- VEP annotation -> ALoFT run

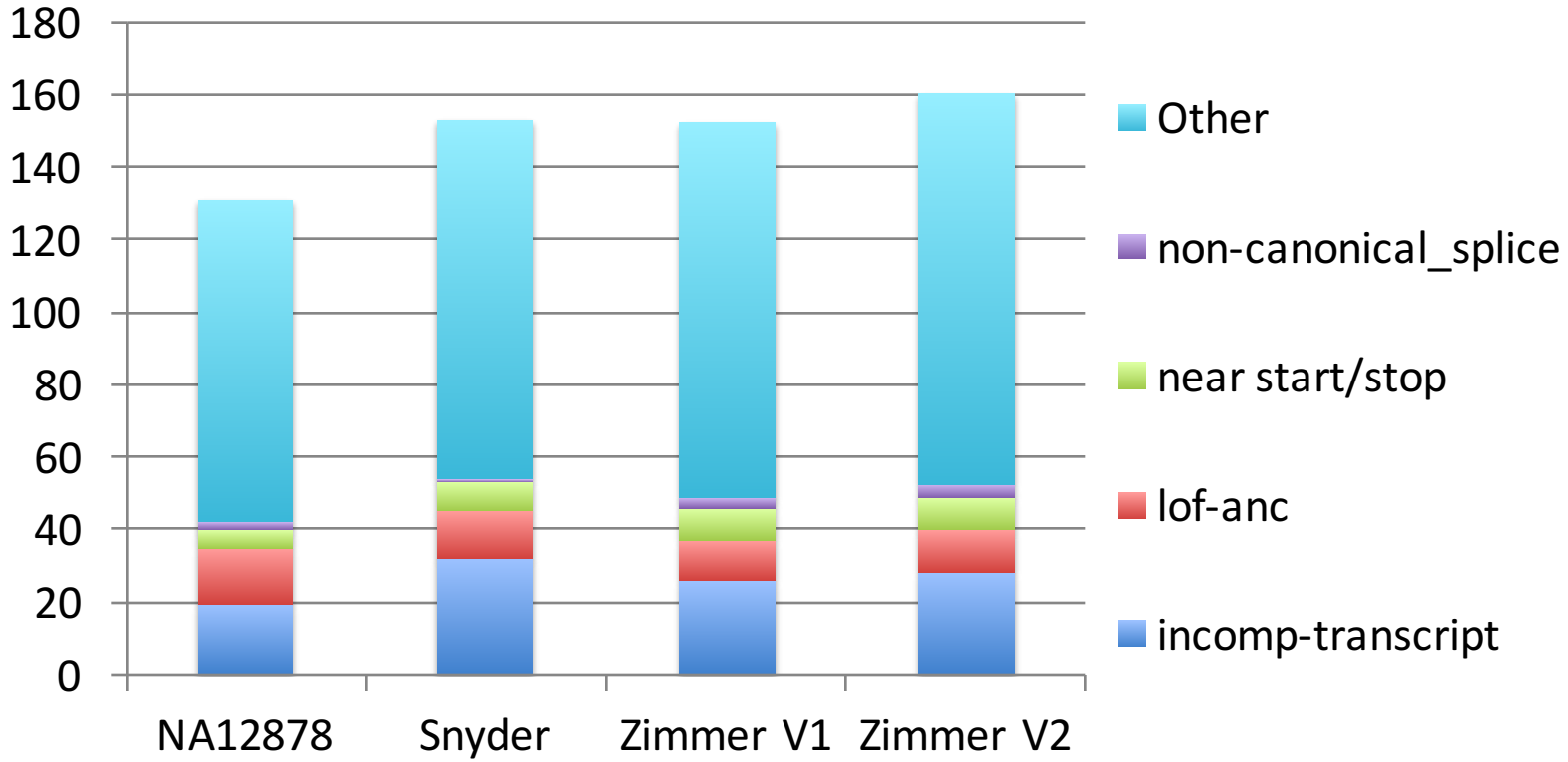
- # of mutations

	# LOF (without flags)	Splice (without flags)
NA12878	80 (49)	308 (28)
Snyder	91 (53)	357 (57)
Zimmer V1	86 (55)	302 (24)
Zimmer V2	90 (57)	310 (26)

