

# Genetic Conditions & GeneProb

## NZ Angus



agricultural business  
research institute



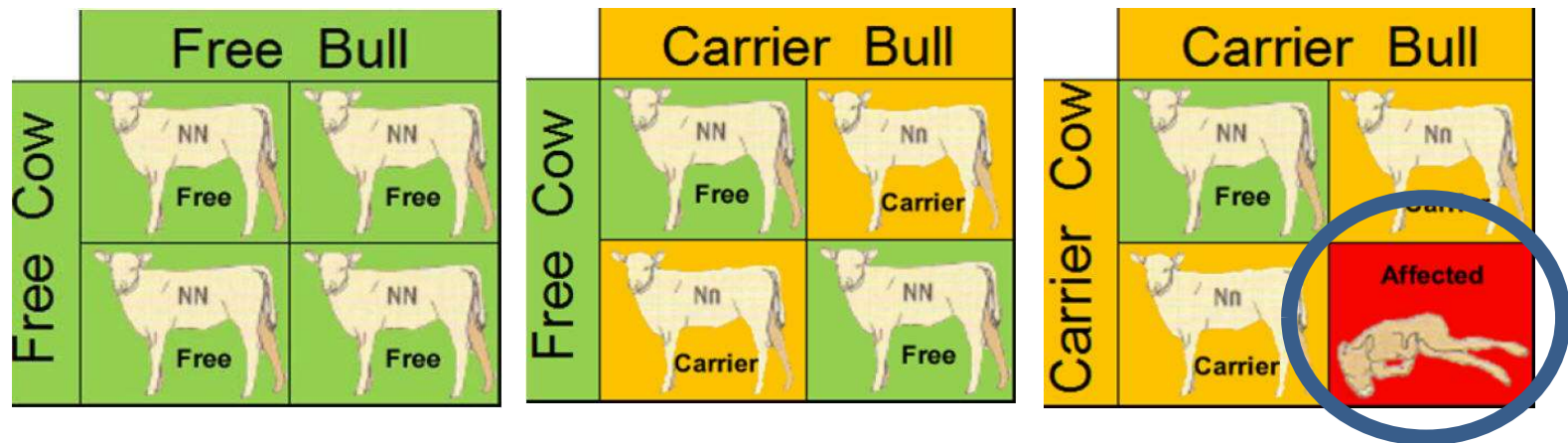
# What are Genetic Conditions?

- Exist in all species including humans
- Caused by an abnormality in an individual's DNA
- Abnormality can range from a single gene mutation to addition or loss of an entire chromosome
- While incidence of a genetic condition is normally rare in a population, the existence of genetic conditions is common:
  - over 400 genetic conditions identified in cattle
  - few have genetic tests commercially available



# Recessive Inheritance

- **Most Genetic Conditions:**
  - are controlled by a single gene
  - and have simple recessive inheritance
- **Three possible genotypes:**
  1. **Free** (zero copies of the disease allele; does not have the disease)
  2. **Carrier** (one copy of the disease allele; does not have the disease)
  3. **Affected** (two copies of the disease allele; has the disease)



# NZ Angus: genetic condition test results

Status	AM	CA	DD	NH
Free	8,267	9,265	8,943	9,250
Carrier	951	1,077	1,343	1,178
Affected	0	0	10	0
Total	9,218	10,342	10,296	10,428
% testing of calves born 2011-21	2.8	3.2	4.3	3.2



