

The background is a dark blue gradient with glowing, out-of-focus binary code (0s and 1s) and abstract, curved lines of light blue and white, suggesting a digital or technological theme.

Pronalaženje de novo varijanti deteta

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Cilj projekta

Ulazni podaci:

- FASTQ fajlovi sa genomima majke, oca i deteta

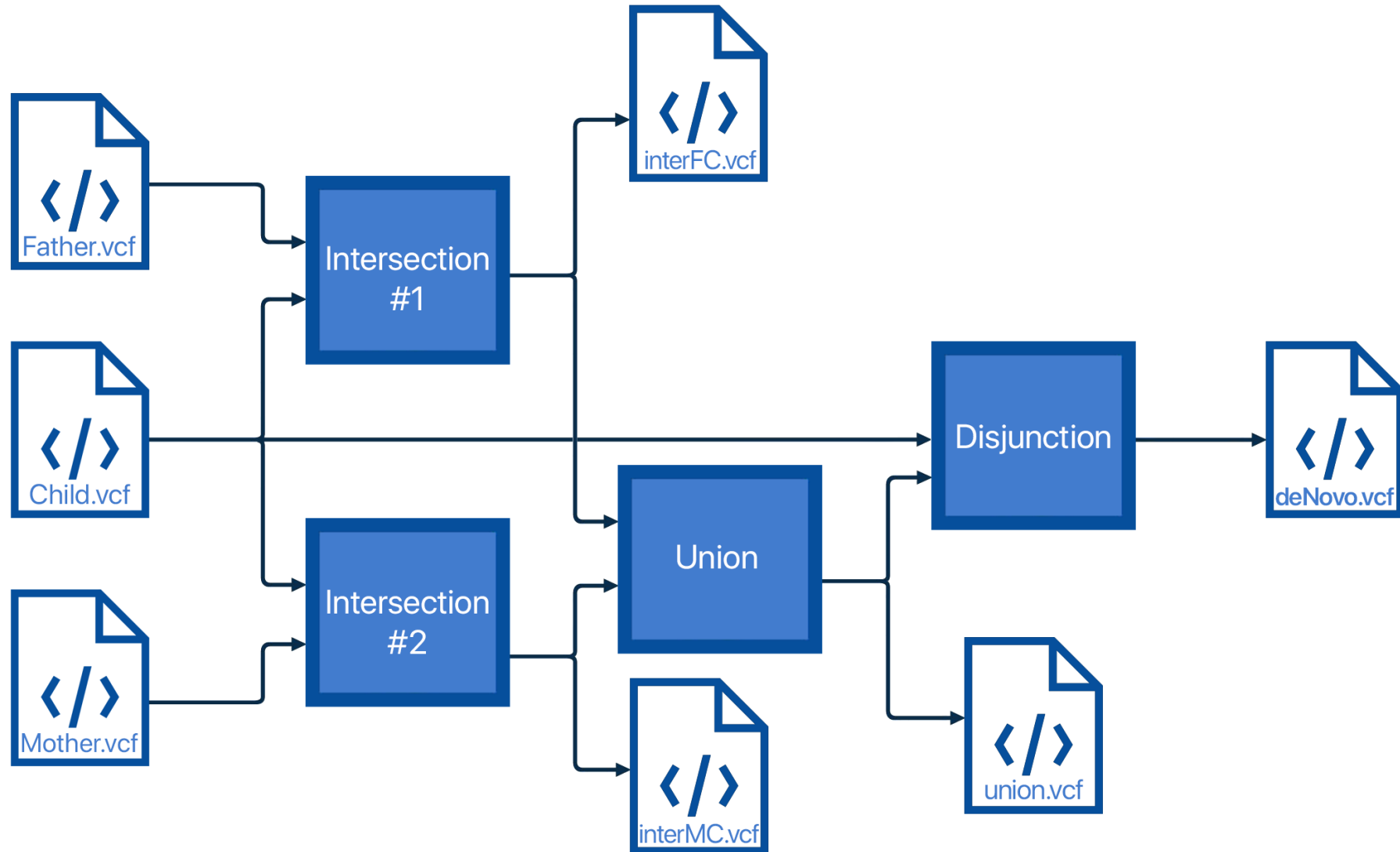
Izlazni podaci:

