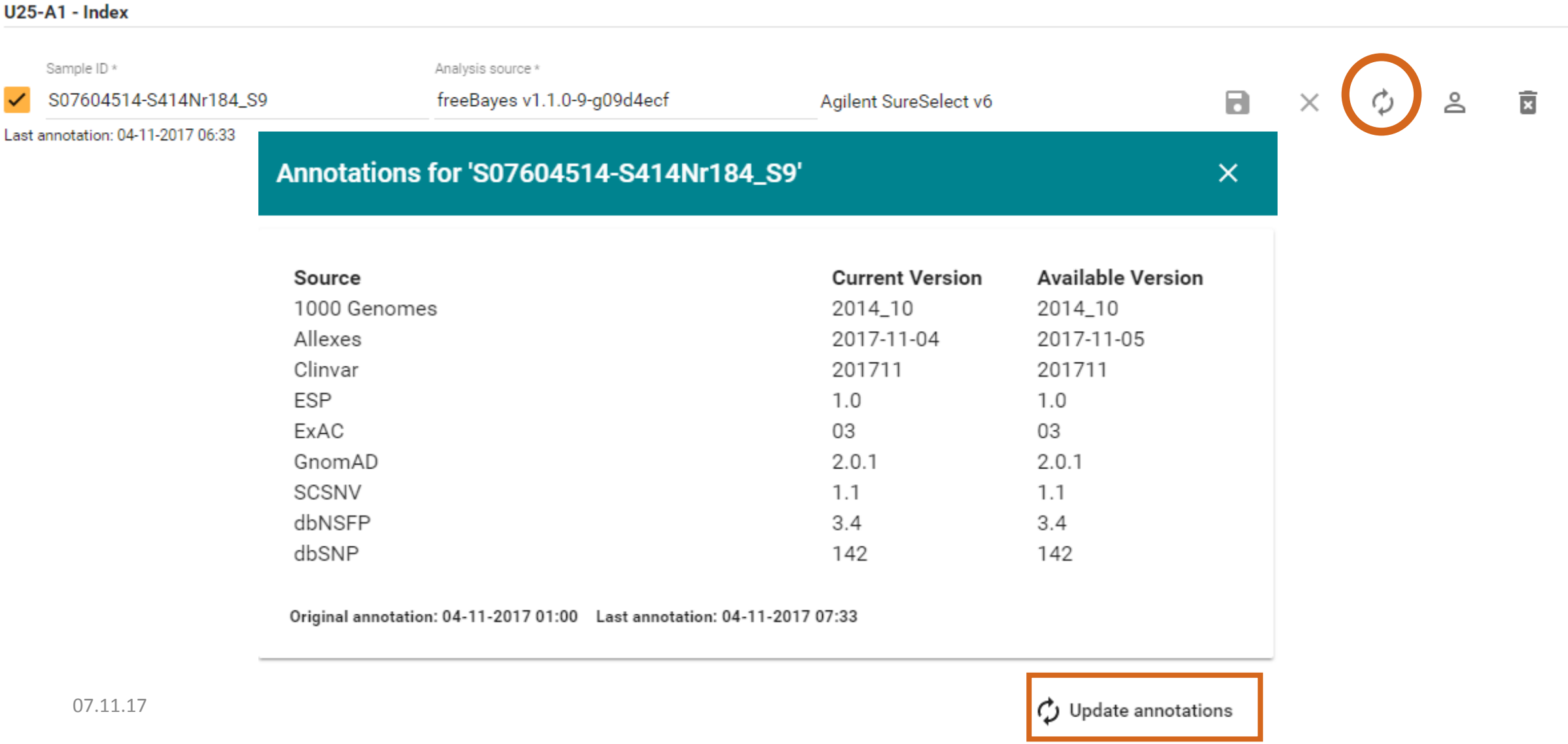
The **varvis™** genomics platform is a complete solution for clinical diagnostics