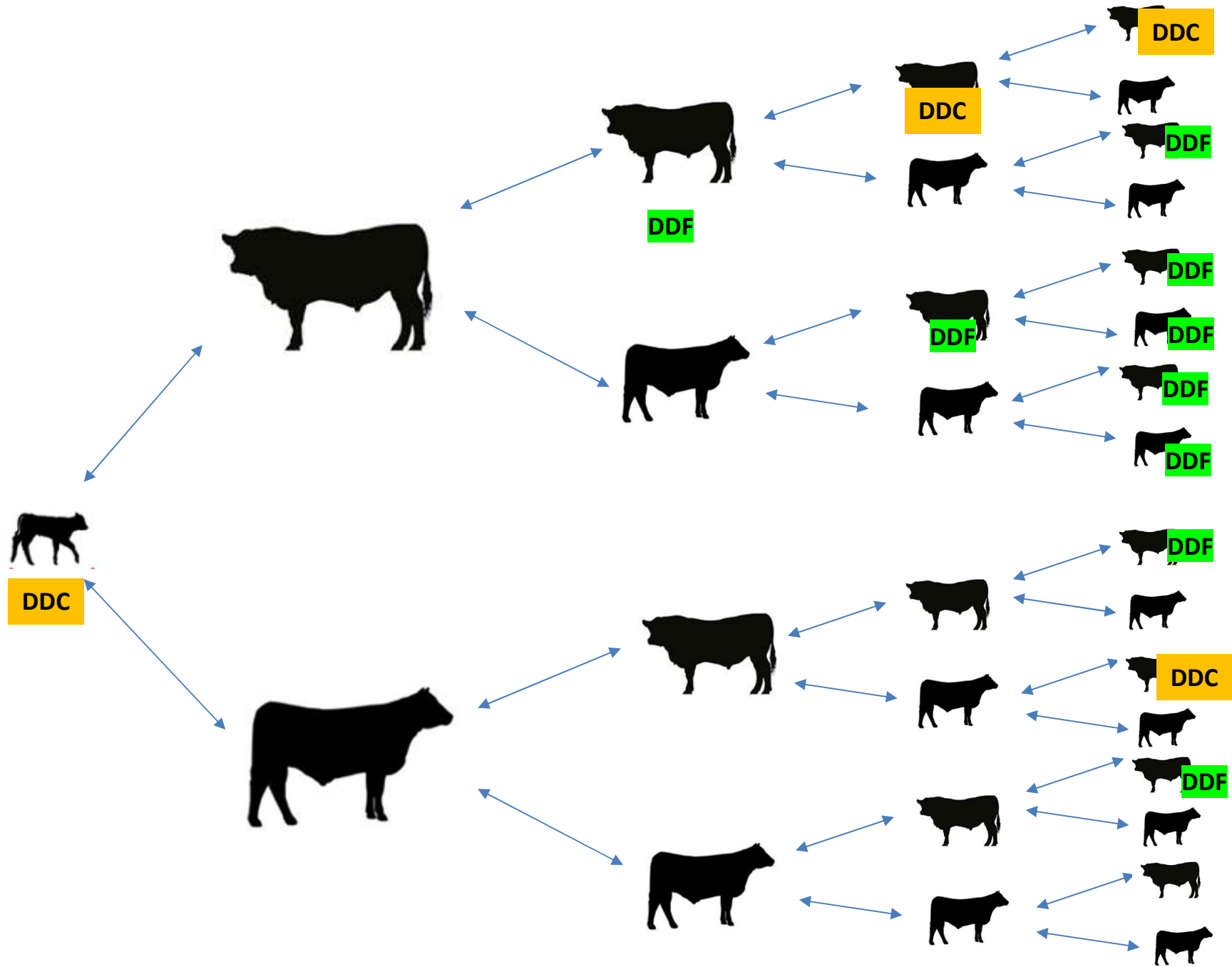
# GeneProb

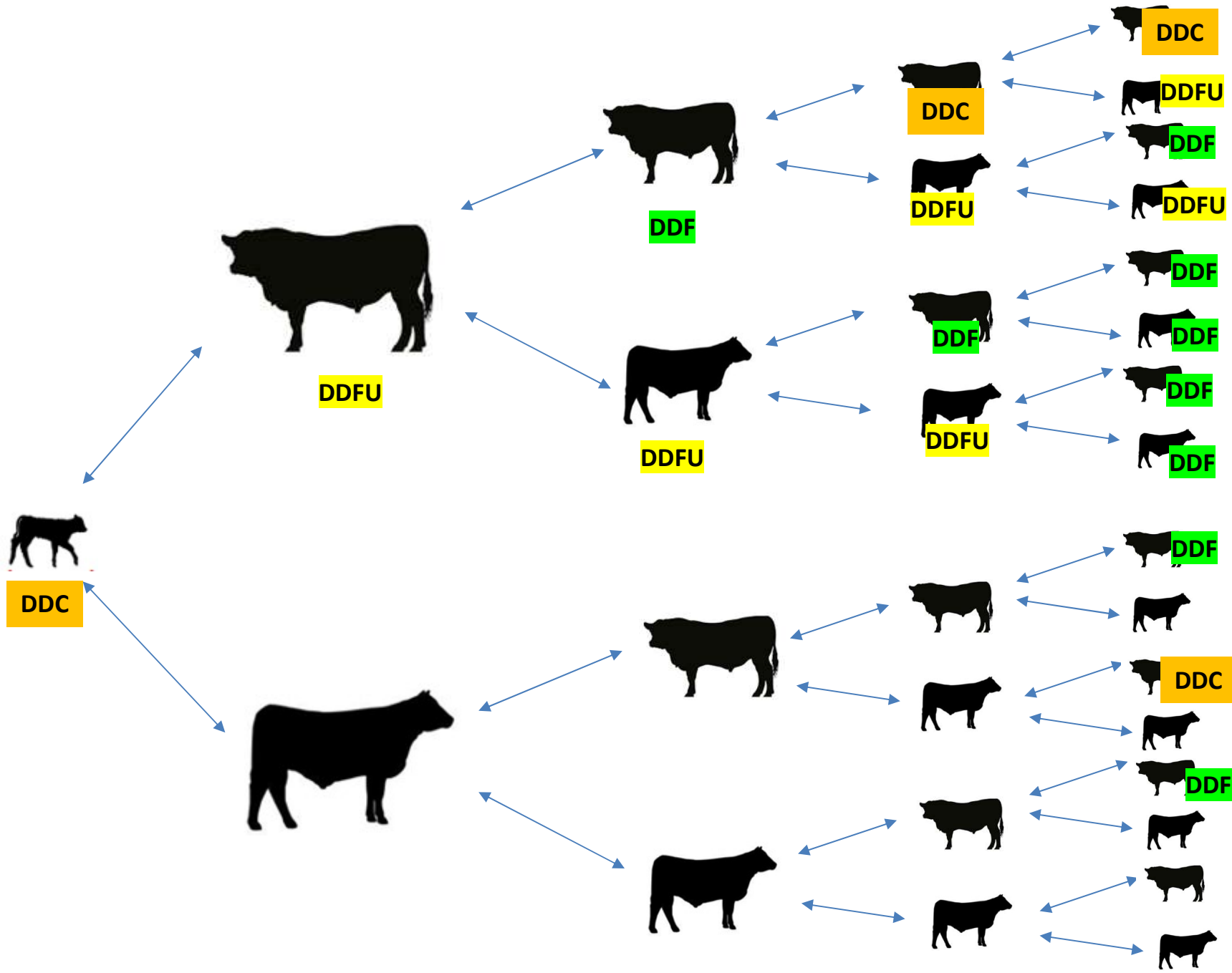
- Written by Prof. Brian Kinghorn (UNE)
  - Underpinned by scientific review and publication
  - Not species specific
- Algorithm uses **pedigree** information and genetic condition **test results** to calculate the probability of untested individuals being carriers
- Licenced to ABRI as
  - BREEDPLAN companion product
  - Integrated with ILR2

