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				4/20	4/291	PSEN1 NM 000021.4	chr14.72136423 O ISV	rs1802839		SNV	upstream gene variant	48	128	67:65	0/1		
	369-21	0	2	5/20	5/291	PSEN1 NM.000021.4	chr14/22219602 C) 10V	0262284	0.52120-8	SNV	3 prime UTR variant	44	170	95:75	0/1		
	369-21	•	1	14/20	14/291	PSEN1	chr14/73220436	m165935	e.*11470+T	SNV	3 prime UTR variant	100	135	0:135	1/1		
	369-21	۰	1	14/20	14/291	PSEN1	chr14.73198145	m165932	c.868+16G-T	SNV	intron variant	100	138	0:138	1/1		
	369-21	0		18 / 20	18/291	PSEN1 NM.000021.4	chr14.73163930 Q. KV	rs774839177	0.88.6855del	deletion	intron variant		106	95:10	011		
	369-21	•	۰	12/20	12/291	PSEN1 NM,000021.4	chr14.73142943 Q. KV	11214225	6-135-48520×A	SNV	intron variant	100	113	0:113	1/1		
	369-21	•	۰	16/20	16/291	PSEN1 NM.000021.4	chr14.73135689 Q. KSV			SNV	upstream gene variant	4	140	134:6	en.		
	369-21	0	•	8/20	8/291	PSEN1 NML000021.4	chr14.73173930 Q. IGV	0214269	c.480+223A+G	SNV	intron variant	99	112	0:112	1/1		
	369-21	0	۰	17/20	17/291	PSEN1 NM.000021.4	chr14.73141316 Q KSV	rs146372308	c-135+4745del	deletion	intron variant	10	104	92:12	01		
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	•	1 369-21 369-21 369-21 369-21 369-23 369-23 369-23 369-23 369-23	34927     C       34929     C       700     dr4.4     v       4997     C       700     dr4.4     v       4000     T     C	198-27 1 1 1 1 198-27 2 4 198-27 2 4 199-28	MA21     T     M.23       MA2     T     M.24       MA2     T     M.25       MA2     M.25     M.25       MA2     M.25     M.25	No.21     V     V     V     V     V     V     NO     NO <td>ματο     ματο     &lt;</td> <td>Apr.2     D     L/J     L/J     M/J     M/J<td>Mark     Mark     <thmark< th="">     Mark     Mark     <thm< td=""><td>MAR     Mar     Mar<td>MAD     M     V     V/2     MADE     CONS     MADE     MADE<td>BAD B 1 1.0.0 10.00</td><td>BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3 BAD 3</td><td>MAD MAD MAD<td>ABAS ABAS ABAS</td><td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td><td>MAR M&lt;</td></td></td></td></thm<></thmark<></td></td>	ματο     <	Apr.2     D     L/J     L/J     M/J     M/J <td>Mark     Mark     <thmark< th="">     Mark     Mark     <thm< td=""><td>MAR     Mar     Mar<td>MAD     M     V     V/2     MADE     CONS     MADE     MADE<td>BAD B 1 1.0.0 10.00</td><td>BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3 BAD 3</td><td>MAD MAD MAD<td>ABAS ABAS ABAS</td><td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td><td>MAR M&lt;</td></td></td></td></thm<></thmark<></td>	Mark     Mark <thmark< th="">     Mark     Mark     <thm< td=""><td>MAR     Mar     Mar<td>MAD     M     V     V/2     MADE     CONS     MADE     MADE<td>BAD B 1 1.0.0 10.00</td><td>BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3 BAD 3</td><td>MAD MAD MAD<td>ABAS ABAS ABAS</td><td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td><td>MAR M&lt;</td></td></td></td></thm<></thmark<>	MAR     Mar <td>MAD     M     V     V/2     MADE     CONS     MADE     MADE<td>BAD B 1 1.0.0 10.00</td><td>BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3 BAD 3</td><td>MAD MAD MAD<td>ABAS ABAS ABAS</td><td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td><td>MAR M&lt;</td></td></td>	MAD     M     V     V/2     MADE     CONS     MADE     MADE <td>BAD B 1 1.0.0 10.00</td> <td>BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3 BAD 3</td> <td>MAD MAD MAD<td>ABAS ABAS ABAS</td><td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td><td>MAR M&lt;</td></td>	BAD B 1 1.0.0 10.00	BAD 3 I U.J.0 U.J.00 W.L.MILL O.W SAME 1 L.D.10.5 BAD 3	MAD <td>ABAS ABAS ABAS</td> <td>MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000</td> <td>MAR M&lt;</td>	ABAS	MAR 2 M MO 00 MO 0000 MO 00000 MO 000000 MO 000000 MO 0000000 MO 0000000 MO 0000000 MO 00000000 MO 00000000 MO 000000000 MO 000000000 MO 000000000000000000000000000000000000	MAR M<

**GENOVESA** – cloud-based database bioinformatics and interpretation system

Analysis of NGS genetic data can be a challenging task that often requires specialized user skills. We developed a cloud-based bioinformatics software – GENOVESA as an easy solution for everyone and for all NGS data regardless of the sample preparation kit used.



## **Key features**

- User friendly
- Automatized bioinformatics analysis of NGS data
- Advanced quality control of sequencing data
- Easy variants filtration
- Local Clinical Variants Database
- Visualization of NGS data
- Internal interpretation of variants and sharing between clinics
- Clinical report generation

# From FASTQ to clinical report

GENOVESA enables evaluation of data from small panels, through clinical exome (CES) whole exome data (WES) to whole genome sequencing (WGS) data. It includes a wide range of annotation databases and the possibility of individual customization. FASTQ, BAM and VCF data quality control. Possibility of own interpretation of variants and comments. Complete clinical management with integration of International Classification of Diseases (ICD) and HPO terminology.

### Sequencing technology

GENOVESA enables analysis of conventional sequencing data (FASTQ, BAM, VCF) regardless of what sequencing technology was used – Illumina, MGI, PacBio, Oxford Nanopore Technologies, Genapsys and many others.

### **Databases and data sharing**

GENOVESA also serves as a database for storing variants, with the possibility of sharing data between individual clinics.

#### Security

GENOVESA prioritizes data security through HTTPS encryption. We do not rely on third-party providers like Google or Amazon; instead, we host your data in our secure data center located in Prague. We do not collect any sensitive patient data, GENOVESA is fully GDPR compliant.

**Contact us:** 

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### Genovesa is a joint project of the companies:



BioVendor MDx

BioVendor MDx a.s.

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