Continuous Distributions

1.8-1.9: Continuous Random Variables

1.10.1: Uniform Distribution (Continuous)

1.10.4-5 Exponential and Gamma Distributions: Distance between crossovers

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Continuous distributions

Example

- Pick a real number x between 20 and 30 with all real values in $[20, 30]$ equally likely.
- Sample space: $S = [20, 30]$
- Number of outcomes: $|S| = \infty$
- Probability of each outcome: $P(X = x) = \frac{1}{\infty} = 0$
- Yet, $P(X \leqslant 21.5) = 15\%$

Continuous distributions

- The *sample space* S is often a subset of $\mathbb{R}^n$.
  We'll do the 1-dimensional case $S \subset \mathbb{R}$.
- The *probability density function (pdf)* $f_X(x)$ is defined differently than the discrete case:
  - $f_X(x)$ is a real-valued function on S with $f_X(x) \geqslant 0$ for all $x \in S$.
  - $\int_S f_X(x)\,dx = 1 \quad$ (vs. $\sum_{x \in S} P_X(x) = 1$ for discrete)
  - The probability of event $A \subset S$ is $P(A) = \int_A f_X(x)\,dx \quad$ (vs. $\sum_{x \in A} P_X(x)$).
  - In n dimensions, use n-dimensional integrals instead.

Uniform distribution

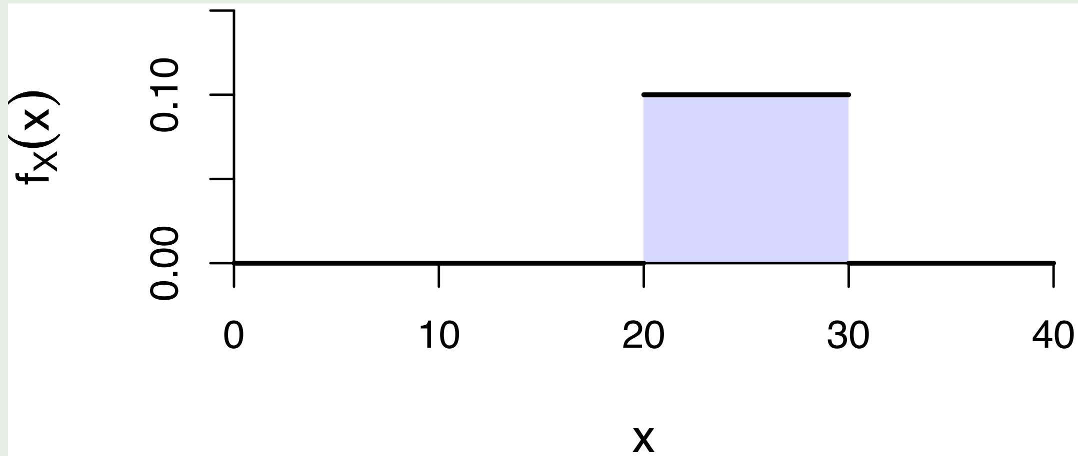
- Let $a < b$ be real numbers.
- The *Uniform Distribution* on $[a, b]$ is that all numbers in $[a, b]$ are "equally likely."
- More precisely, $f_X(x) = \begin{cases} \frac{1}{b-a} & \text{if } a \leqslant x \leqslant b; \\ 0 & \text{otherwise.} \end{cases}$

Uniform distribution (real case)

The uniform distribution on $[20, 30]$

We could regard the sample space as $[20, 30]$, or as all reals.

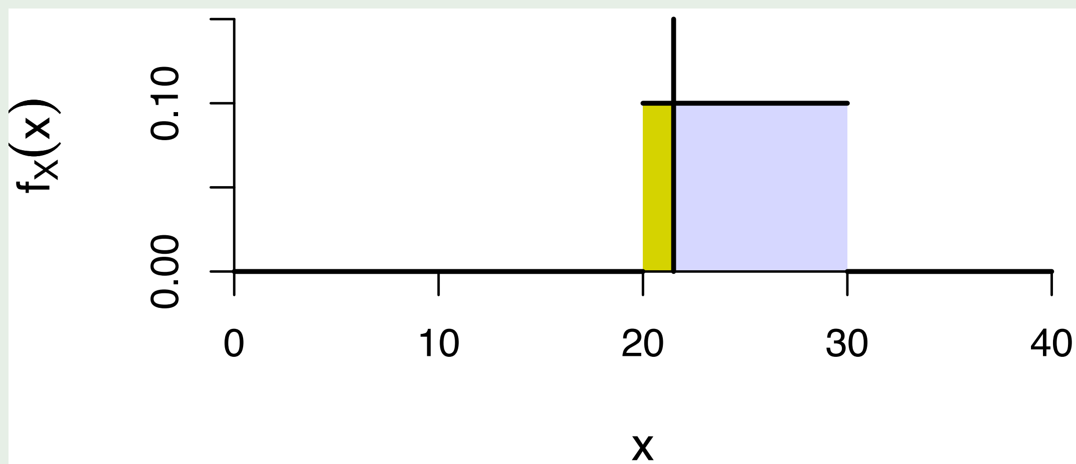
$$f_X(x) = \begin{cases} 1/10 & \text{for } 20 \leqslant x \leqslant 30; \\ 0 & \text{otherwise.} \end{cases}$$



$$P(X \leqslant 21.5) = \int_{-\infty}^{20} 0\,dx + \int_{20}^{21.5} \frac{1}{10}dx = 0 + \left.\frac{x}{10}\right|_{20}^{21.5}$$

$$= \frac{21.5 - 20}{10}$$

$$= .15 = 15\%$$



Cumulative distribution function (cdf)

