Genetic Interactions and Linkage

Lecture 3

Applied Animal and Plant Breeding

GENE 251/351

School of Environment and Rural Science (Genetics)

Topics

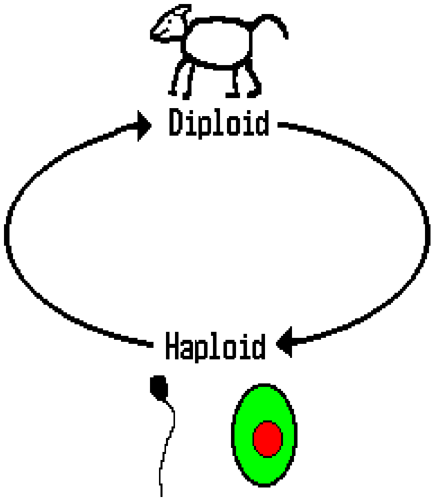
- Relating Genotype to Phenotype

- Mendelian genetics and the single locus model
- Interactions between alleles at one locus
- Interactions between alleles at different loci
- Sex specific traits

- Linkage
- Comparative mapping of genes

- Polyploidy

Remember

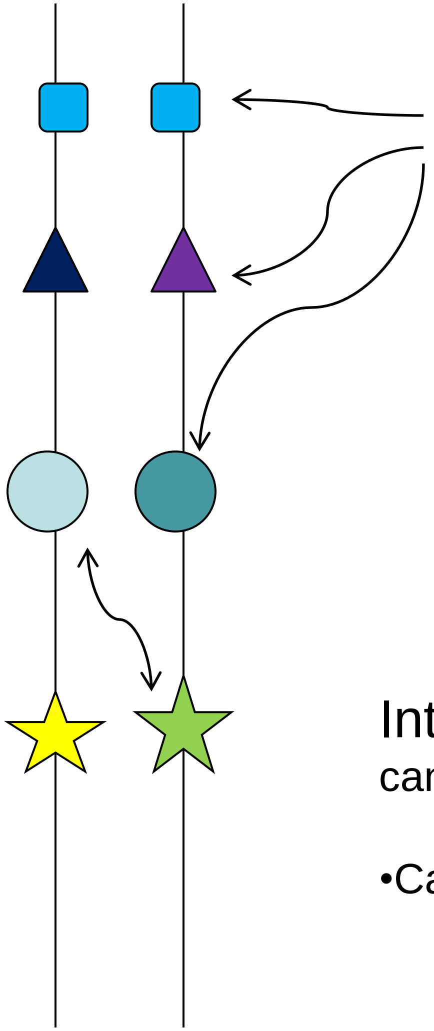


Recombination occurs in the gametes to give rise to new combinations of alleles

Chromosome number is halved in the gametes

Parents pass on only 1 allele of a pair
Parents transmit 50% of their genes
Progeny receive 2 alleles (chromosome sets)
one from each parent

Gene interactions



Interactions between alleles at one locus

- Dominant/recessive
- Co-dominant
- Incomplete dominance

Interaction between alleles at different genes (loci) can result in Epistatis

- Can have varying effects on the phenotype

Examples of inheritance model

- dominance /recessive model
- polledness in cattle, 2 alleles: P and p

  PP = polled

  Pp= polled

  pp = horned

- co-dominant
  - (ABO blood groups)
  - phenotypes are: A, AB and B (and O)

- sex linked

– females	CC	Cc	cc
– males	C-	c-	

example: sex linked genetic defects

Additional Examples of Co-dominant traits

Colour in Carnations

White carnations crossed with red carnations results in pink carnations

Feather colour in birds

White feathered chickens crossed with black feathered chickens gives chickens with black and white feathers

Dominant and Recessive Alleles

- For example: Two heterozygous dogs with black coats are crossed. What would their pups look like? (Alternative is brown coat)

<u>Parents</u>

Phenotype	Black	x	Black
Genotype	Bb		Bb
Gametes	½ B, ½ b		½ B, ½ b

Offspring		Male Gametes	
		B	b
Female gametes	B	BB black	Bb black
	b	Bb black	bb brown

Punnett Square




- Genotype summary
    - ¼ BB :½ Bb :¼ bb
- Phenotype summary
    - ¾ Black dogs : ¼ Brown dogs

or

3 Black dogs : 1 Brown dog

Sex determination

- In mammals
- Females are the homogametic sex, i.e. XX
- Males are the heterogametic sex, i.e. XY

- In birds,
- Females are the heterogametic sex, ZW
- Males are the homogametic sex, ZZ.

How would we discover the model of inheritance?

- Red Bull x Black Cow → ?
- Red Bull x Red Cow → ?
- Black Bull x Black Cow → ?