# GeneProb Codes

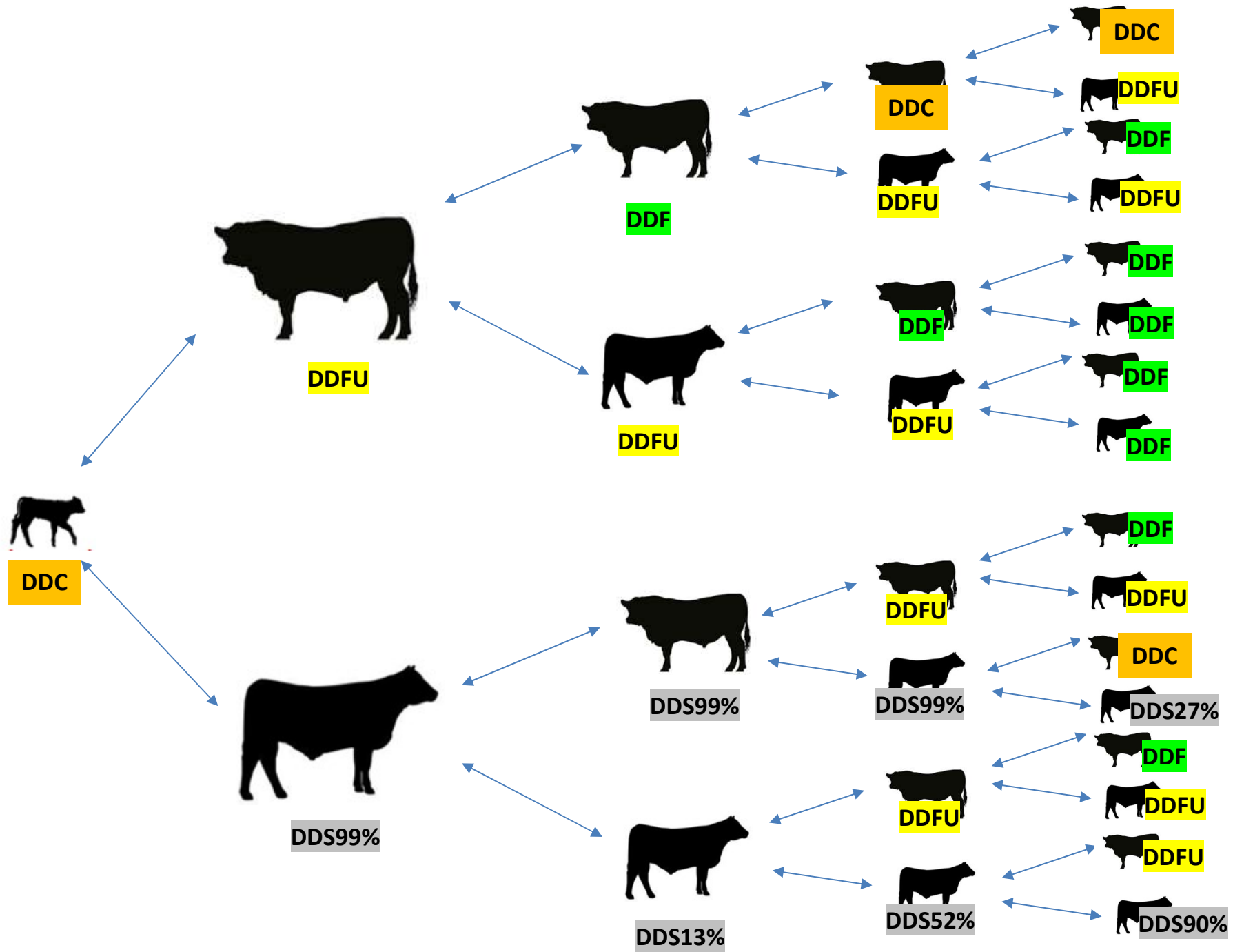
Code	Description
_F	<b>Tested free</b>
_FU	Not tested, based on pedigree expected to be free
_C	<b>Tested carrier</b>
_XX%	Not tested, based on pedigree the animal has the indicated percentage chance of being a carrier
_A	<b>Tested affected</b>