The *Cumulative Distribution Function (cdf)* of a random variable X is

$$F_X(x) = P(X \leqslant x)$$

For a continuous random variable,

$$F_X(x) = P(X \leqslant x) = \int_{-\infty}^{x} f_X(t)\,dt \qquad \text{and} \qquad f_X(x) = F_X{}'(x)$$

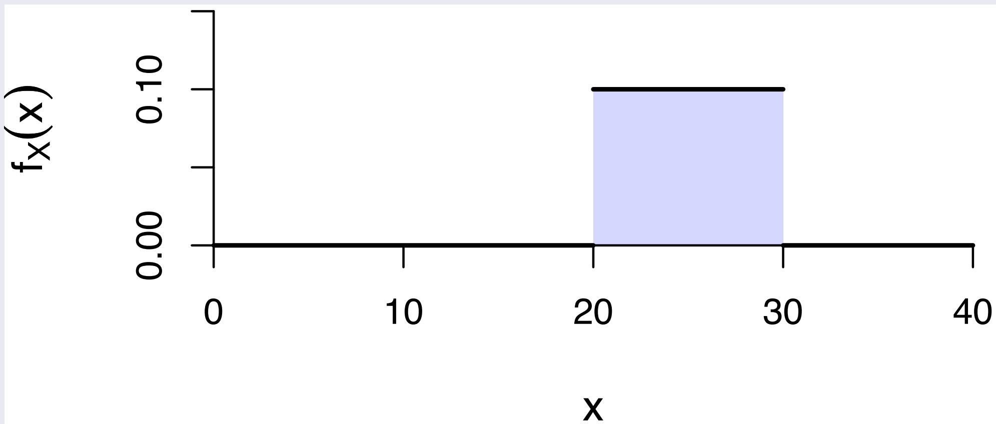
Uniform distribution on $[20, 30]$

- For $x < 20$: $\quad F_X(x) = \int_{-\infty}^{x} 0\,dt = 0$
- For $20 \leqslant x < 30$: $\quad F_X(x) = \int_{-\infty}^{20} 0\,dt + \int_{20}^{x} \frac{1}{10}dt = \frac{x-20}{10}$
- For $30 \leqslant x$: $\quad F_X(x) = \int_{-\infty}^{20} 0\,dt + \int_{20}^{30} \frac{1}{10}\,dt + \int_{30}^{x} 0\,dt = 1$
- Together:

$$F_X(x) = \begin{cases} 0 & \text{if } x < 20 \\ \frac{x-20}{10} & \text{if } 20 \leqslant x \leqslant 30 \\ 1 & \text{if } x \geqslant 30 \end{cases} \qquad f_X(x) = F_X{}'(x) = \begin{cases} 0 & \text{if } x < 20 \\ \frac{1}{10} & \text{if } 20 \leqslant x \leqslant 30 \\ 0 & \text{if } x \geqslant 30 \end{cases}$$

