POPULATION GENETICS OF THE CRITICALLY ENDANGERED SPOON-BILLED SANDPIPER

Anastasia Lyulina

Institute of Science and Technology Austria

Laboratory of Evolutionary Genomics

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photo by Gerrit Vyn

Wildlife is experiencing a global decline in population size, possibly on a way to a massive extinction event. Birds are not an exception, especially migratory birds.

RUSSIA

CHUKOTKA

KAMCHATKA

SOUTHEAST ASIA

EAST ASIAN–AUSTRALASIAN FLYWAY

100 – 200
breeding pairs

spoon-billed sandpiper

of least concern

red-necked stint

Pimm S.L. et al. *Science* 2014
Barnosky A.D. et al. *Nature* 2011
Bairlein F. *Science* 2016
Clark N.A. *Oryx* 2016

photo by Ekaterina Maksimova

Phylogeny of sequenced *Calidris species*

little stint — 1 genome sequenced

long-toed stint — 1 genome sequenced

red-necked stint — 9 genomes of unrelated individuals sequenced

spoon-billed sandpiper — 7 genomes of unrelated individuals sequenced

red knot — 1 genome sequenced

0.005
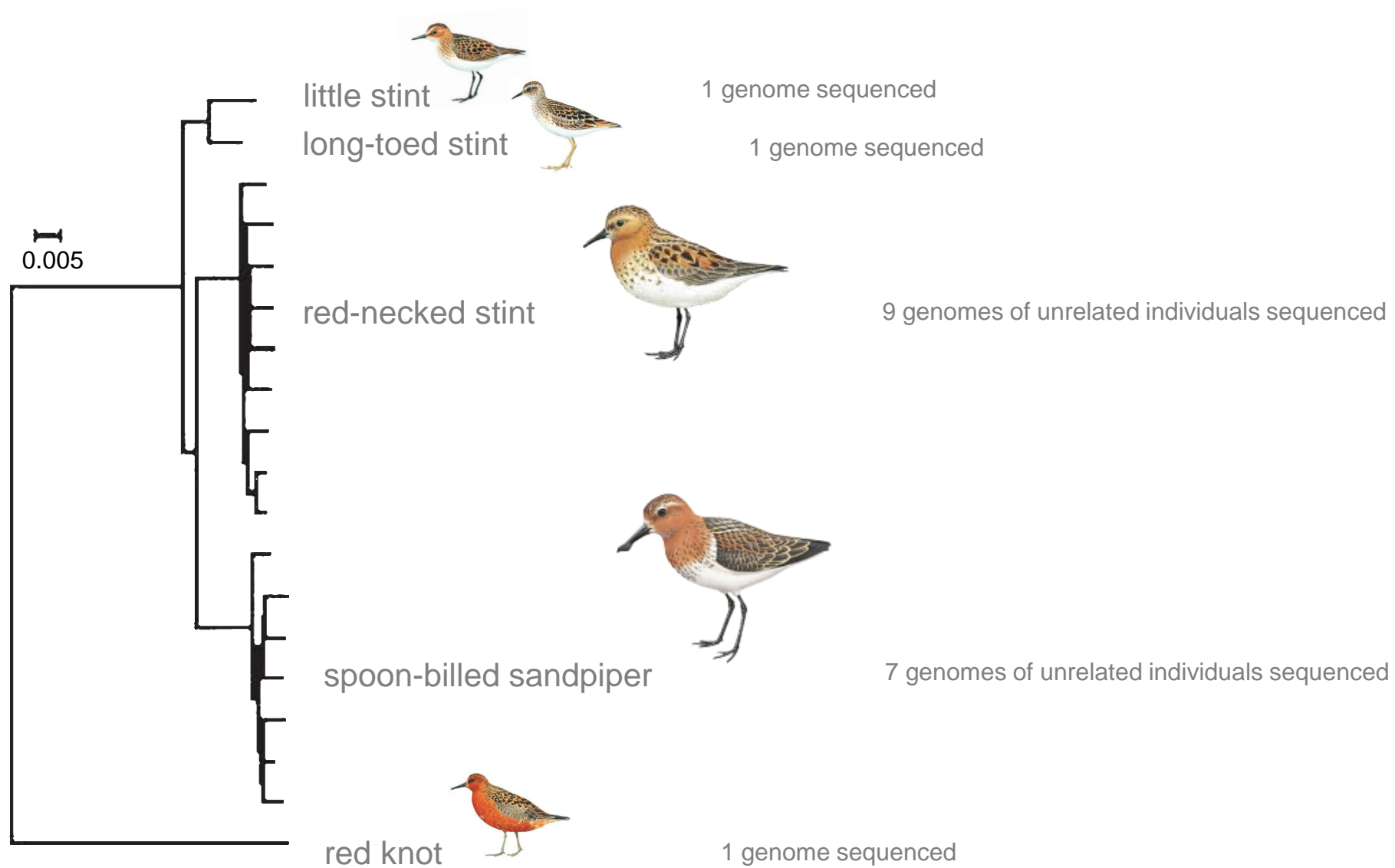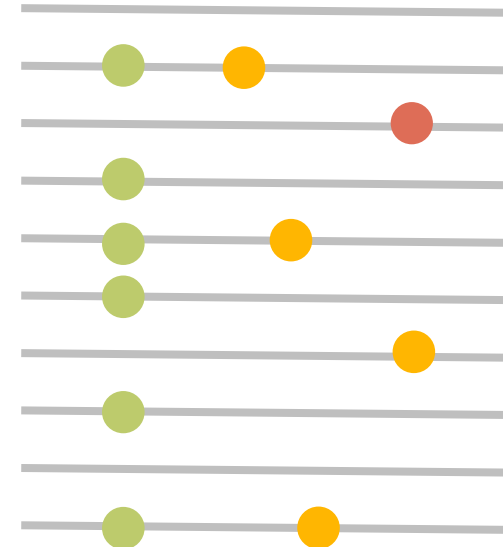
Demographic history reconstructed with PSMC
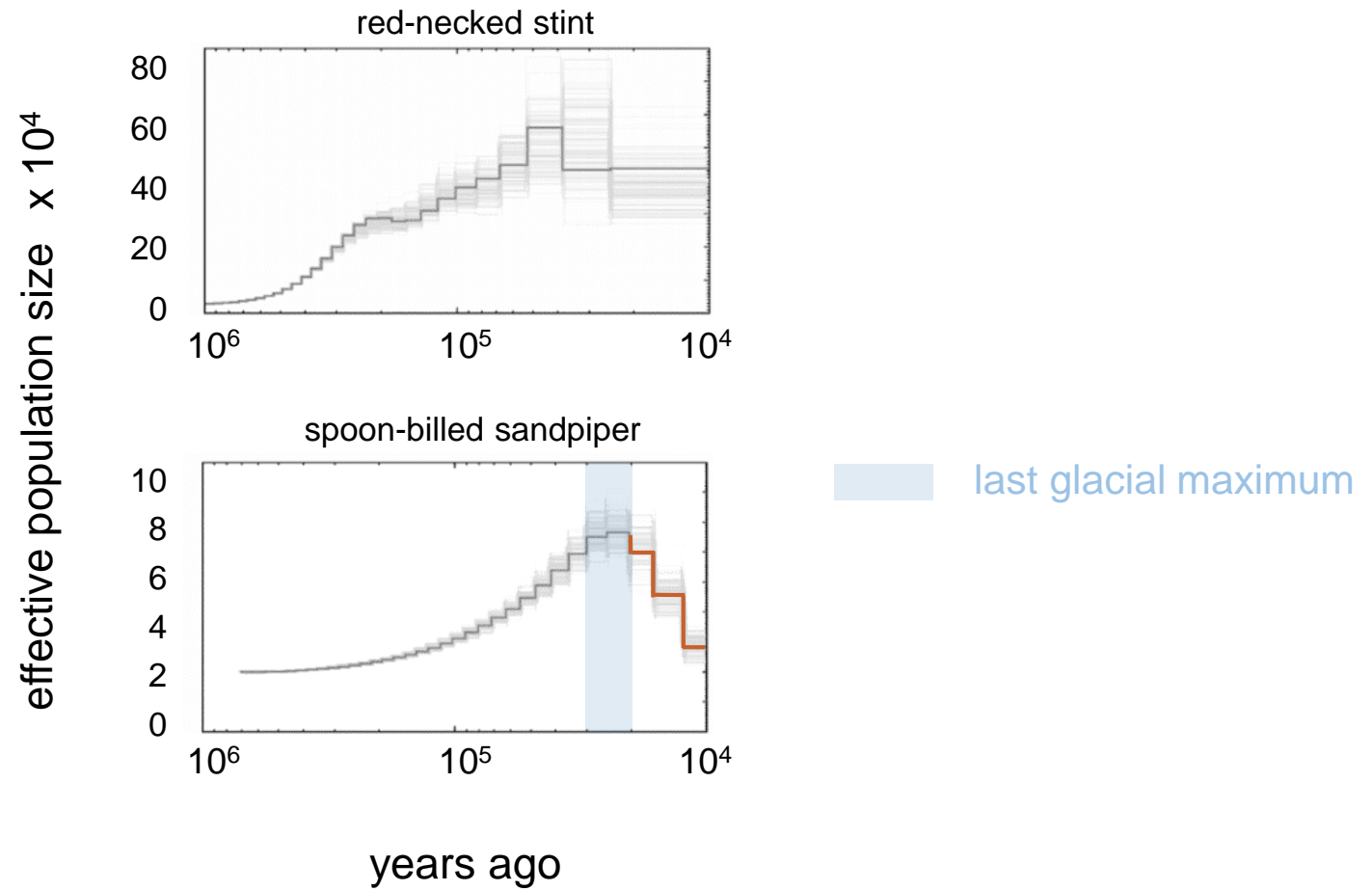
What happens when a population grows?