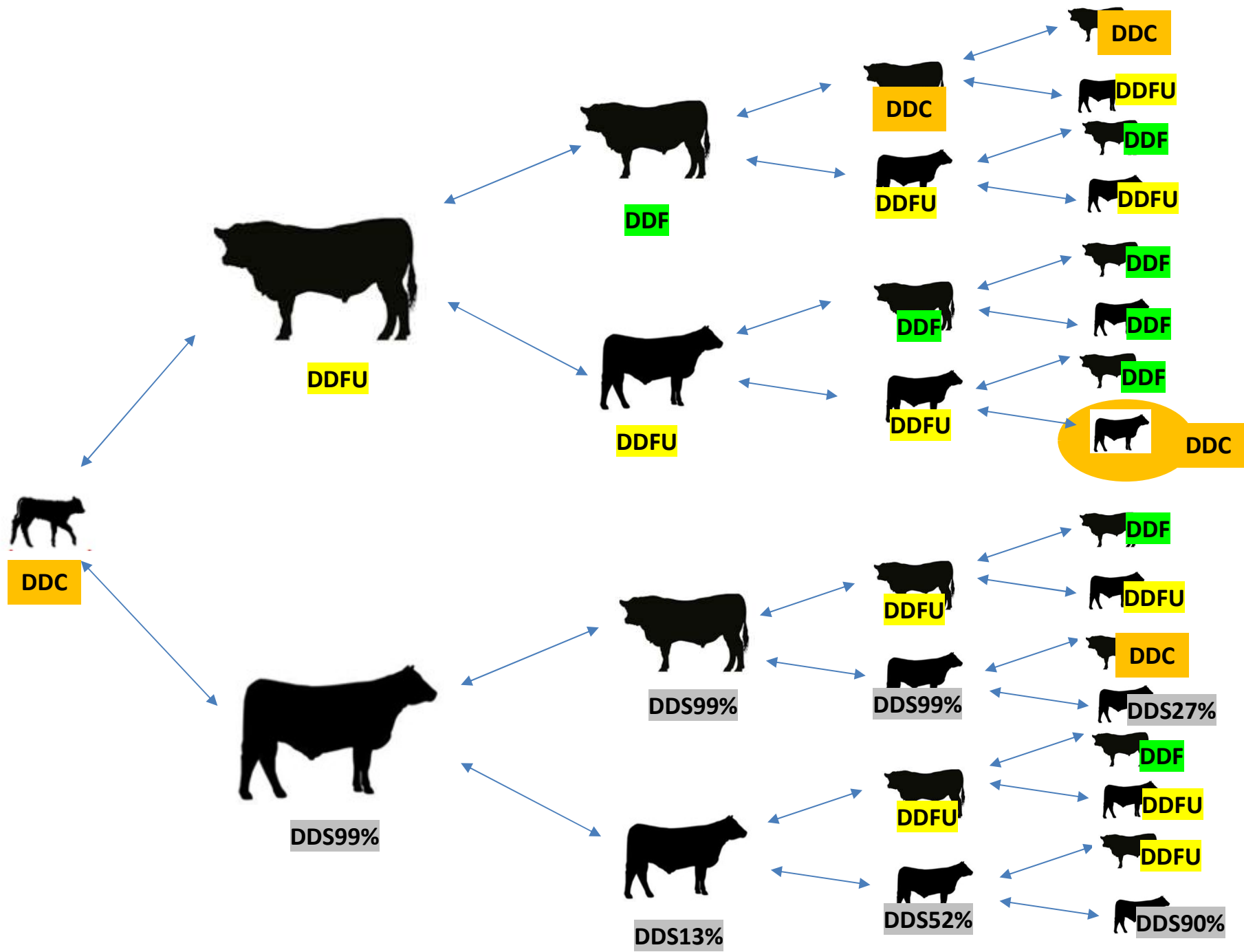