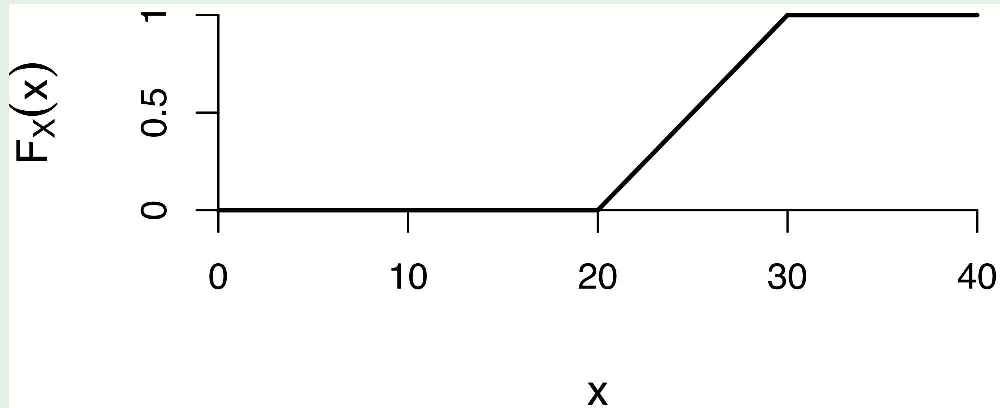
PDF vs. CDF

Probability density function



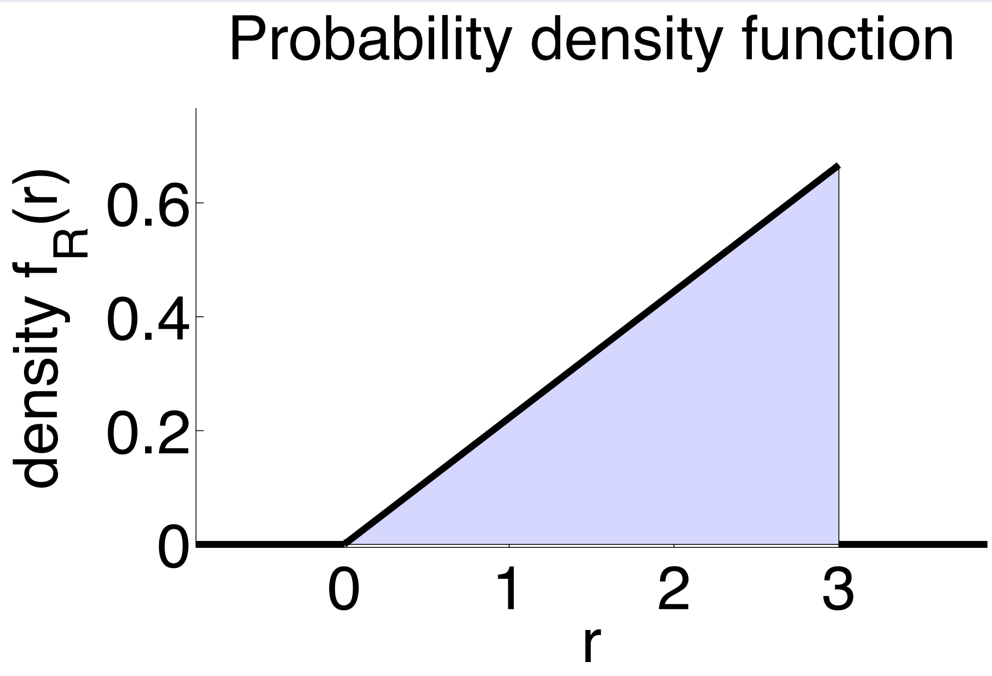
- $f_X(x) = \begin{cases} .1 & \text{if } 20 \leqslant x \leqslant 30; \\ 0 & \text{otherwise.} \end{cases}$
  It's discontinuous at $x = 20$ and 30.
- **PDF is derivative of CDF:**
  $f_X(x) = F_X('x)$

Cumulative distribution function



- $F_X(x) =$
  $\begin{cases} 0 & \text{if } x < 20; \\ (x-20)/10 & \text{if } 20 \leqslant x \leqslant 30; \\ 1 & \text{if } x \geqslant 30. \end{cases}$
- **CDF is integral of PDF:**
  $$F_X(x) = \int_{-\infty}^{x} f_X(t)\,dt$$

PDF vs. CDF: Second example

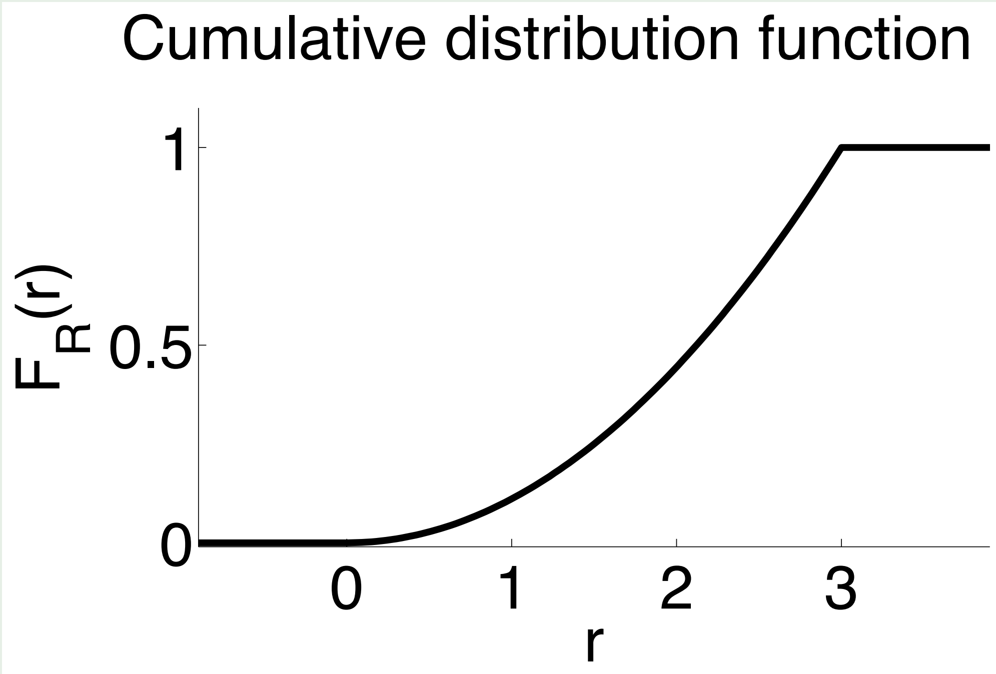


- $f_R(r) = \begin{cases} 2r/9 & \text{if } 0 \leqslant r < 3; \\ 0 & \text{if } r \leqslant 0 \text{ or } r > 3 \end{cases}$

  It's discontinuous at $r = 3$.

- **PDF is derivative of CDF:**

  $f_R(r) = F_R{}'(r)$



- $F_R(r) = \begin{cases} 0 & \text{if } r < 0; \\ r^2/9 & \text{if } 0 \leqslant r \leqslant 3; \\ 1 & \text{if } r \geqslant 3. \end{cases}$