- VCF fajl sa de novo varijantama deteta

Implementacija rešenja

1. Rekonstrukcija genoma – GRAF Germline Variant Detection Workflow
2. Pravljenje alata za pronalaženje de novo varijanti deteta
3. Pronalaženje de novo varijanti pomoću RTG Tools VCFEval
4. Upoređivanje rezultata i korišćenje SBG VCF Benchmark alata za vizuelizaciju razlika

Pravljenje alata za pronalaženje de novo varijanti deteta



Somatski hromozomi

- U svakom paru hromozoma deteta – jedan hromozom od majke, drugi od oca
- Mutacija na jednom ili na oba hromozoma roditelja – veliki broj kombinacija za dete

Parent	Child	Intersection
0/0	0/0	Not detected
0/0	0/1	Skip
0/0	1/1	Skip
0/0	1/2	Skip
0/1	0/0	Skip
0/1	0/1	if ref1==ref2 and alt1==alt2 write 0/1, else skip
0/1	1/1	if ref1==ref2 and alt1==alt2 write 0/1, else skip
0/1	1/2	if ref1==ref2 and alt1[0]==alt2[0 or 1] write 0/1, else skip
1/1	0/0	Skip (not possible?)
1/1	0/1	if ref1==ref2 and alt1==alt2 write 0/1, else print(error)
1/1	1/1	if ref1==ref2 and alt1==alt2 write 1/1, else skip
1/1	1/2	if ref1==ref2 and alt1[0]==alt2[0 or 1] write 0/1, else skip
1/2	0/0	Skip (not possible?)
1/2	0/1	if ref1==ref2 and alt1[0 or 1]==alt2[0] write 0/1, else skip
1/2	1/1	if ref1==ref2 and alt1[0 or 1]==alt2[0] write 0/1, else skip
1/2	1/2	if ref1==ref2 and alt1[0 or 1]==alt2[0 or 2] write 0/1 or ½, else skip