gain of rare alleles,
common ones are not affected

generation 1

generation *t*

What happens when a population goes through a bottleneck?

loss of rare alleles,
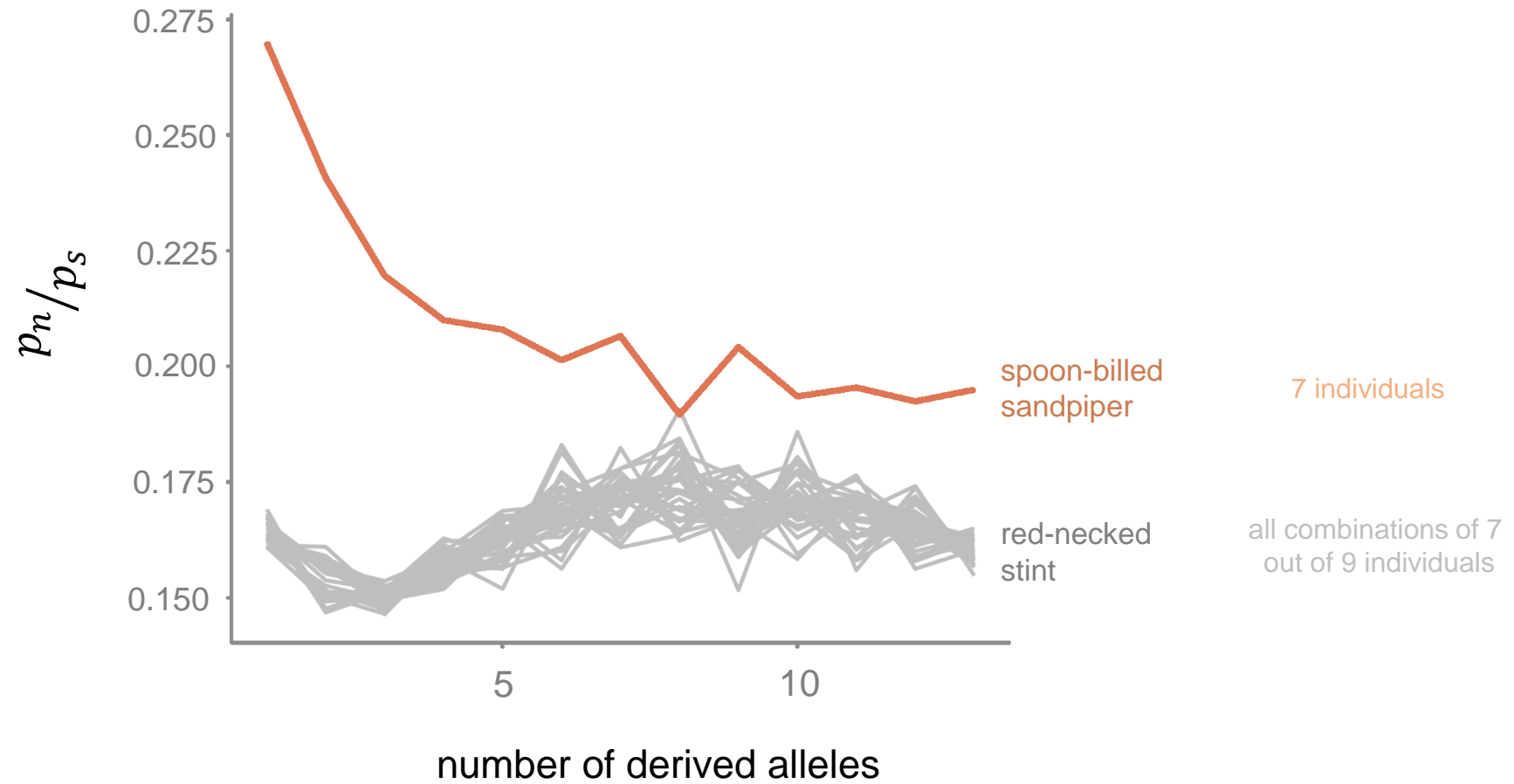common ones are not affected

generation 1

generation *t*

What do we see from the data?