- **CDF is integral of PDF:**

  $F_R(r) = \int_{-\infty}^{r} f_R(t)\,dt$

Probability of an interval

Compute $P(-1 \leqslant R \leqslant 2)$ from the PDF and also from the CDF

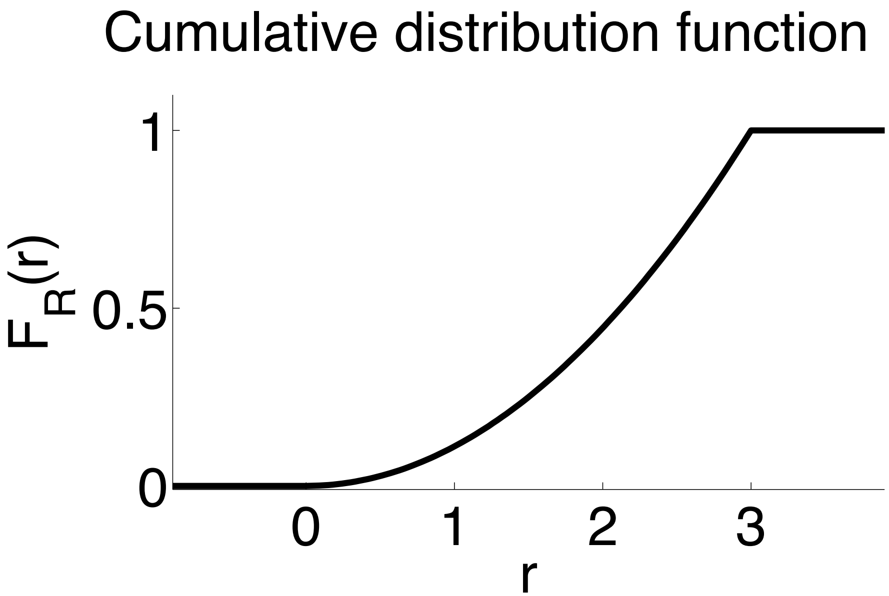
Computation from the PDF

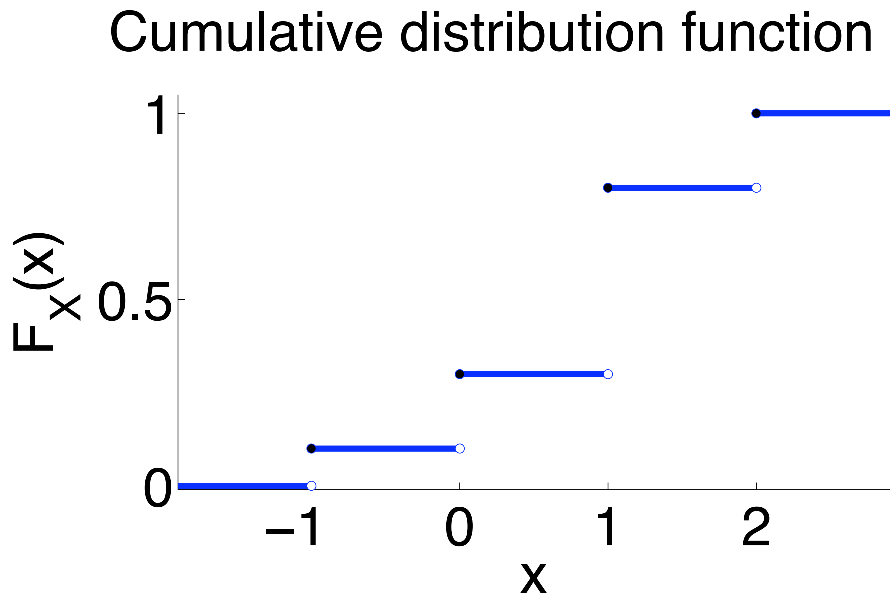
$$\begin{aligned}
P(-1 \leqslant R \leqslant 2) &= \int_{-1}^{2} f_R(r)\,dr = \int_{-1}^{0} f_R(r)\,dr + \int_{0}^{2} f_R(r)\,dr \\
&= \int_{-1}^{0} 0\,dr + \int_{0}^{2} \frac{2r}{9}\,dr \\
&= 0 + \left(\left. \frac{r^2}{9} \right|_{r=0}^{2} \right) = \frac{2^2 - 0^2}{9} = \boxed{\mathbf{\frac{4}{9}}}
\end{aligned}$$

Computation from the CDF

$$\begin{aligned}
P(-1 \leqslant R \leqslant 2) &= P(-1^- < R \leqslant 2) \\
&= F_R(2) - F_R(-1^-) = \frac{2^2}{9} - 0 = \boxed{\mathbf{\frac{4}{9}}}
\end{aligned}$$

Continuous vs. discrete random variables





In a continuous distribution:

- The probability of an individual point is 0: $P(R = r) = 0$.
  So, $P(R \leqslant r) = P(R < r)$, i.e., $F_R(r) = F_R(r^-)$.
- The CDF is continuous.
  (In a discrete distribution, the CDF is discontinuous due to jumps at the points with nonzero probability.)
- $$P(a < R < b) = P(a \leqslant R < b) = P(a < R \leqslant b) = P(a \leqslant R \leqslant b) = F_R(b) - F_R(a)$$

Cumulative distribution function (cdf)

The *Cumulative Distribution Function (cdf)* of a random variable X is

$$F_X(x) = P(X \leqslant x)$$

Continuous case

- $F_X(x) = \int_{-\infty}^{x} f_X(t)\,dt$
- Weakly increasing.
- Varies smoothly from 0 to 1 as x varies from $-\infty$ to ∞.
- To get the pdf from the cdf, use $f_X(x) = F_X{}'(x)$.

Discrete case

- $F_X(x) = \sum_{t \leqslant x} P_X(t)$
- Weakly increasing.
- Stair-steps from 0 to 1 as x goes from $-\infty$ to ∞.
- The cdf jumps where $P_X(x) \neq 0$ and is constant in-between.
- To get the pdf from the cdf, use $P_X(x) = F_X(x) - F_X(x^-)$ (which is positive at the jumps, 0 otherwise).

