

# Comparative Studies of Copy Number Variation Detection Methods for Next-Generation Sequencing Technologies

This paper deals with various CNV detection methods and their performance on various variables and their combinations.

To measure performance the variables we use are:

Parameter of interest	Values	Fixed parameters
$l$ (kbp)	0.8,1,2,6	$n=6, c=5$
$n$	0,1,3,6	$l=6$ kbp, $c=5$
$c$	3,5,10,30	$l=6$ kbp, $n=5$

Here,  $l$  = length of a single copy number

$n$  = number of blocks in a fixed length

$c$  = coverage of copy numbers

We examine the methods over two types of data. Simulated data and real data. Simulated data are made with maintenance of the above mentioned variables. The performance graphs are below:

The means and standard deviations of estimation error of break point position.						
	CNV-seq	FREEC	readDepth	CNVnator	SegSeq	EWT
$l=0.8$ kbp, $n=6$ , $c=5$	395±1.67e3	729±87	32±10	NA	686±1.78e3	36±17
$l=2$ kbp, $n=6$ , $c=5$	449±218	793±752	29±92	2±91	884±552	83±82
$l=6$ kbp, $n=6$ , $c=5$	100±138	479±251	28±22	2±48	1.22e3±1.37e3	31±14
$l=6$ kbp, $n=0$ , $c=5$	116±270	359±548	33±0	113±51	417±9.07e3	5.13e3±5.05e3
$l=6$ kbp, $n=1$ , $c=5$	613±838	274±136	25±80	177±442	194±5.28e3	30±194
$l=6$ kbp, $n=3$ , $c=5$	1.28e3±1.57e3	269±178	1.6e3±2.15e3	79±404	561±1.09e3	1.71e3±2.05e3
$l=6$ kbp, $n=6$ , $c=3$	643±458	669±183	NA	49±363	2.76e3±1.48e3	75±88
$l=6$ kbp, $n=6$ , $c=10$	203±81	729±50	32±10	59±290	88±9.15e3	36±17
$l=6$ kbp, $n=6$ , $c=30$	24±28	734±0	48±426	12±41	477±3.32e3	33±0

The means and standard deviations of estimation error of copy number.

	CNV-seq	FREEC	readDepth	CNVnator	SegSeq	EWT
$l=0.8$ kbp, $n=6$ , $c=5$	$2.44\pm1.46$	$2.53\pm0.26$	$0.89\pm0.68$	NA	$3.46\pm1.08$	$1.38\pm0.93$
$l=2$ kbp, $n=6$ , $c=5$	$1.61\pm0.22$	$2.3\pm0.51$	$1.23\pm1.08$	$0.85\pm0.22$	$2.86\pm1.07$	$1.30\pm0.79$
$l=6$ kbp, $n=6$ , $c=5$	$0.87\pm0.19$	$1.56\pm0.53$	$0.82\pm0.78$	$0.50\pm0.08$	$2.46\pm1.56$	$1.10\pm0.93$
$l=6$ kbp, $n=0$ , $c=5$	$0.15\pm0.06$	$1.12\pm1.41$	$0.06\pm0.01$	$0.02\pm0.01$	$1.73\pm0.70$	$1.27\pm2.08$
$l=6$ kbp, $n=1$ , $c=5$	$0.14\pm0.05$	$0.15\pm0.53$	$0.02\pm0.13$	$0.17\pm0.05$	$0.49\pm0.46$	$0.36\pm0.89$
$l=6$ kbp, $n=3$ , $c=5$	$0.21\pm0.08$	$0.00\pm0.00$	$0.46\pm0.99$	$0.12\pm0.08$	$0.63\pm0.59$	$0.03\pm0.30$
$l=6$ kbp, $n=6$ , $c=3$	$1.12\pm0.23$	$1.76\pm0.48$	NA	$0.45\pm0.14$	$2.43\pm1.33$	$1.13\pm0.82$
$l=6$ kbp, $n=6$ , $c=10$	$0.74\pm0.14$	$1.69\pm0.32$	$0.66\pm0.45$	$0.49\pm0.07$	$3.27\pm1.47$	$0.89\pm0.96$
$l=6$ kbp, $n=6$ , $c=30$	$0.73\pm0.08$	$1.96\pm0.30$	$0.84\pm0.57$	$0.70\pm0.04$	$4.10\pm0.69$	$1.24\pm0.90$