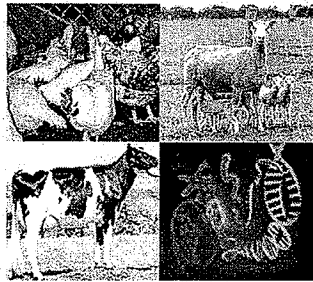


Genetics of small populations



Karen Marshall

UNE
THE UNIVERSITY
OF NEWCASTLE

IAEA, Korea, April, 2006

Issues relevant to small populations

Loss of genetic diversity

Characterizing population size (previous session)

Inbreeding (previous session)

Drift

Population fragmentation

Breeding strategies for conservation

Small populations

Loss of genetic diversity

Less genetic diversity → less ability to evolve in response to environmental change

Threats include:

- ❖ Extinction of populations or species
- ❖ Inbreeding resulting in less heterozygosity
- ❖ Loss of alleles due to sampling
- ❖ Selection leading to allele fixation

(Frankham, 2002)

The main reason for loss of genetic diversity is restricted effective population size (N_e) over a number of generations

Small populations

Creation of genetic diversity

Genetic variation can be created by

- ❖ New mutations
- ❖ New epistatic gene combinations

Small populations

Small populations

Small population size lead to inbreeding and drift

Inbreeding results from the joining of gametes with alleles that are IBD & leads to:

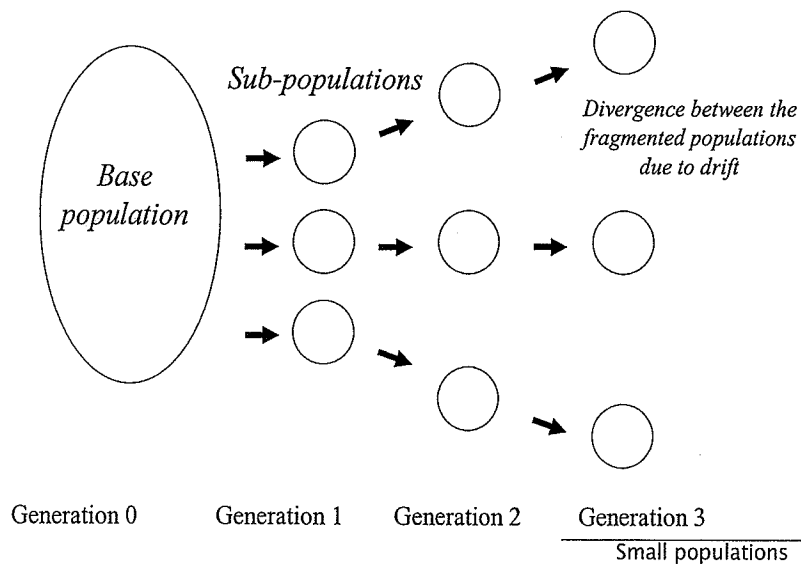
- ❖ Genetic similarity within small populations

Drift results from random sampling of gametes & leads to:

- ❖ Random change in allele frequencies
- ❖ Genetic divergence *between* fragmented populations

Small populations

Population fragmentation



Drift at a single locus level

At the single locus level, genetic divergence (drift) is the variance in allele frequency between sub-populations

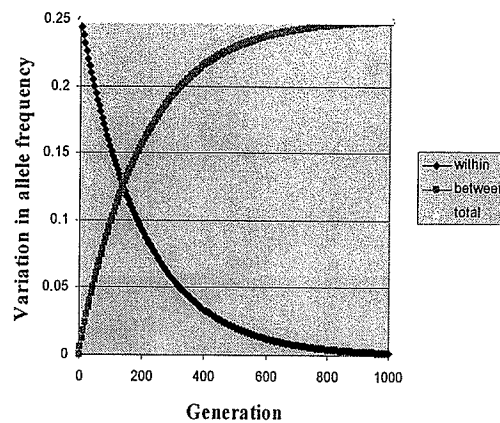
For example $p=q=0.5$, $N_{e(\text{sub-pop})}=10$

Generation	0	t = 1	t = 10	t = 100	t = infinity
$V_{q \text{ Between}}$	0	0.0125 ($pq/2N_e$)	0.10	0.2485	0.25 (pq^*)
$V_{q \text{ Within}}$	0.25 (pq^*)	0.2375	0.15	0.0015	0
$V_{q \text{ Total}}$	0.25	0.25	0.25	0.25	0.25