What to expect when

1. black is dominant
2. red is dominant

Autosomal vs Sex Linked

- Genes located on the sex chromosomes are said to be sex-linked, usually X-linked and they display a different inheritance pattern to autosomal genes.

- In humans most of these genes are on the X chromosome.

- Only 27 genes so far have been identified on the Human Y chromosome.

X-linked inheritance,
e.g. deleterious recessive mutations

Cross between heterozygous female for X-linked trait and a particular male

e.g. Recessive genetic defect: HH = healthy, Hh = healthy, hh = sick, H- = healthy, h- = sick

Male Gametes

Normal male

Carrier Female

Female Gametes

	½ X^H	½ Y
½ X^H	¼ X^HX^H	X^H Y
½ X^h	X^HX^h	X^h Y
	Female offspring	Male offspring

All normal

50% sick

Sex-limited genes

- The sex of an organism can influence the expression of autosomal genes via hormonal feedback systems. These are called *sex-limited* genes.
- Examples:
  - milk production genes, genes for reproductive traits

- Individuals of both sexes carry copies of all of these genes even if they are not expressed in a particular sex!

Sex-influenced genes

- The dominance relationship of alleles for these autosomal genes change depending on the sex of the individual.

- Example:
  - horned Dorset sheep crossed with hornless Suffolk

Dorset h^+h^+ x Suffolk hh

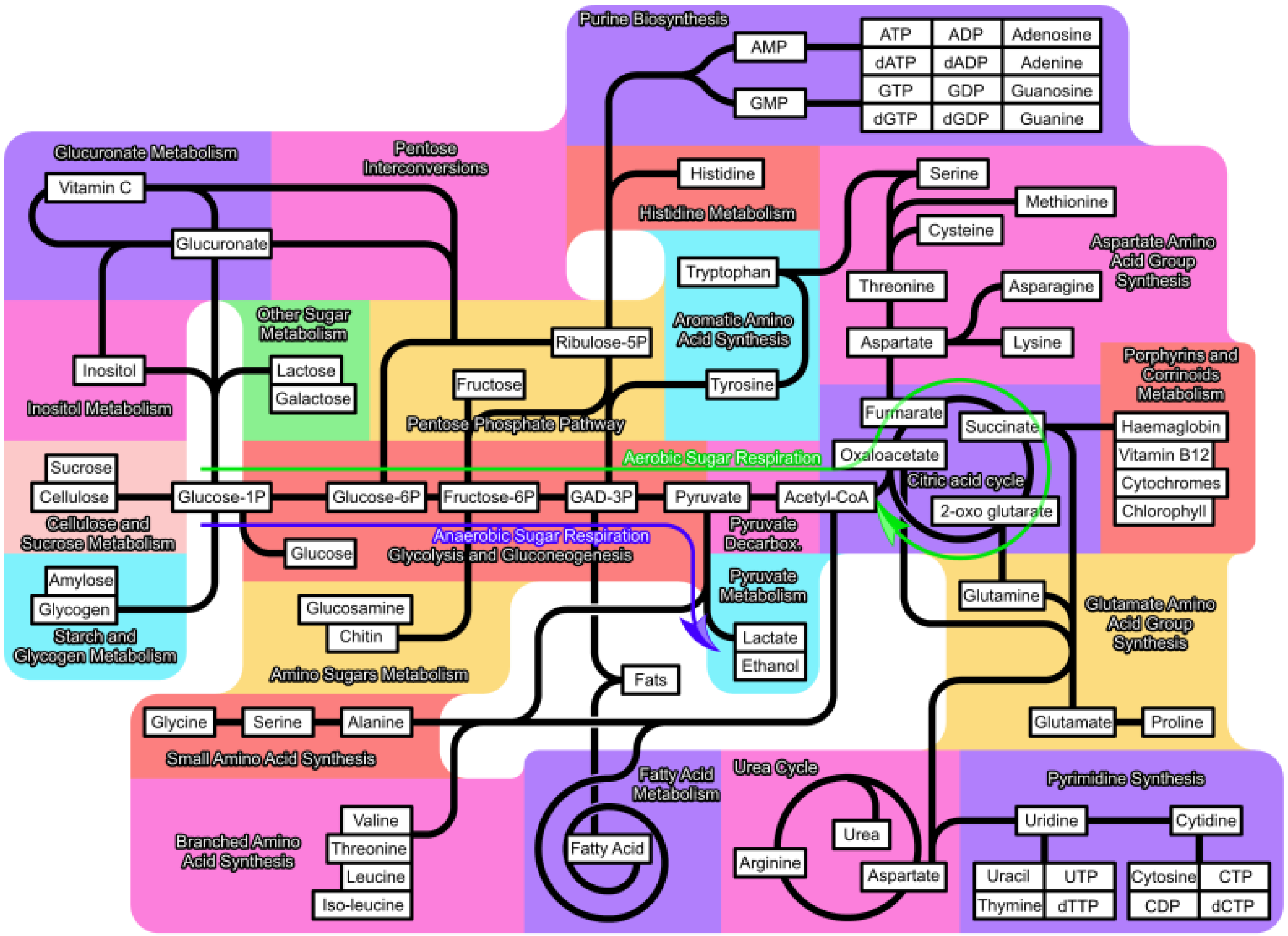
Females h^+h = hornless

Males h^+h = horned

Epistasis - interactions between different loci

- Alleles for one gene can influence the expression of alleles for another gene
- Complex biochemical pathways determine production of various chemicals / phenotypes eg hair or coat colour
- For every step at least one gene product is needed
- Epistasis results when genes are involved in the same biochemical pathway