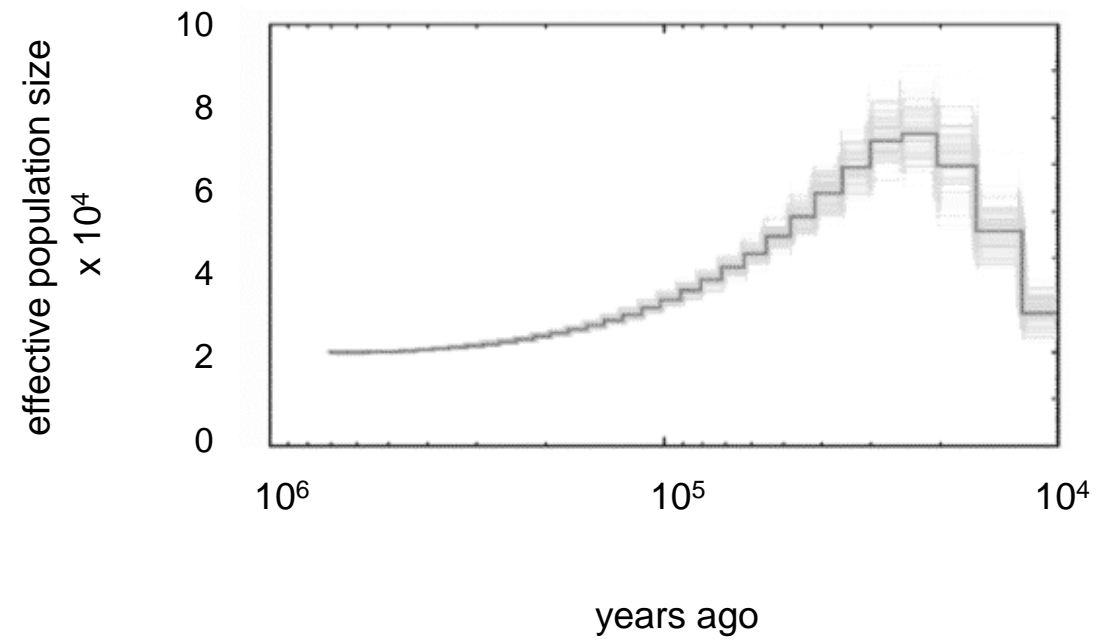# The overall level of nucleotide diversity indeed did not change much

