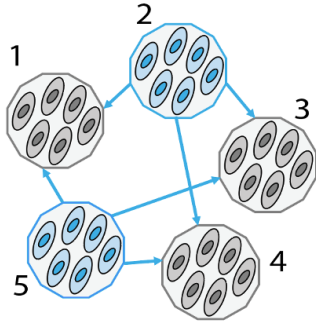


Number of cells (N) = 5

Number of times variant is seen (n) = 6

$$f \approx 0.5 - \sqrt{0.25 - n/N^2} = 0.4$$



Mosaic mutation example

Calculation of mosaic score:

Number of cells carrying mutation (N') = $f * N = 0.4 * 5 = 2$

So, we take the 2 rows with maximum hits and count it

$$n_m = \sum_{i=1,2} nr_i = \sum_{i=1,5} nr_i = \sum_{i=2,3} nr_i = \sum_{i=2,4} nr_i = \sum_{i=3,4} nr_i = \sum_{i=3,5} nr_i = 3$$

$$n_m = \sum_{i=1,3} nr_i = \sum_{i=1,4} nr_i = \sum_{i=3,4} nr_i = 0$$

$$n_m = \sum_{i=2,5} nr_i = 6 \quad \text{We take the maximum possible } n_m$$

	1	2	3	4	5
1					
2					
3					
4					
5					

$$\text{Mosaic score} = n_m/n = \frac{6}{6} = 1$$

Calculation of germline score:

Number of cells not carrying germline variant (N') = $f * N = 0.4 * 5 = 2$

So, we take the 2 columns with maximum hits and count it

$$n_g = \sum_{i=1,2} nc_i = \sum_{i=1,5} nc_i = \sum_{i=2,3} nc_i = \sum_{i=2,4} nc_i = \sum_{i=3,5} nc_i = \sum_{i=4,5} nc_i = 2$$

$$n_g = \sum_{i=2,5} nc_i = 0$$

$$n_g = \sum_{i=1,3} nc_i = \sum_{i=1,4} nc_i = \sum_{i=3,4} nc_i = 4 \quad \text{We take the maximum possible } n_g$$

$$\text{Germline score} = n_g/n = \frac{4}{6} = 0.6$$

Fig S2. Example of calculating mosaic and germline scores for a mosaic variant.