Numerous biochemical pathways for epistasis to be present



Summary of Epistatic ratios

When epistasis is operative between two gene loci, the number of phenotypes appearing in the offspring from dihybrid parents will be less than four. Reference only

Genotypes	A-/B-	A-/bb	aa/B-	aa/bb
Classical Ratio	9	3	3	1
Dominant epistasis	12		3	1
Recessive epistasis	9	3	4	
Duplicate genes with cumulative effect	9	6		1
Duplicate dominant genes	15			1
Duplicate Recessive genes	9	7		
Dominant and Recessive interaction	13		3	

Epistasis example

In European Simmentals coat colour is either deep red or yellow, both with white face and markings.

Red = $E^r E^r$ DD; Yellow = E^rE^r Dd or E^rE^r dd

Here D – the dilution allele shows epistasis and dilutes the expression of the red allele even though the individuals are all homozygous for that allele.

The alternative to the red phenotype is black – E^D-

D is a diluter gene where

DD=dark, Dd=medium colour, and dd=pale colour

Red and black calves

Diluted red = yellow

diluted black = 'mouse'

Linkage, Recombination and Mapping

Parent animal

A ***B***

a ***b***

Non-recombinant gametes
Frequency: (1 – r)

AB

ab

Recombinant gametes
Frequency: r

Ab

aB

Classical Mapping: Using a test cross

	Parent 1	AABB	x aabb	Parent 2	
F1	AaBb (100%)				
F1-gametes		AB	Ab	aB	ab
	frequencies				
A and B are unlinked:		25	25	25	25
A and B linked:		35	15	15	35
A and B tightly linked		48	2	2	48

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Classical Mapping: Using a test cross

Parent 1 AABB x aabb Parent 2

F1 AaBb (100%)

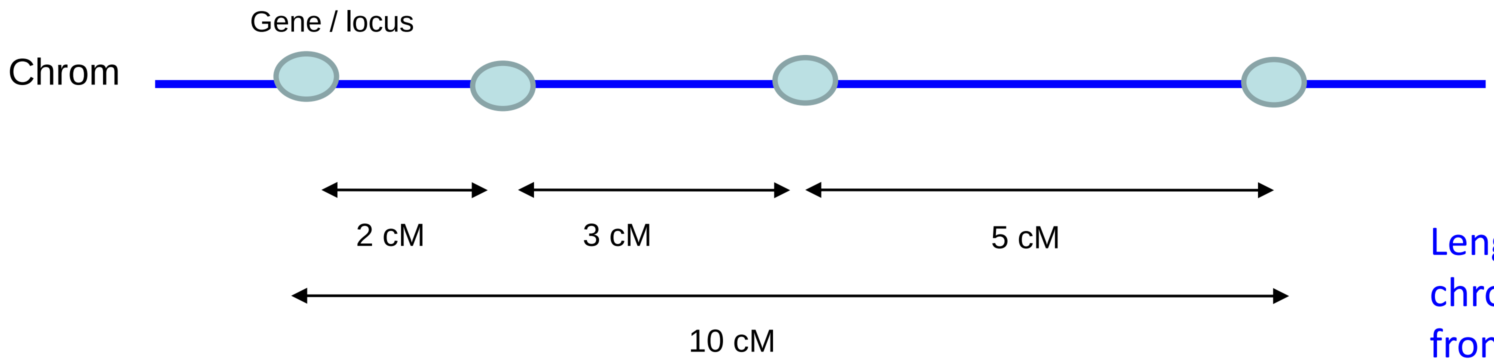
F1-gametes frequencies	AB	Ab	aB	ab	Recombination Frequency
A and B are unlinked:	25	25	25	25	50
A and B linked:	35	15	15	35	30
A and B tightly linked	48	2	2	48	4

Recombinants (Ab, aB)

