

Supplemental Material

Justification of Equation 1

The Poisson distribution describes the probability of a droplet containing k number of molecules where the average number of molecules per droplet is λ :

$$P(k; \lambda) = \frac{\lambda^k e^{-\lambda}}{k!}.$$

The average number of molecules per droplet, called the “occupancy” throughout the text, can be easily found from the fraction of empty droplets as follows. If P and N are the numbers of PCR(+) and PCR(-) droplets respectively, the probability of an empty droplet is

$$P(0; \lambda) = \frac{N}{N + P}.$$

Combining the two equations above,

$$\left[P(0; \lambda) = \frac{\lambda^0 e^{-\lambda}}{0!} = e^{-\lambda} \right] = \frac{N}{N + P}.$$

Solve for λ :

$$e^{-\lambda} = \frac{N}{N + P}$$
$$\lambda = \ln\left(\frac{N + P}{N}\right).$$

This equation for calculating occupancy works best when the fraction of empty droplets is high, typically at least $\frac{1}{2}$ (Pohl & Shih, 2004).

<i>Patient number</i>	<i>Coriell cell line</i>	<i>Patient number, cont'd.</i>	<i>Coriell cell line, cont'd.</i>
1	NA14638	11	NA13714
2	NA14637	12	NA13712
3	NA14097	13	NA13709
4	NA14096	14	NA13707
5	NA14094	15	NA13705
6	NA14093	SMA carrier	NA03814
7	NA14092	SMA 1	NA03813
8	NA14091	SMA 2	NA00232
9	NA14090	SMA 3	NA09677
10	NA13715	SMA 4	NA10684

Supplementary Table 1 Map of patient numbers used in the text to Coriell cell lines.

Supplementary Reference

G. Pohl and le-M. Shih, *Expert Rev Mol Diagn.*, 2004, **4**, 41-47.