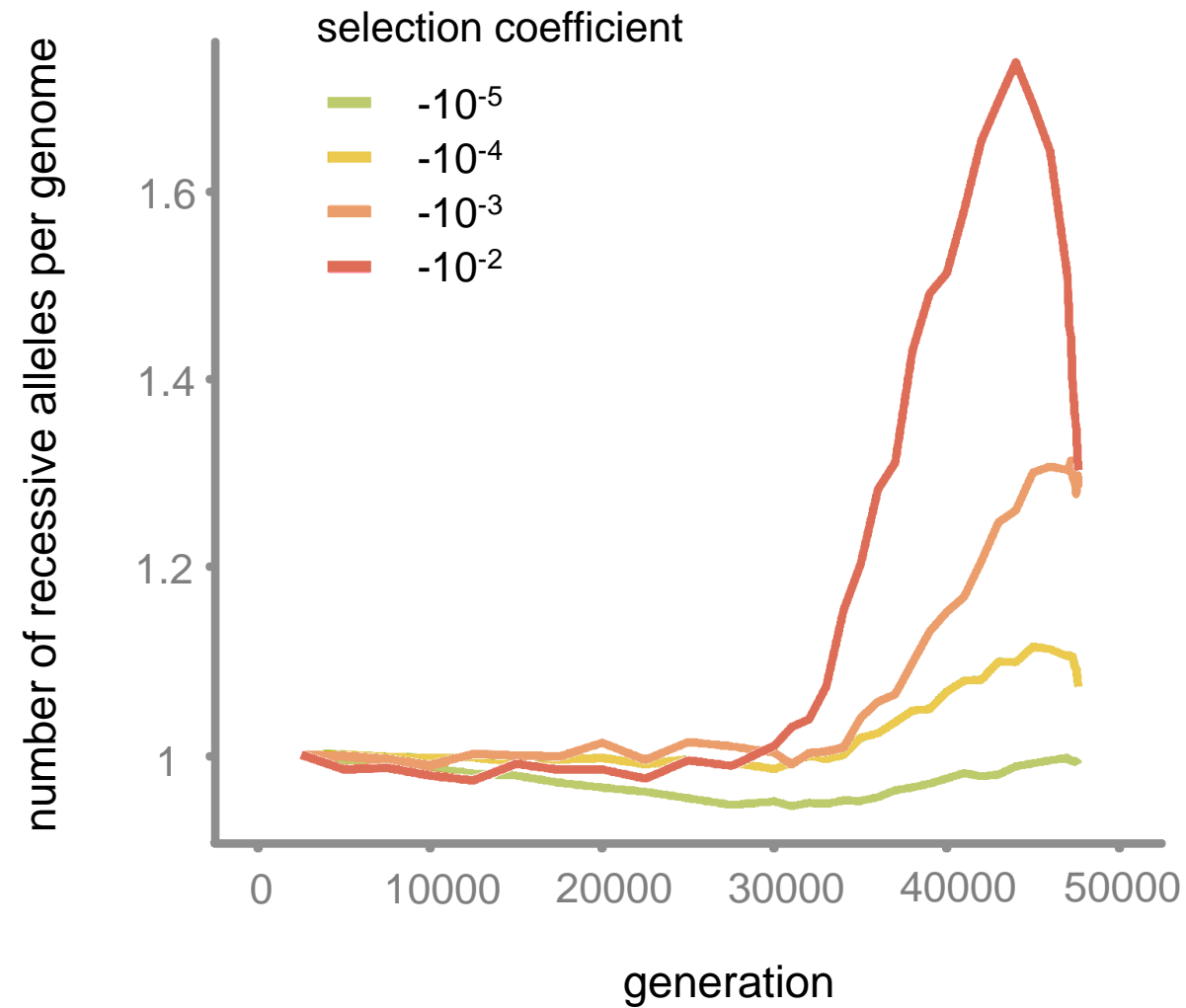# But the proportion of deleterious polymorphisms increased
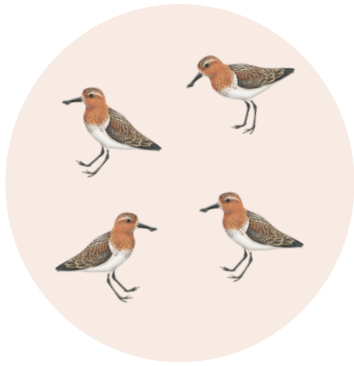
What happened with the spoon-billed sandpiper?