CDF, percentiles, and median

The *kth percentile* of a distribution X is the point x where k% of the probability is up to that point:

$$F_X(x) = P(X \leqslant x) = k\% = k/100$$

Example: $F_R(r) = P(R \leqslant r) = r^2/9 \quad$ (for $0 \leqslant r \leqslant 3$)

- $r^2/9 = (k/100) \quad \Rightarrow \quad r = \sqrt{9(k/100)}$
- 75th percentile: $r = \sqrt{9(.75)} \approx 2.60$
- Median (50th percentile): $r = \sqrt{9(.50)} \approx 2.12$
- 0th and 100th percentiles:
  $r = 0$ and $r = 3$ if we restrict to the range $0 \leqslant r \leqslant 3$.

  But they are not uniquely defined, since
  $F_R(r) = 0$ for all $r \leqslant 0$ and $F_R(r) = 1$ for all $r \geqslant 3$.

Expected value and variance (continuous r.v.)

Replace sums by integrals. It's the same definitions in terms of "$E(\cdot)$":

$$\mu = E(X) = \int_{-\infty}^{\infty} x \cdot f_X(x)\,dx$$

$$E(g(X)) = \int_{-\infty}^{\infty} g(x) f_X(x)\,dx$$

$$\begin{aligned}\sigma^2 &= \mathrm{Var}(X)\\ &= E((X-\mu)^2) = E(X^2) - (E(X))^2\end{aligned}$$

μ and σ for the uniform distribution on $[a,b]$ (with $a < b$)

$$\mu = E(X) = \int_a^b x \cdot \frac{1}{b-a}\,dx = \left.\frac{x^2/2}{b-a}\right|_{x=a}^{b} = \frac{(b^2-a^2)/2}{b-a} = \frac{b+a}{2}$$

$$E(X^2) = \int_a^b x^2 \cdot \frac{1}{b-a}\,dx = \left.\frac{x^3/3}{b-a}\right|_{x=a}^{b} = \frac{(b^3-a^3)/3}{b-a} = \frac{b^2+ab+a^2}{3}$$

$$\sigma^2 = \mathrm{Var}(X) = E(X^2) - (E(X))^2 = \frac{b^2+ab+a^2}{3} - \left(\frac{b+a}{2}\right)^2 = \frac{(b-a)^2}{12}$$

$$\sigma = \mathrm{SD}(X) = (b-a)/\sqrt{12}$$

Exponential distribution

- How far is it from the start of a chromosome to the first crossover?
- How far is it from one crossover to the next?
- Let D be the random variable giving either of those. It is a real number > 0, with the *exponential distribution*

$$f_D(d) = \begin{cases} \lambda\, e^{-\lambda\, d} & \text{if } d \geqslant 0; \\ 0 & \text{if } d < 0. \end{cases}$$

  where crossovers happen at a rate $\lambda = 1$ M^{-1} $= 0.01$ cM^{-1}.

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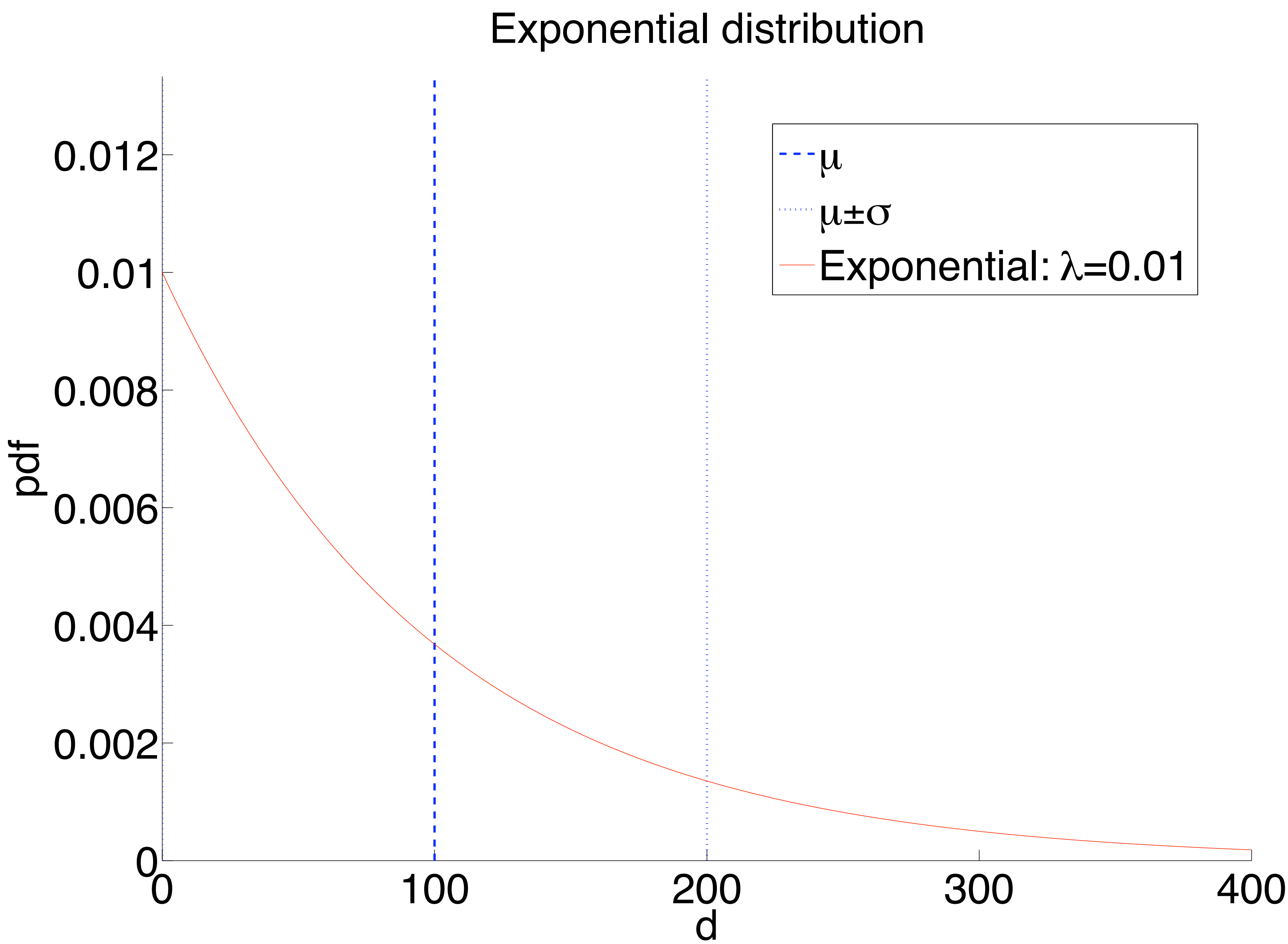
	General case	**Crossovers**
Mean	$E(D) = 1/\lambda$	$= 100$ cM $= 1$ M
Variance	$\mathrm{Var}(D) = 1/\lambda^2$	$= 10000$ cM^2 $= 1$ M^2
Standard Dev.	$\mathrm{SD}(D) = 1/\lambda$	$= 100$ cM $= 1$ M