- # of genes

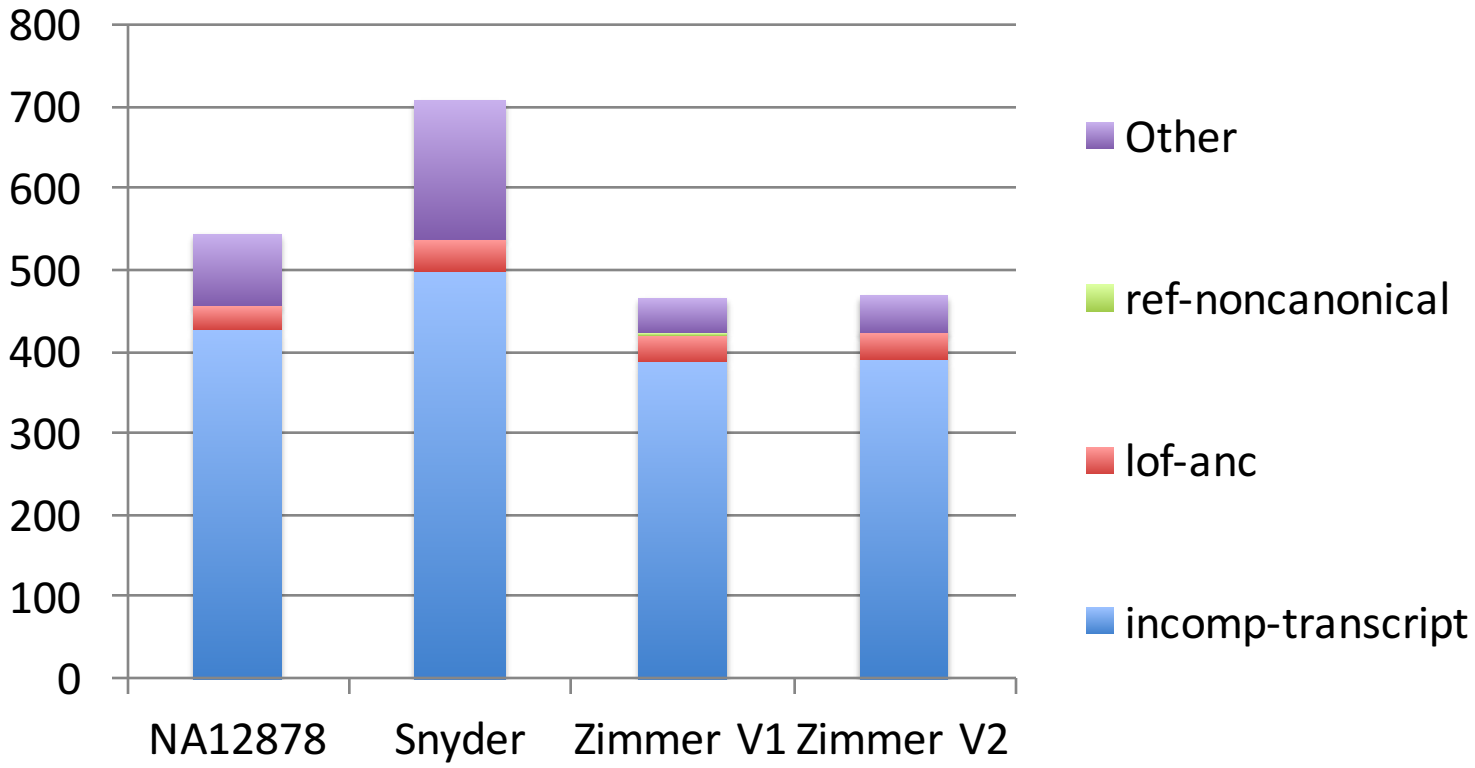
	# LOF (without flags)	Splice (without flags)
NA12878	80 (49)	275 (29)
Snyder	89 (51)	319 (56)
Zimmer V1	85 (54)	268 (25)
Zimmer V2	88 (55)	274 (27)