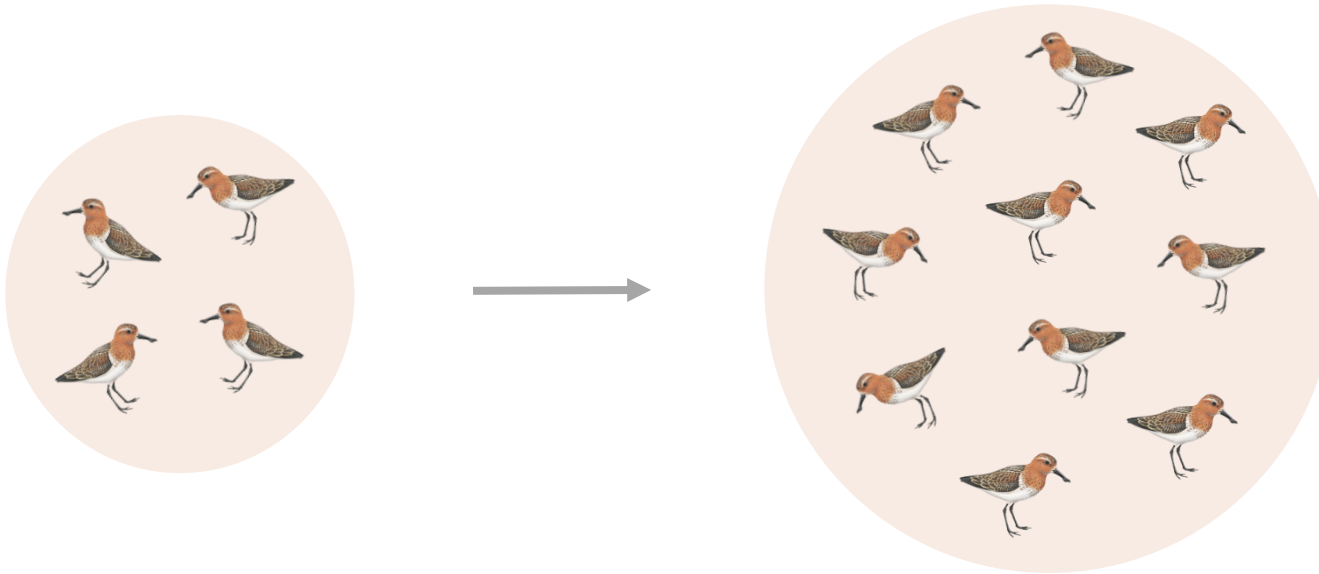# What happened with the spoon-billed sandpiper?
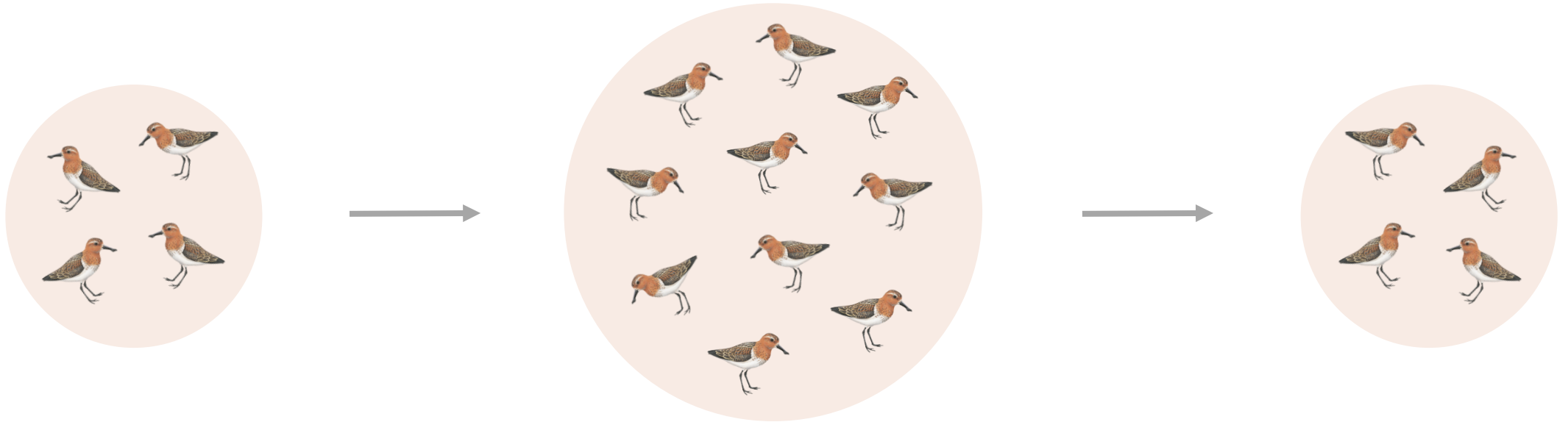
What happened with the spoon-billed sandpiper?

In a small population, genetic drift helps remove bad recessive variants.

What happened with the spoon-billed sandpiper?

When a population is growing, genetic drift stops, allowing to accumulate new deleterious variants.

What happened with the spoon-billed sandpiper?

When a population declines immediately after a population growth, it maintains some of the new recessive deleterious variants.

Can we find a distribution of selection coefficients
that explains what we see?
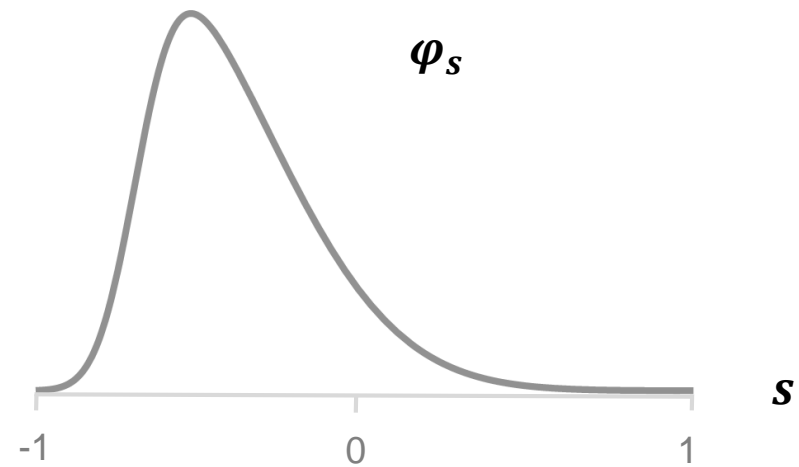
spoon-billed sandpiper

