Vers une application en routine de la cartographie optique du génome (OGM)

Présentation d'études récentes et des développements produits

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OGM results are concordant with SOC and increase reportable findings in unresolved cases

Multisite Study with OGM in Postnatal

>1,000	**Datapoints study** SOC (Karyotyping, FISH, microarray, Southern Blotting and PCR)
99.6%	**Concordance between OGM and SOC results** 98.7% full concordance
10.1% to 14.8%	**Increase in variants reported**

Broeckel U et al. Multisite Study of Optical Genome Mapping of Retrospective and Prospective Constitutional Disorder Cohorts. medRxiv 2022.12.26.22283900; doi: https://doi.org/10.1101/2022.12.26.22283900

Combining NGS and OGM for unresolved cases

50	**Unresolved cases** Following CMA +/- WES
40%	**Increase in pathogenic findings** (35% exome negative cases)
76%	**Pathogenic or candidate variant identified** (57.9% WES and CMA negative cases)

Shieh, J.T., Penon-Portmann, M., Wong, K.H.Y. *et al.* Application of full-genome analysis to diagnose rare monogenic disorders. *npj Genom. Med.* **6**, 77 (2021). https://doi.org/10.1038/s41525-021-00241-5

Faster and More Complete Genomic Analysis Solutions for Clinical Research

DNA isolation

Ionic Automated Extraction System

OGM

SVs and CNVs (>500 bp)

Stratys
High Throughput system
(>480 samples)

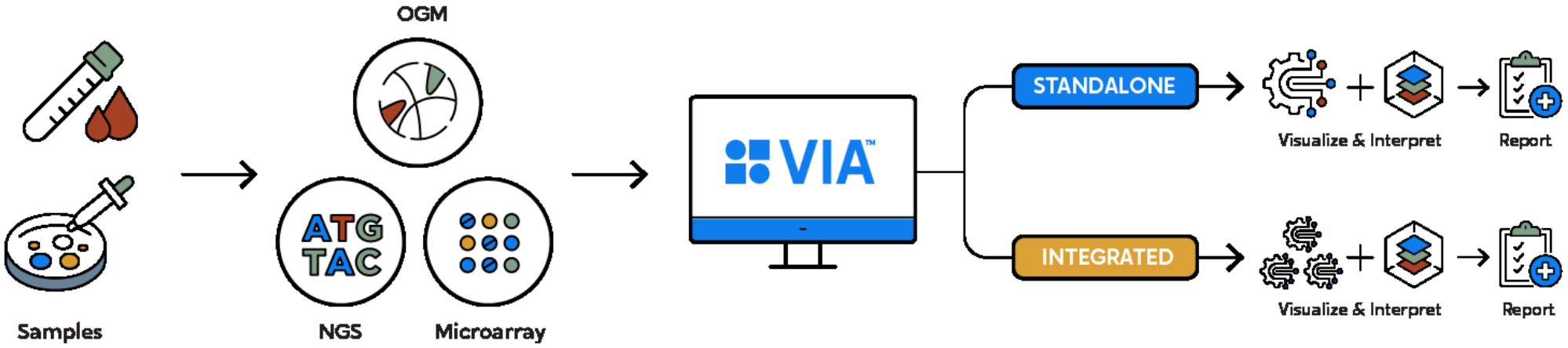
+

Sequencing or Arrays (not a Bionano solution)
SNVs & Indels (< 500 bp) or CNVs

Data Analysis

VIA software, automated & agnostic

VIA CNV Constitutional workflow overview



External + Internal **Databases**

Phenotype Based **Variant Prioritization**

Familial /Inheritance analyses

Annotate Variants based on characterized previous results

Automated **Classification**

Integrated database & knowledgebase

Thank You

Questions?

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