- **Transcript-level Flags**
- **LoF**

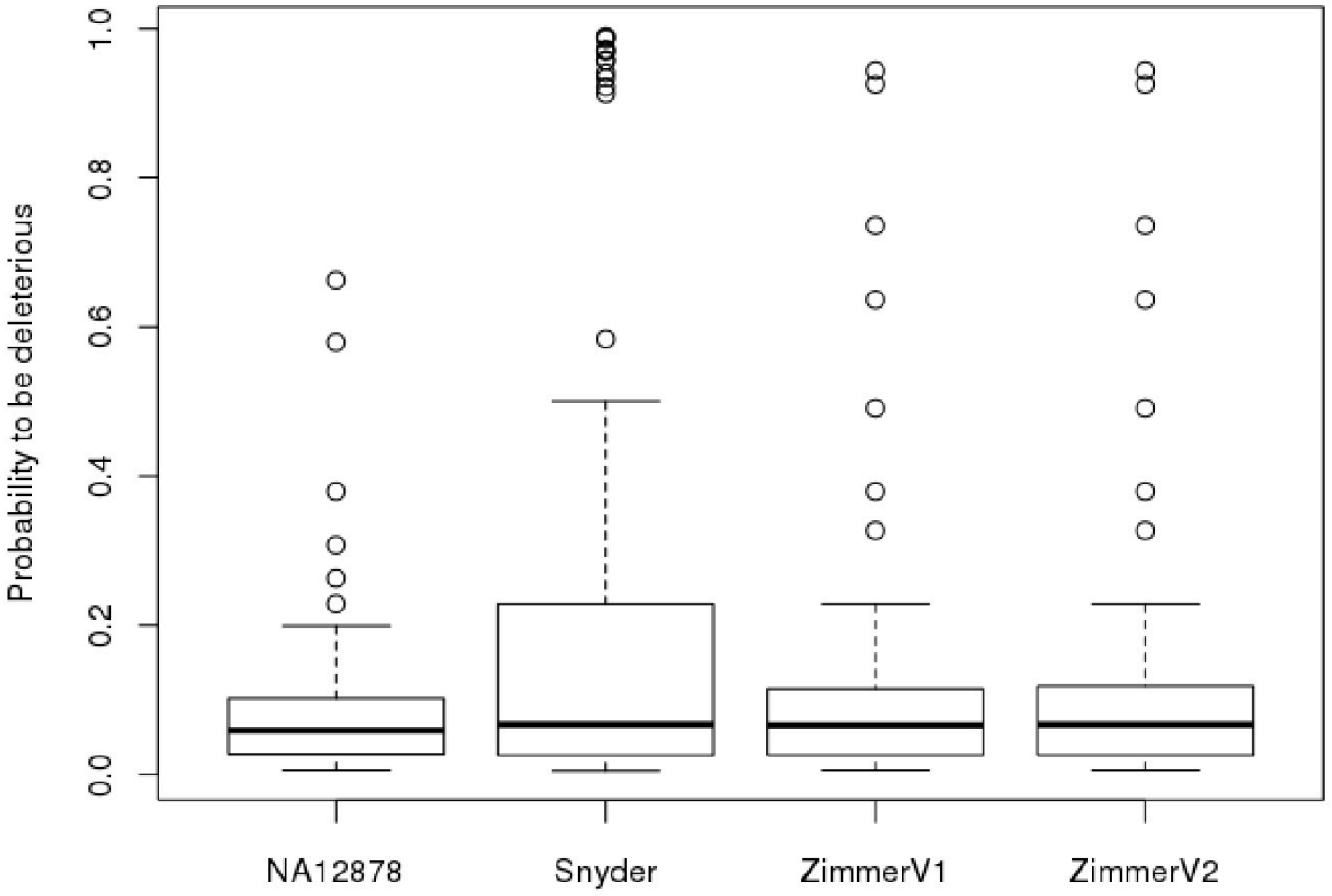


	Other
NA12878	89 (56 NMD)
Snyder	99 (67 NMD)
Zimmer V1	103 (58 NMD)
Zimmer V2	108 (63 NMD)

Transcript-level Flags
Splice



LOF variants



Potential deleterious variant
Zimmer

chr	pos	ref	alt	gene	Score	genotype	Gene function
6	17606162	C	T	FAM8A1	0.94365	0/1	Unknown, Autism related ??? Pubmed: 22495306
6	155577717	T	A	TIAM2	0.63655	0/1	Cell migration
17	61829719	A	C	CCDC47	0.92540	0/1	unknown
19	759925	C	A	MISP	0.73605	0/1	Mitotic spindle positioning

No disease associations in OMIM

CCDC47 associated with Schizophrenia

Potential deleterious variant

Snyder **....** something is wrong ... (metabolism ???)

chr	pos	ref	alt	gene	Score	genotype	OMIM
2	44079970	C	A	ABCG8	0.92190	0/1	Sitosterolemia
2	215854316	T	A	ABCA12	0.97240	0/1	Ichthyosis
2	216240022	G	T	FN1	0.98975	0/1	fibronectin deficiency
9	111718091	G	T	CTNNAL1	0.98845	0/1	
9	130635074	G	T	AK1	0.96915	0/1	Hemolytic anemia
10	29581479	C	A	LYZL1	0.58365	0/1	
11	64056777	C	A	GPR137	0.94075	0/1	
12	18800840	G	T	PIK3C2G	0.95735	0/1	
12	122400030	C	A	WDR66	0.93380	0/1	
14	71570264	C	A	PCNX	0.98635	0/1	
15	68504073	G	T	CLN6	0.97080	0/1	Ceroid lipofuscinosiss
15	93007504	C	A	ST8SIA2	0.91290	0/1	
20	5157344	C	A	CDS2	0.95755	0/1	