Genetic linkage maps

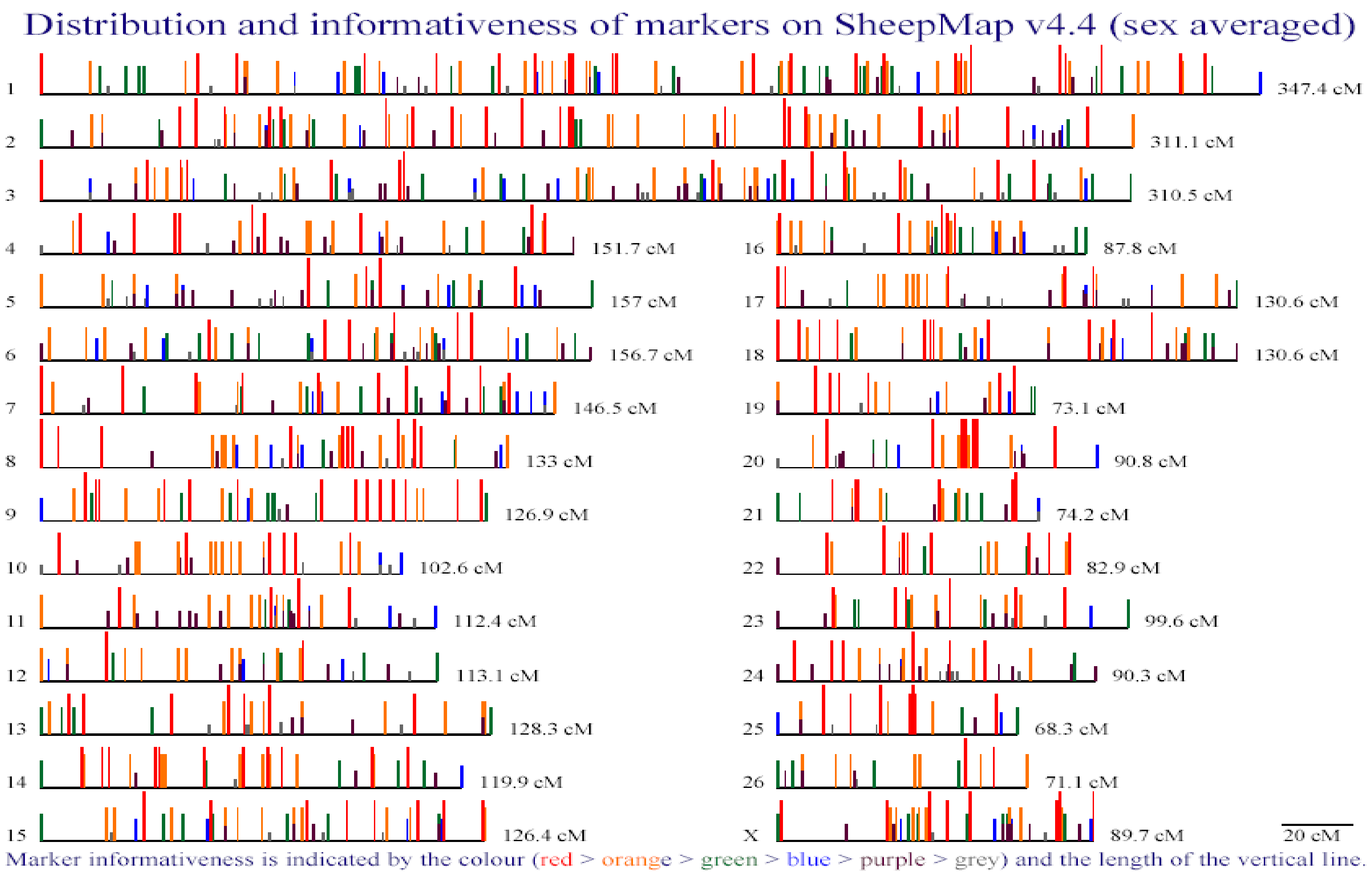
- Genetic distances determined according to recombination fraction between each pair of loci

- The unit of measure is Morgans (or centimorgans, cM)



Length of chromosomes vary from 50 - 350 cM

Sheep map-microsatellite markers



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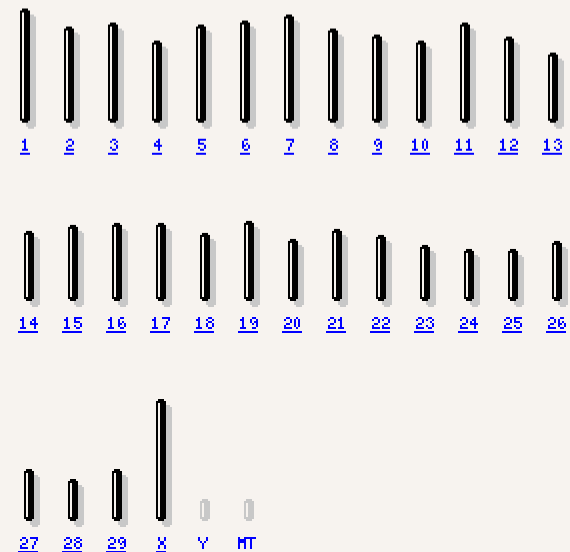
lturally important animal for beef and milk production.