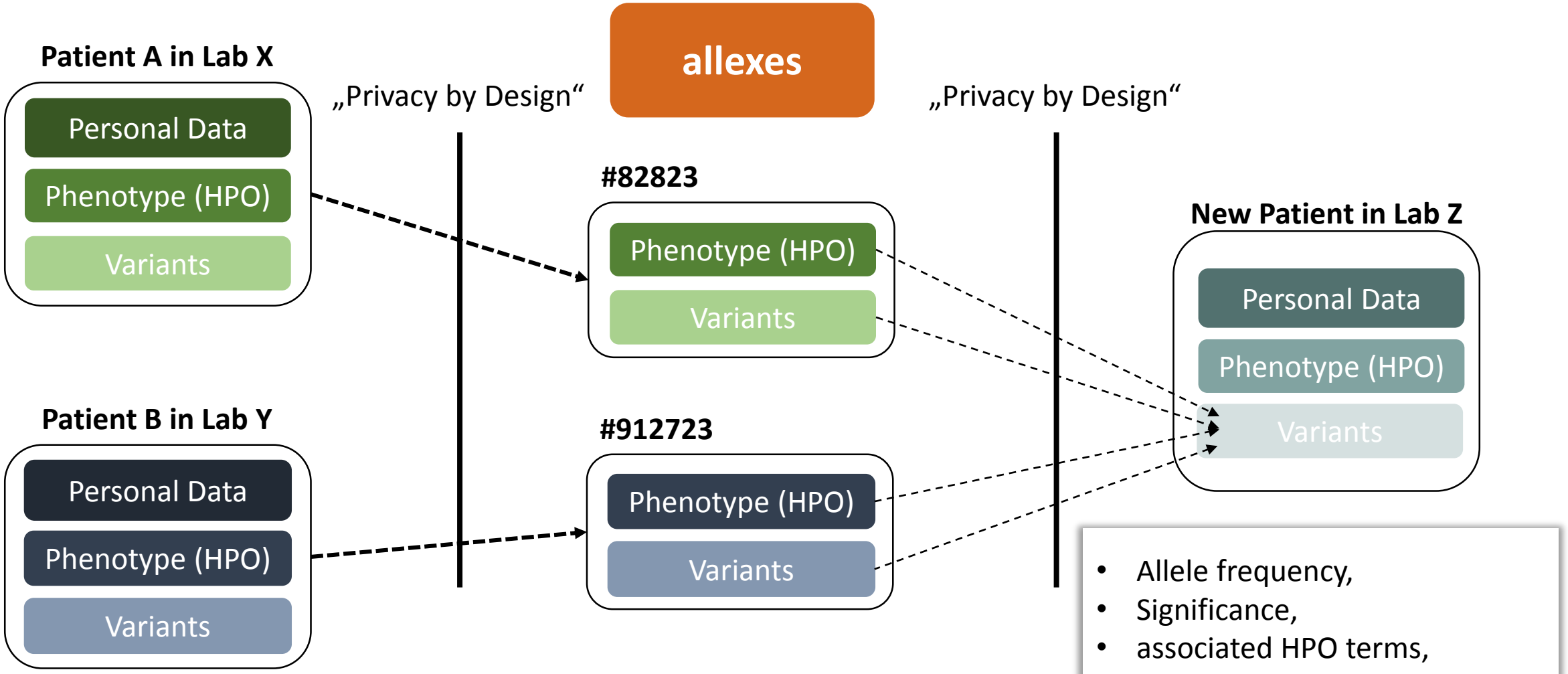
Public data: Annotation sources + Version

- **1000 Genomes: 2014_10** Afrian, American, EastAsian, European, SouthAsian, Total Frequencies
- **Clinvar: 201711:** Accession Number, Significance, Status
- **ESP: 1.0** EVS Total Frequency
- **ExAC: 03** African, American, EastAsian, European NonFinnish, Finnish, Other, SouthAsian, Total Frequencies
- **GnomAD: 2.0.1** Female, HomInd, Male, Total **GnomExs**
- **SCSNV: 1.1** ScSnvAda, ScSnvRf
- **dbNSFP: 3.4** Insilico Prediction Rankings, including GERP++, Mutation Taster, Sift and many more
- **dbSNP: 142**

Public data: Annotation sources + Version



allexes Protects Patient Data by Design



- Allele frequency,
- Significance,
- associated HPO terms,
- **no** access to patient identifiers