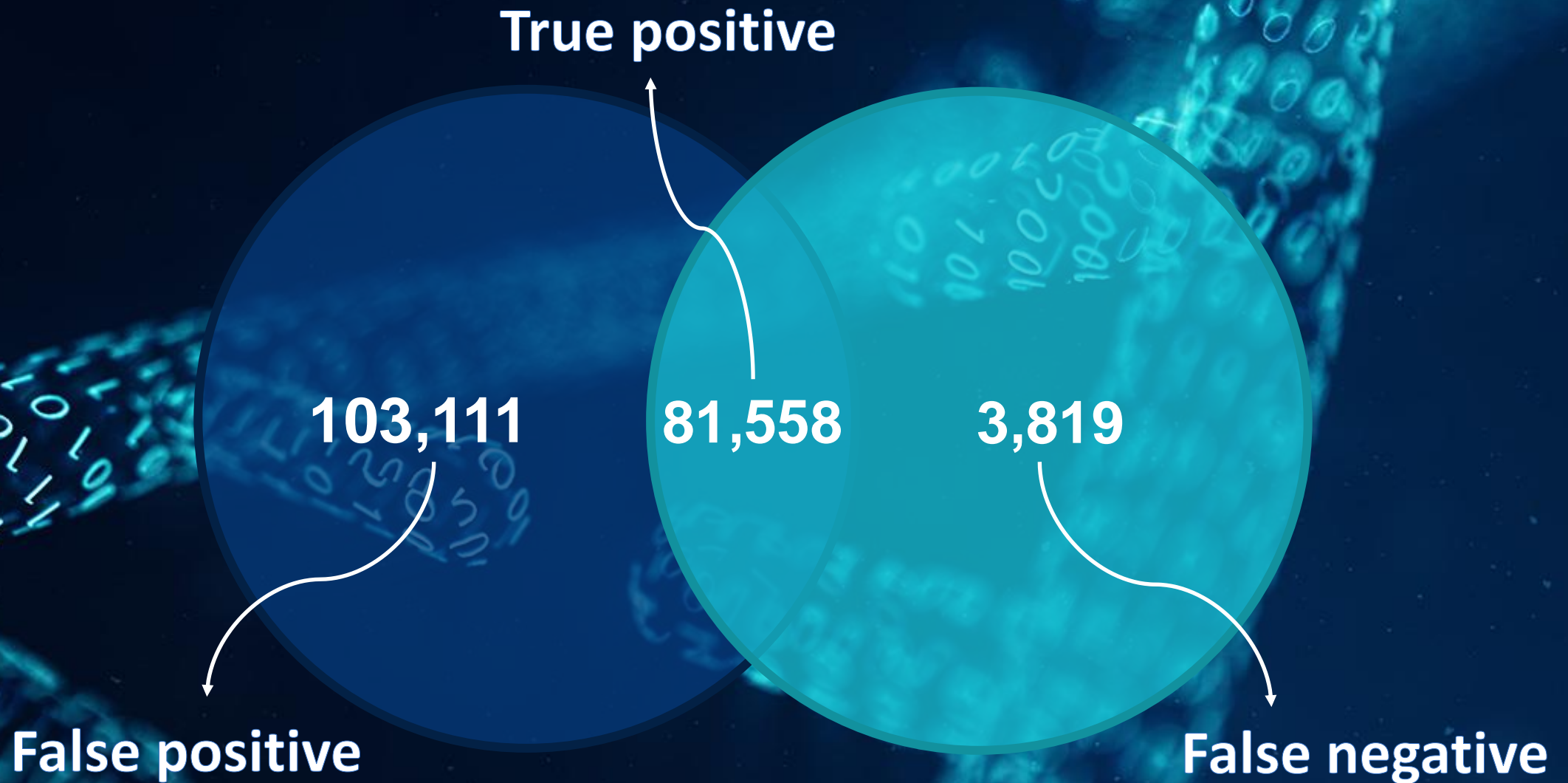
X i Y hromozomi

- Dete (sin) nasleđuje X od majke, Y od oca
- U VCF fajlu majka ima Y hromozom?
- Neke varijante na polnim hromozomima imaju genotip 1 / 2 ?
- Posebna logika za presek polnih hromozoma

Rezultati

	This project	Partial	RTG Tools
Number of de novo variations detected	184,689	131,570	85,381
Percentage [%]	3.765	2.682	1.740

Rezultati



Rezultati

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	NA24385
chr1	66521	.	T	TATATA	608.27	PASS	...	GT:...	0/1:...
chr1	66522	.	A	ATATAT	378.36	PASS	...	GT:...	0/1:...

VCF	#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	NA24385
Child	chr1	3719890	.	T	TTG,TTGTG	173.64	PASS	...	GT:...	1/2:...
Mother	chr1	3719890	.	TTG	T,TTGTGTG	173.64	PASS	...	GT:...	1/2:...
Father	chr1	3719890	.	TTG	T,TTGTG	173.64	PASS	...	GT:...	1/2:...

VCF	#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	NA24385
Child	chr1	775840	.	CA	C,CAA	182.92	PASS	...	GT:...	1/2:...
Mother	chr1	775840	.	CA	CA	182.92	PASS	...	GT:...	1/1:...
Father	chr1	775840	.	CA	CA	182.92	PASS	...	GT:...	1/1:...

The background is a dark blue gradient with several glowing, translucent blue lines that curve and flow across the frame. These lines are composed of binary digits (0s and 1s) in a monospace font, giving the impression of digital data or code being transmitted or processed. The overall aesthetic is futuristic and tech-oriented.

Hvala na pažnji!