Small populations

Change in allele frequency due to drift

For example, $p=q=0.5$, $N_e(\text{sub-pop})=100$



Small populations

Drift increases trait variation across lines

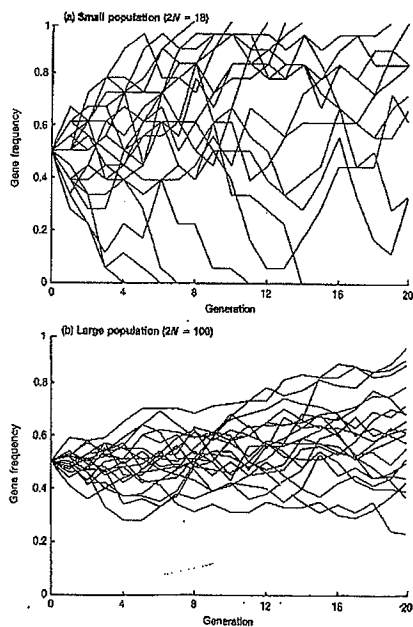
In previous graph, total variation in allele frequency remained stable

However, each sub-population is moving towards fixation (i.e. aa or AA, not aA), which results in an increase in trait variance

At the continuous trait level, genetic divergence (drift) is the variance between the trait means of the different sub-populations

❖ this can be predicted for traits of neutral fitness

Small populations



Bristle number for sub-populations of fruit fly.

Effect of population size (N) on the divergence and the rate of fixation of populations

<http://www.wooster.edu/biology/mloveless/Drift.html>

Small populations

Effects of drift and F following population fragmentation

Sub-populations will become divergent due to drift.

- ❖ increased genetic variance between the sub-populations

Within a sub-population individuals will become more alike due to inbreeding.

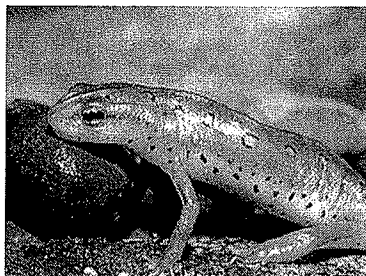
- ❖ reduced genetic variance within the sub-populations

Small populations

Problem

The 'lesser spotted blue newt' inhabit the marshy environment of the Forgotten Flatlands.

The flatlands were drained, resulting in fragmentation into 6 small sub-populations.



Given that migration is not possible, what are the likely outcomes ?

Small populations

Answer: fate of the newts

Sub-populations become distinctive from each other: some are more blue, some have more spots

Variance within a sub-population is lower than in the original population: newts within a location look similar

Whilst total variation is now greater than before, F within each sub-population is increasing. This may lead to extinction of a sub-population, and possibly a net reduction in variance.

Small populations

Maintaining genetic variation within populations

Attention to merit

- ❖ Merit may be adaptability, disease resistance, fecundity
- ❖ Can select parents of the next generation to result in a change in merit (directional selection) or not

Attention to N_e / diversity

- ❖ Both F and drift are effected by N_e
- ❖ Equalising the contribution of each family to the next generation increases N_e
- ❖ Can apply concept of optimal contribution theory
- ❖ Migration is also important

Small populations

Balancing genetic merit & diversity

Select few individuals as parents of the next generation:

- ❖ Strong directional selection (high genetic gain) *but*
- ❖ Low N_e → high inbreeding and drift

Select many individuals as parents of the next generation:

- ❖ Weak directional selection (low genetic gain) *but*
- ❖ High N_e → low inbreeding and drift

Small populations

Maintaining genetic variation over populations

Need to determine both which sub-populations to select, and which individuals within these sub-populations

In some cases crossing between sub-populations may be advantageous

- ❖ e.g. when one population has few animals of one sex
- ❖ to re-create genetic variance

But it can also be detrimental (loss of adaptive alleles - 'outbreeding depression')

Small populations

IAEA regional training course on selective breeding & gene technologies

GENUP for WINDOWS: default.pgp [Default data set: Wool Sheep]

File Edit Modules View Language Window Help

Diversity (2)

Open saved file Save Number of populations available: 6 Number of marker loci: 5 New simulation Update results
Total number of individuals conserved: 100 Alleles per locus: 2

Pop. No.	Merit	Number Available	Number Saved	Change Number Saved	Fix No. Saved	Allele frequencies Locus					Variance within pop.
						1	2	3	4	5	
1	92	77	0			.65	.1	.82	.86	.1	.0991
2	103	95	34			.44	0	0	.07	.42	.11102
3	106	65	65			0	.86	0	.06	.31	.10334
4	98	112	0			.44	0	0	.93	.68	.10508
5	100	145	1			.74	0	0	0	.92	.0532
6	98	137	0			.3	0	0	.87	0	.04082

Results for conserved population:

- Between population variance: 0495
- Within population variance: .2103
- Total allele variance: .2605
- Weighting on between variance: 1
- Weighted genetic variance: .2605
- Mean merit: 104.92

Scatter plot: Genetic variance vs. Merit. FIT = 98.232 percent. Show distance figures More Fit.