Exponential distribution

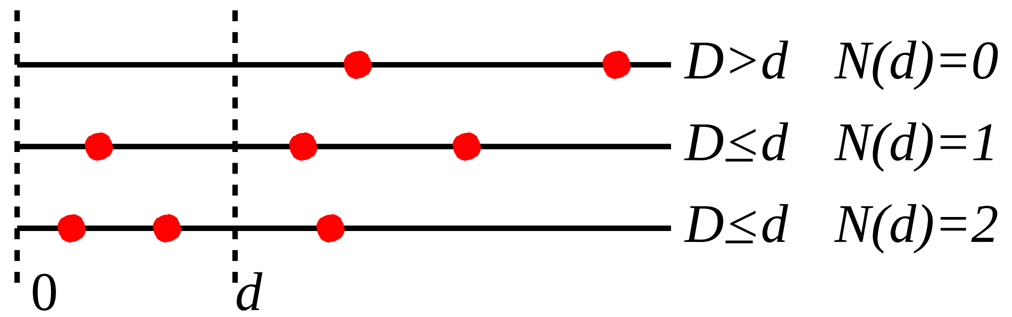


Exponential distribution

- In general, if events occur on the real number line $x \geqslant 0$ in such a way that the expected number of events in all intervals $[x, x+d]$ is $\lambda\, d$ (for $x > 0$), then the exponential distribution with parameter λ models the time/distance/etc. until the first event.
- It also models the time/distance/etc. between consecutive events.
- Chromosomes are finite; to make this model work, treat "there is no next crossover" as though there is one but it happens somewhere past the end of the chromosome.

Proof of pdf formula

- Let $d > 0$ be any real number.
- Let $N(d)$ be the # of crossovers that occur in the interval $[0, d]$.



  - If $N(d) = 0$ then there are no crossovers in $[0, d]$, so $D > d$.
  - If $D > d$ then the first crossover is after d so $N(d) = 0$.
  - Thus, $D > d$ is equivalent to $N(d) = 0$.
- $P(D > d) = P(N(d) = 0) = e^{-\lambda d}(\lambda d)^0/0! = e^{-\lambda d}$
  since $N(d)$ has a Poisson distribution with parameter λd.
- The cdf of D is

$$F_D(d) = P(D \leqslant d) = 1 - P(D > d) = \begin{cases} 1 - e^{-\lambda d} & \text{if } d \geqslant 0; \\ 0 & \text{if } d < 0. \end{cases}$$

- Differentiating the cdf gives pdf $f_D(d) = F_D{}'(d) = \lambda e^{-\lambda d}$ (if $d \geqslant 0$).

Discrete and Continuous Analogs

	Discrete	**Continuous**
"Success"	Coin flip at a position is heads	Point where crossover occurs
Rate	Probability p per flip	λ (crossovers per Morgan)
# successes	Binomial distribution: # heads out of n flips	Poisson distribution: # crossovers in distance d
Wait until 1st success	Geometric distribution	Exponential distribution
Wait until rth success	Negative binomial distribution	Gamma distribution