Projec

Eukaryota; *Metazoa*; *Chordata*; *Craniata*; *Vertebrata*; *Euteleostomi*; *Mammalia*; *Eutheria*; *Laurasiatheria*; *Cetartiodactyla*; *Ruminantia*; *Pe*
Bovinae; *Bos*; *Bos taurus*

Photo: courtesy of Terri Hobbs (www.crazyforcows.com)



- unknown chromosome size

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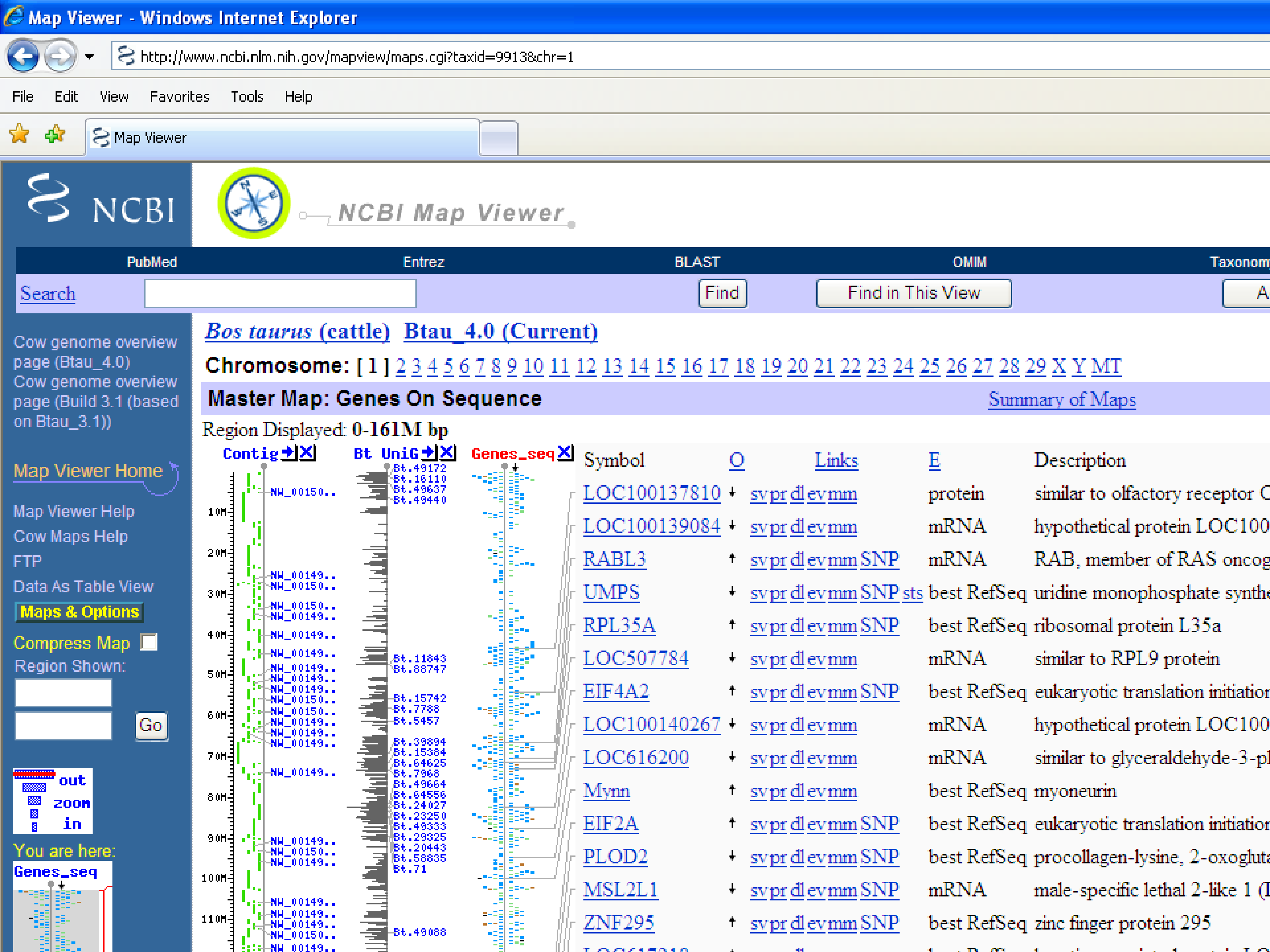
naps:

Sequence Maps	Genetic maps	RH maps
15 maps	MARC	ILTX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Cow genome overview page (Btau_4.0)
Cow genome overview page (Build 3.1 (based on Btau_3.1))
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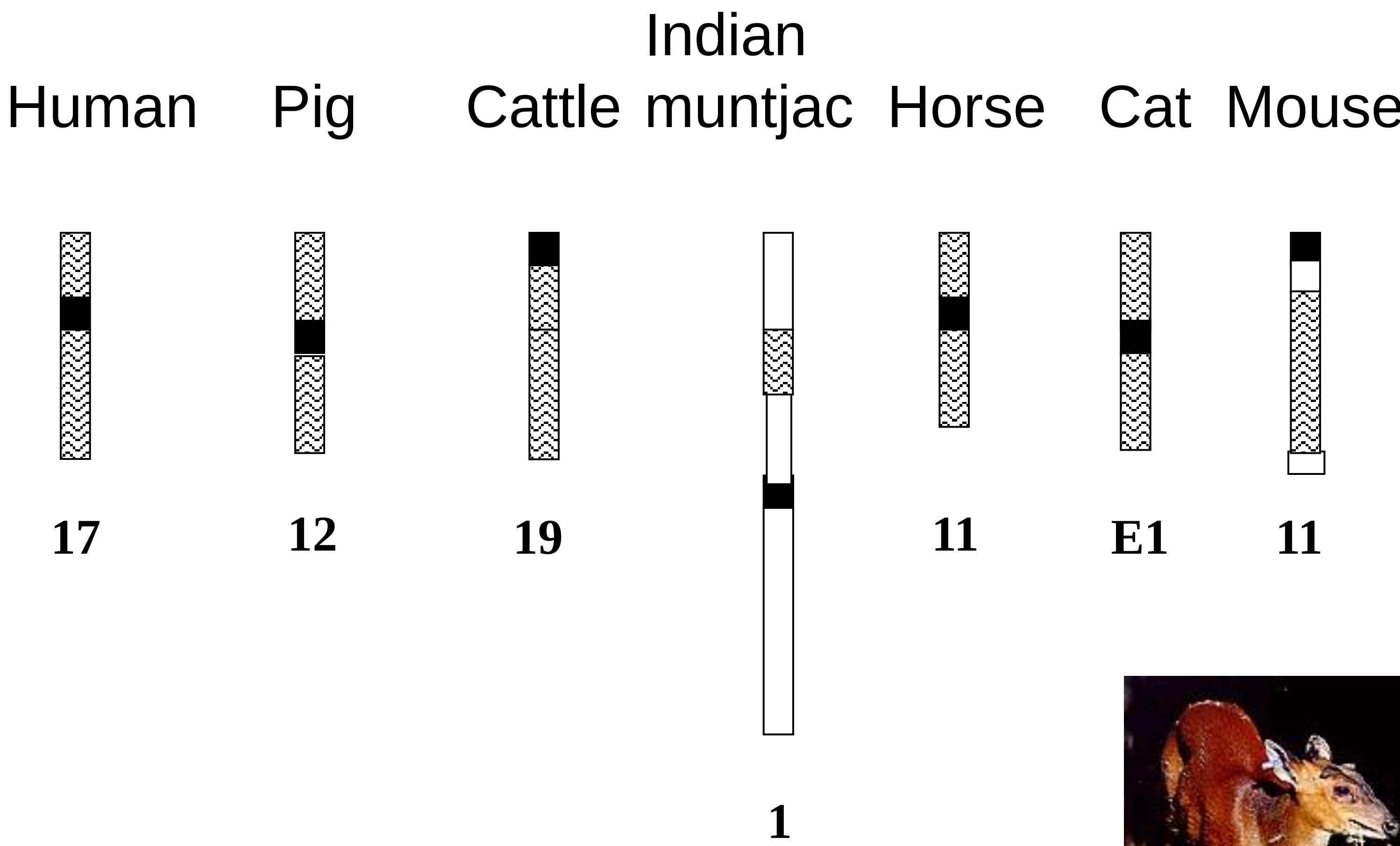
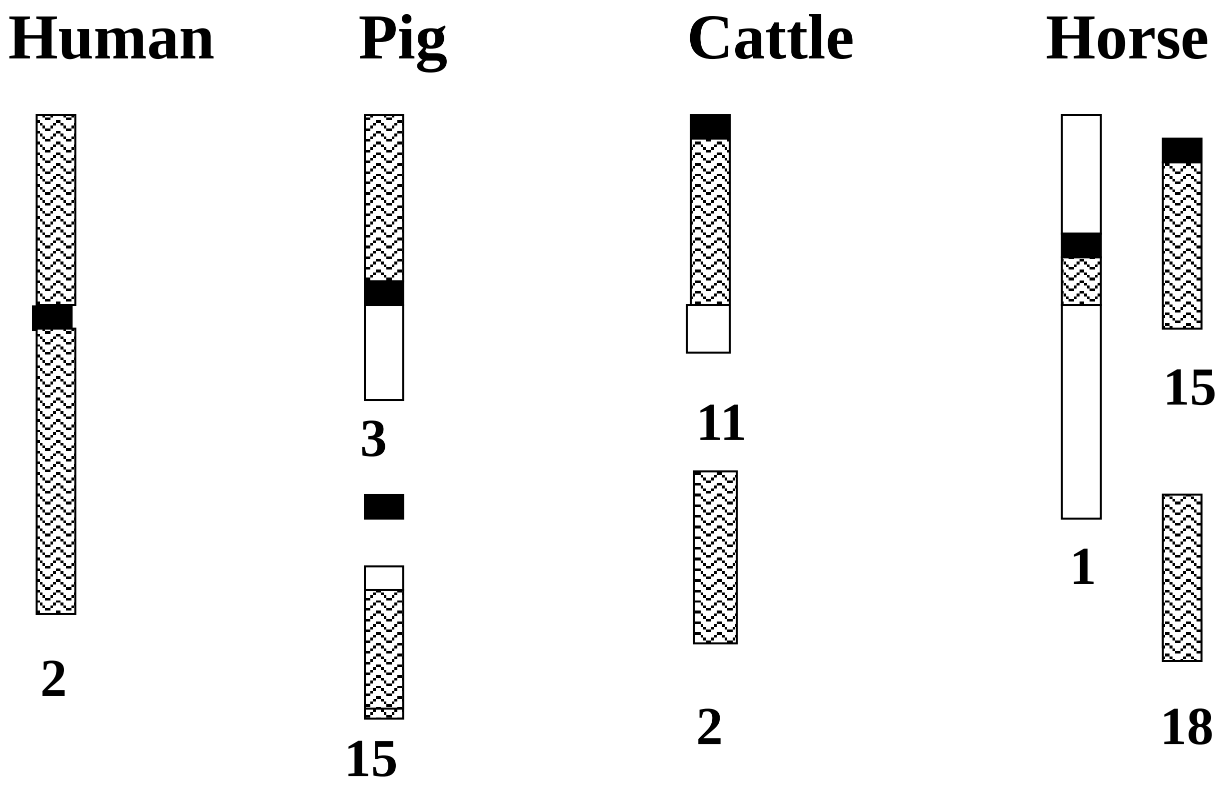
Maps & Options
Compress Map
Region Shown:
Go
out
zoom
in
You are here:
Genes_seq
Bos taurus (cattle) Btau_4.0 (Current)
Chromosome: [1] 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 X Y MT
Master Map: Genes On Sequence
Summary of Maps
Region Displayed: 0-161M bp
Contig
Bt UniG
Genes_seq
Symbol O Links E Description
LOC100137810 sv pr dl ev mm protein similar to olfactory receptor
LOC100139084 sv pr dl ev mm mRNA hypothetical protein LOC100
RABL3 sv pr dl ev mm SNP mRNA RAB, member of RAS oncog
UMPS sv pr dl ev mm SNP sts best RefSeq uridine monophosphate synthe
RPL35A sv pr dl ev mm SNP best RefSeq ribosomal protein L35a
LOC507784 sv pr dl ev mm mRNA similar to RPL9 protein
EIF4A2 sv pr dl ev mm SNP best RefSeq eukaryotic translation initiation
LOC100140267 sv pr dl ev mm mRNA hypothetical protein LOC100
LOC616200 sv pr dl ev mm mRNA similar to glyceraldehyde-3-p
Mynn sv pr dl ev mm best RefSeq myoneurin
EIF2A sv pr dl ev mm SNP best RefSeq eukaryotic translation initiation
PLOD2 sv pr dl ev mm SNP best RefSeq procollagen-lysine, 2-oxoglut
MSL2L1 sv pr dl ev mm SNP mRNA male-specific lethal 2-like 1
ZNF295 sv pr dl ev mm SNP best RefSeq zinc finger protein 295