Selection for merit

Small populations

GENUP for WINDOWS: default.pgp [Default data set: Wool Sheep]

File Edit Modules View Language Window Help

Diversity (2)

Open saved file Save Number of populations available: 6 Number of marker loci: 5 New simulation Update results
Total number of individuals conserved: 100 Alleles per locus: 2

Pop. No.	Merit	Number Available	Number Saved	Change Number Saved	Fix No. Saved	Allele frequencies Locus					Variance within pop.
						1	2	3	4	5	
1	92	77	40			.65	.1	.82	.86	.1	.0991
2	103	95	29			.44	0	0	.07	.42	.11102
3	106	65	21			0	.86	0	.06	.31	.10334
4	98	112	0			.44	0	0	.93	.68	.10508
5	100	145	1			.74	0	0	0	.92	.0532
6	98	137	15			.3	0	0	.87	0	.04082

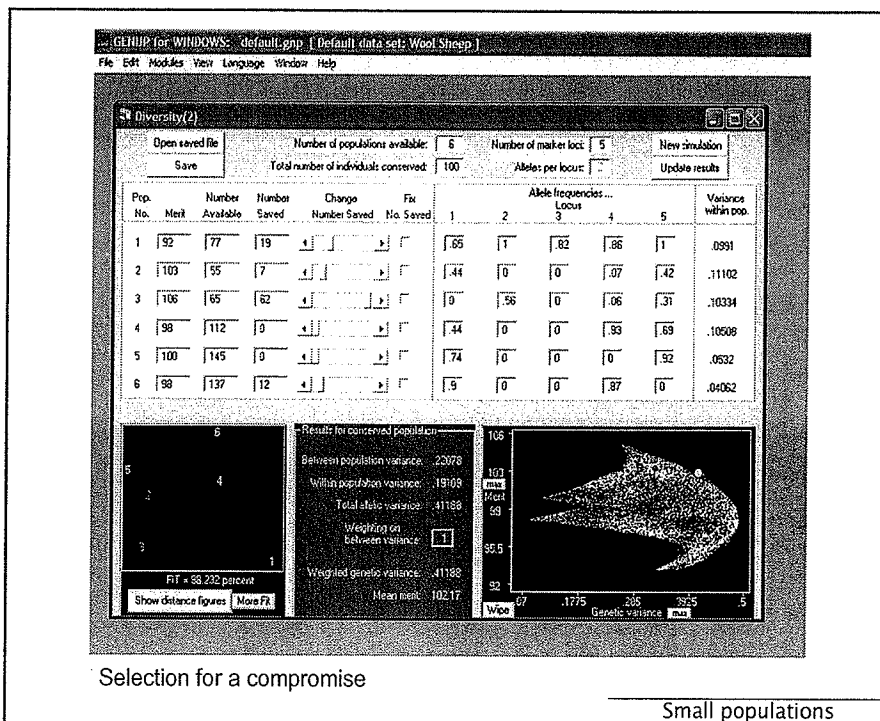
Results for conserved population:

- Between population variance: .2902
- Within population variance: .187
- Total allele variance: .4802
- Weighting on between variance: 1
- Weighted genetic variance: .4802
- Mean merit: 98.45

Scatter plot: Genetic variance vs. Merit. FIT = 98.232 percent. Show distance figures More Fit.

Selection for genetic variance

Small populations



A simulated example

- ❖ 12 breeds with 1000 animals available in each
- ❖ Resources to conserve 120 animals only
- ❖ Genetic variation both within and between breed was determined by molecular markers (5 microsatellite loci with a maximum of 5 alleles each)
- ❖ Diversity is taken as: $V_{\text{within-breed}} + 5 \times V_{\text{between-breeds}}$
- ❖ Merit of each breed has been quantified
- ❖ Need to determine which breed to select and which animals within the breed: maximise merit + λ diversity

Chaiwong and Kinghorn, AAABG 13: 365

Small populations

Results – # of individuals selected from each population

weight on diversity $y(\lambda)$	Population number											
	1	2	3	4	5	6	7	8	9	10	11	12
<i>Merit</i>	108	99	101	107	109	114	111	104	112	95	102	104
0	-	-	-	-	-	120	-	-	-	-	-	-
15	-	-	-	4	14	63	2	-	37	-	-	-
23	-	-	-	18	15	45	28	-	14	-	-	-
34	1	-	-	25	15	39	30	-	8	-	2	-
48	6	-	-	31	8	31	-	-	-	-	11	-
69	6	2	-	32	4	26	-	-	-	-	18	-
110	12	10	-	33	-	22	-	-	-	-	22	-
225	11	16	-	33	-	21	-	-	-	-	24	-

λ is Weight on diversity

Small populations

Summary

Breeding strategies for conservation aim to balance genetic merit and diversity but should also accommodate other constraints e.g. cost of transportation

Within a sub-population this can be managed by the application of optimal contribution theory

Over populations you need to consider which sub-population and which animals within a sub-population

Small populations