Gamma distribution

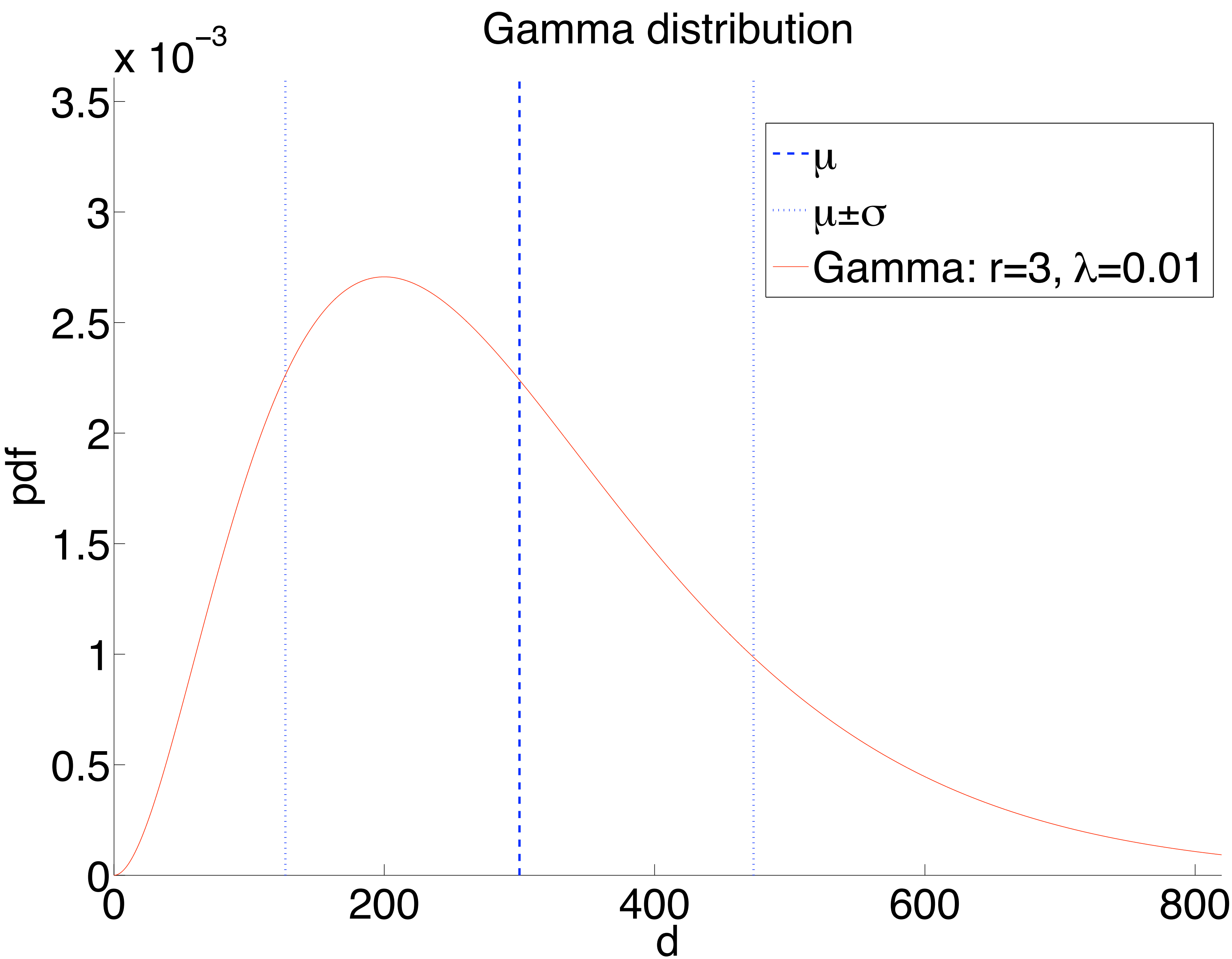
- How far is it from the start of a chromosome until the rth crossover, for some choice of $r = 1, 2, 3, \ldots$?
- Let D_r be a random variable giving this distance.
- It has the *gamma distribution* with pdf

$$f_{D_r}(d) = \begin{cases} \frac{\lambda^r}{(r-1)!} d^{r-1} e^{-\lambda d} & \text{if } d \geqslant 0; \\ 0 & \text{if } d < 0. \end{cases}$$

Mean	$E(D_r) = r/\lambda$
Variance	$\text{Var}(D_r) = r/\lambda^2$
Standard deviation	$\text{SD}(D_r) = \sqrt{r}/\lambda$

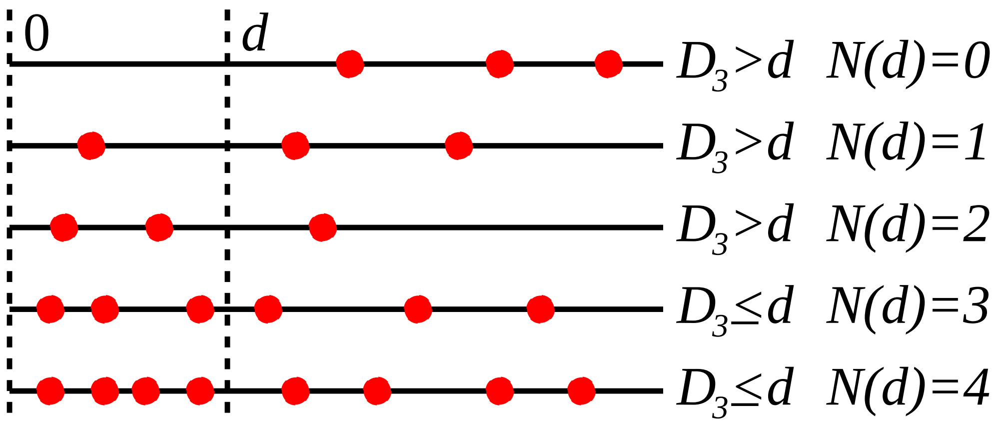
- The gamma distribution for $r = 1$ is the same as the exponential distribution.
- The sum of r i.i.d. exponential variables, $D_r = X_1 + X_2 + \cdots + X_r$, each with rate λ, gives the gamma distribution.