Synteny of Human chromosome 17



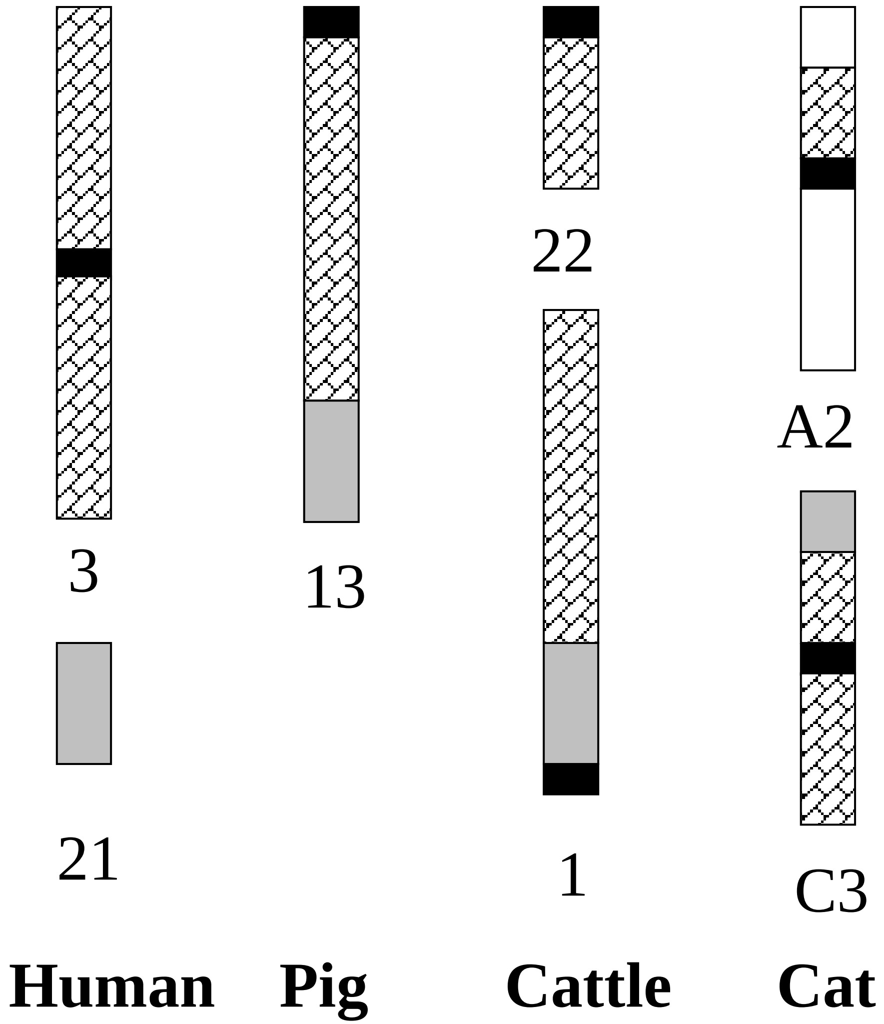
Synteny of entire chromosomes conserved

Synteny of Human chromosome 2.



Synteny of large chromosome segments conserved.

Synteny of Human chromosome 3 and 21



Segments of chromosomes combine to produce new synteny.

Polyploidy

- Majority of animals are diploid = 2n

- A lot of plants (and a small number of animals) can tolerate having multiple copies of the chromosome set therefore are = 3n, 4n, 6n

Polyploidy

- This can either occur due to pollination by a closely related species and subsequent doubling of the chromosomes because the mitotic spindles don't form properly.

- The cell's own mitotic spindle formation fails and you get doubling of the chromosome set.

- Most species survive because they can reproduce vegetatively.

Polyploidy

- If chromosome sets are doubled it means pairing can happen properly at meiosis and so these can reproduce sexually. These are tetraploids (at least).

Examples:

  – roses, chrysanthemums, tulips, coffee, bananas, cotton, wheat.

Octoploid

Tetraploid

Triploid

Polyploidy

- Individuals with multiple copies of the same genome set are AUTOPOLYPLOIDS

- and those with genome sets from different species are ALLOPOLYPLOIDS.

Wheat – a hexaploid

- **Wheat, *Triticum aestivum*, is hexaploid (6n) and is called an allopolyploid**

AA x BB
(n=7) (n=7)
T. urartu x A. searsii

AB
(n=14)
Emmer wheat

AABB x DD
(n=14) (n=7)
T.Dicoccoides x Ae tauschii

1 Two diploid species cross to produce a hybrid with two different sets of chromosomes in its genome.

2 The chromosomes in the hybrid double to form a tetraploid.

3 The tetraploid hybrid crosses with another diploid species to produce a plant with three sets of chromosomes in its genome.

Wheat – a hexaploid

AABB x DD
(n=14) (n=7)

↓

ABD
(n=21)

↓

AABBDD
(n=21)
Bread wheat, spelt

4 The chromosomes in the triple hybrid double to form a hexaploid.

5 Modern bread wheat is a hexaploid – a hybrid of three different species.