red-necked stint

φ_s

s

-1 0 1

probability density function of the distribution of
selection coefficients on new mutations

s	sh	0	relative fitness

$$subs(\varphi_s, N_e, h) = 2N_e ut \cdot \int_s \frac{\int_0^{\frac{1}{2N_e}} e^{2N_e s((2h-1)x^2 - 2hx)} dx}{\int_0^1 e^{2N_e s((2h-1)x^2 - 2hx)} dx} \cdot \varphi_s \, ds$$

number of mutations   fixation probability

x	allele frequency
s	selection coefficient
N_e	effective population size
h	dominance coefficient
u	mutation rate
t	number of generations

$\varphi_s (\mu, \sigma, \alpha)$ probability density function of the distribution of selection coefficients
which we model as a skew normal distribution
with location parameter μ, scale parameter σ, and shape parameter α

$$pols(\varphi_s, N_e, h) = \int\limits_{s} \int\limits_{0}^{1} 2x(1-x) \cdot \frac{1}{c_s} e^{2N_e s\left(x^2 + 2hx(1-x)\right)} x^{4N_e v - 1}(1-x)^{4N_e u - 1} \cdot \varphi_s \, dx ds$$

frequency of
heterozygotes

probability density function
of the distribution
of allele frequency x

probability density function
of the distribution
of selection coefficients s

x allele frequency

s selection coefficient

N_e effective population size

h dominance coefficient

u, v forward and backward mutation rates

c_s normalizing constant

$\varphi_s (\mu, \sigma, \alpha)$ probability density function of the distribution of selection coefficients
which we model as a skew normal distribution
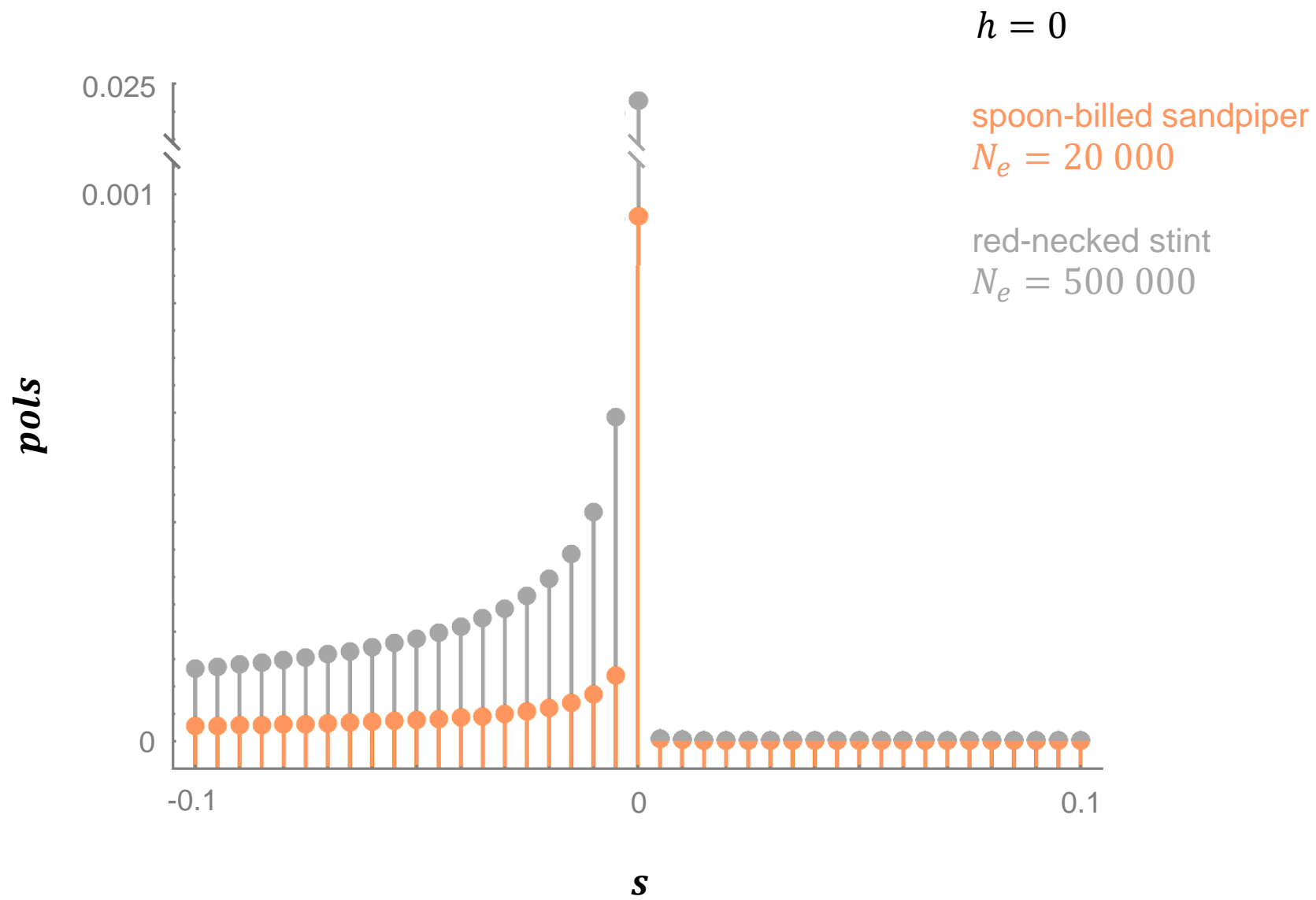with location parameter μ, scale parameter σ, and shape parameter α