Gamma distribution



Proof of Gamma distribution pdf for $r = 3$

- Let $d > 0$ be any real number.
- $D_3 > d$ is the event that the third crossover does not happen until sometime after position d.



- When $D_3 > d$, the number $N(d)$ of crossovers in the chromosome interval $[0, d]$ is less than 3, so it's 0, 1, or 2.
  $D_3 > d$ is equivalent to $N(d) < 3$.
  $D_3 \leqslant d$ is equivalent to $N(d) \geqslant 3$.

Proof of Gamma distribution pdf for $r = 3$

- Let $d > 0$ be any real number.
- $D_3 > d$ is the event that the third crossover does not happen until sometime after position d.
- When $D_3 > d$, the number $N(d)$ of crossovers in the chromosome interval $[0, d]$ is less than 3, so it's 0, 1, or 2:

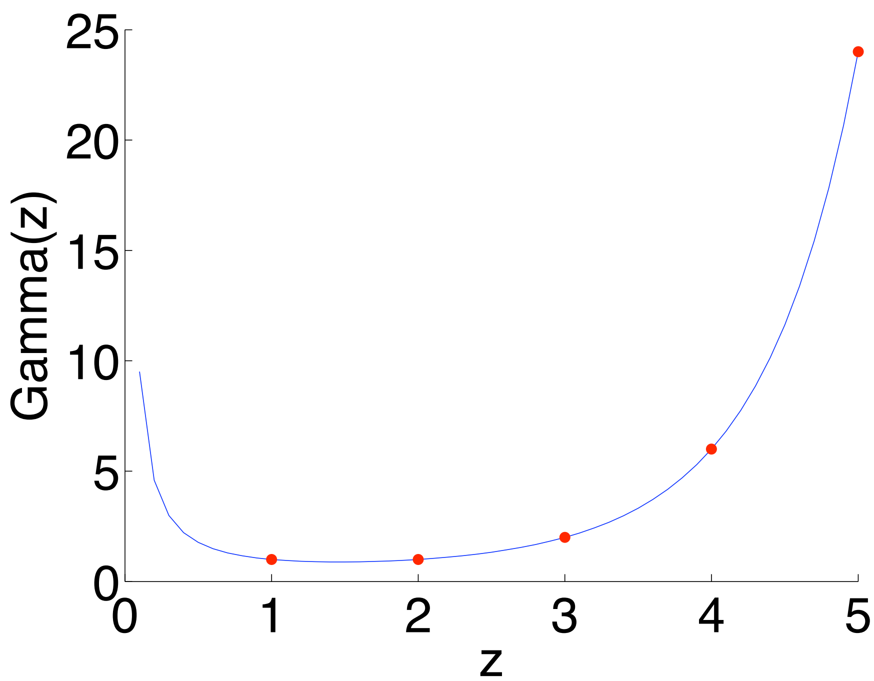
$$\begin{aligned} P(D_3 > d) = & \ P(N(d)=0) + P(N(d)=1) + P(N(d)=2) \\ = & \ e^{-\lambda d}\left(\frac{(\lambda d)^0}{0!} + \frac{(\lambda d)^1}{1!} + \frac{(\lambda d)^2}{2!}\right) \end{aligned}$$

- The cdf of D_3 is $P(D_3 \leqslant d) = 1 - P(D_3 > d)$.
- Differentiating the cdf and simplifying gives the pdf

$$f_{D_3}(d) = \begin{cases} \lambda^3 d^2 e^{-\lambda d}/2! & \text{if } d \geqslant 0; \\ 0 & \text{if } d < 0. \end{cases}$$

The Gamma function and factorials

- The *Gamma function* is a generalization of factorials:
$$\Gamma(z) = \int_0^\infty t^{z-1} e^{-t}\, dt$$
for real $z > 0$.
- $\Gamma(z) = (z-1)!$ for $z = 1, 2, 3, \ldots$
- $\Gamma(z)$ extends to all complex numbers except integers $\leqslant 0$.



$\Gamma(z) = (z-1)!$ for $z = 1, 2, 3, \ldots$.

- $\Gamma(1) = \displaystyle\int_0^\infty t^0 e^{-t}\, dt = -e^{-t}\Big|_0^\infty = -0 + 1 = 1$
- $\Gamma(z) = (z-1)\Gamma(z-1)$ can be shown using integration by parts: differentiate t^{z-1} and integrate up $e^{-t}\, dt$.
- When z is a positive integer, iterate this to
$\Gamma(z) = (z-1)(z-2)\cdots(2)(1)\Gamma(1) = (z-1)! \cdot \Gamma(1) = (z-1)!$ $\square$

Variations of the distributions

- The Gamma distribution is defined for real $r > 0$ rather than just positive integers:

$$f_{D_r}(d) = \begin{cases} \frac{\lambda^r}{\Gamma(r)} d^{r-1} e^{-\lambda d} & \text{if } d \geqslant 0; \\ 0 & \text{if } d < 0. \end{cases}$$

(The denominator $(r-1)!$ was replaced by $\Gamma(r)$.)

- For Poisson, Exponential, and Gamma distributions, instead of the rate parameter λ, some people use the *shape* parameter $\theta = 1/\lambda$:
  - For crossovers, $\theta = 1$ M $= 100$ cM.
  - The Poisson parameter for distance d is $\mu = \lambda d = d/\theta$.