Medical device as cloud platform

Data privacy

- BDSG
- EU Regulation 2016/679

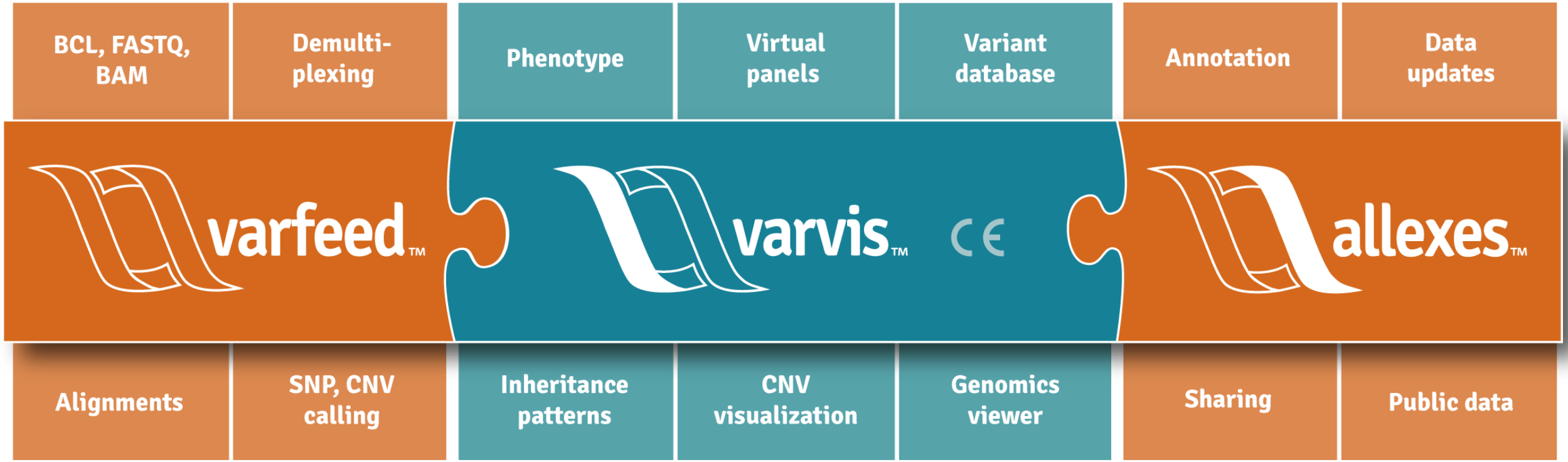
Certified data center
Data security assessment

Medical approval

- Regulation 93/42/EEC
- MPG
- ISO 13485
- ISO 14971

Quality management system
Clinical validation
Risk management

The **varvis™** genomics platform is a complete solution for clinical diagnostics



UNIVERSITÄT LEIPZIG

Medizinische Fakultät

Universitätsklinikum Leipzig

Medizin ist unsere Berufung.

variant evaluation in NGS diagnostics (clinical examples in live demo + Hands-on practical session without slides)

Konrad Platzer, MD

Institute of Human Genetics
University of Leipzig, Germany

Final remarks

Summary – key benefits of **varvis™**

Up to date

Use continuously updated public data from sources like **gnomAD** and **ClinVar**.

80% savings

Our clinically validated CNV analysis provides significant cost savings compared to ligation based CNV detection methods.

6000+

Access more than 6000 diligently curated WES cases including all variants and phenotype descriptions through our network.

CE Approved

varvis™ is a medical device Class I according to MDD 93/42/EEC.

Overnight express

No matter how many samples or how many sequencing runs you have: our fully automated process delivers results overnight.

Deep phenotyping

varvis™ enables standardized phenotyping using ontologies like HPO to improve the accuracy of your diagnosis.

Thank you for attending our workshop!

Do you have **additional questions or want a live demo or varvis**?

Are you interested in the **clinical validation** of your CNV data?

Would you like to test **varvis** with your **own data**?

Please visit www.varvis.com for more information

Or contact yvonne.schmitz@limbus-medtec.com