$$pols(\varphi_s, N_e, h) = \int_S \int_0^1 2x(1-x) \cdot \frac{1}{c_s} e^{2N_e s\left(x^2 + 2hx(1-x)\right)} x^{4N_e v - 1} (1-x)^{4N_e u - 1} \cdot \varphi_s \, dx \, ds$$

$$c_s = \int_0^1 e^{2N_e s\left(x^2 + 2hx(1-x)\right)} x^{4N_e v - 1} (1-x)^{4N_e u - 1} dx$$
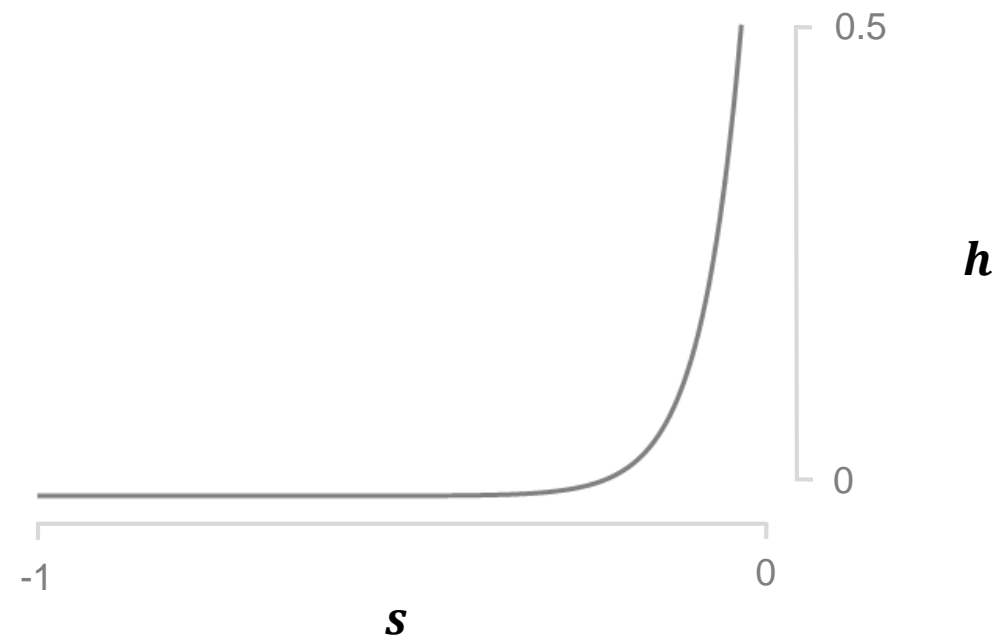
$$h = 0: \qquad \int_0^1 e^{2N_e s x^2} x^{4N_e v - 1} (1-x)^{4N_e u - 1} dx$$

$$h = \frac{1}{2}: \qquad \int_0^1 e^{2N_e s x} x^{4N_e v - 1} (1-x)^{4N_e u - 1} dx$$

$h = 0$

spoon-billed sandpiper
$N_e = 20\,000$

red-necked stint
$N_e = 500\,000$

$$h = \frac{e^{\beta s}}{2}$$

Deng H.W. and Lynch M. Genetics 1996
Balick D.J. et al. PLOS Genetics 2015

Fyodor Kondrashov

Mateusz Konczal

Luis Zapata, Francisco Camara, Anna Vlasova, Carla Bello,
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