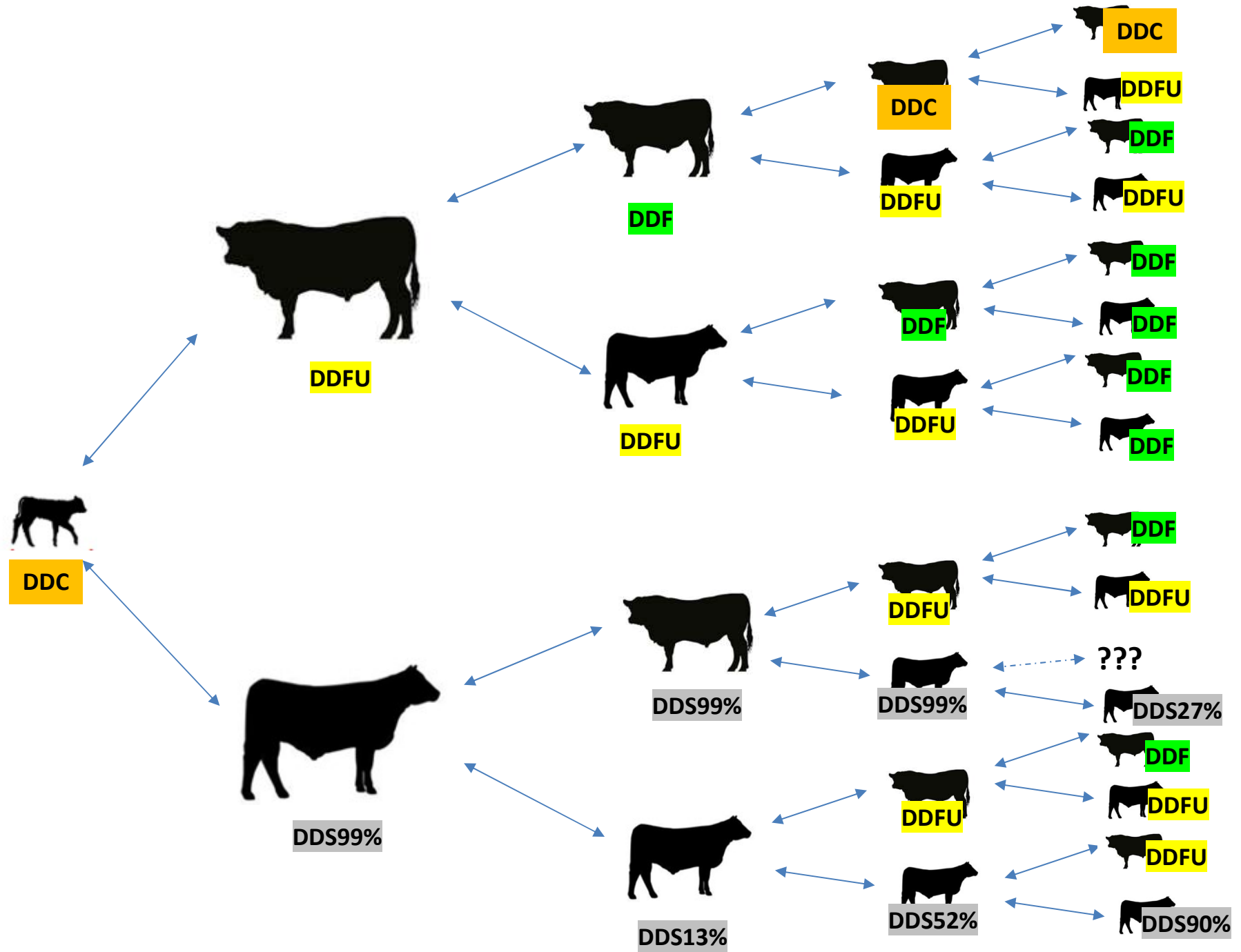










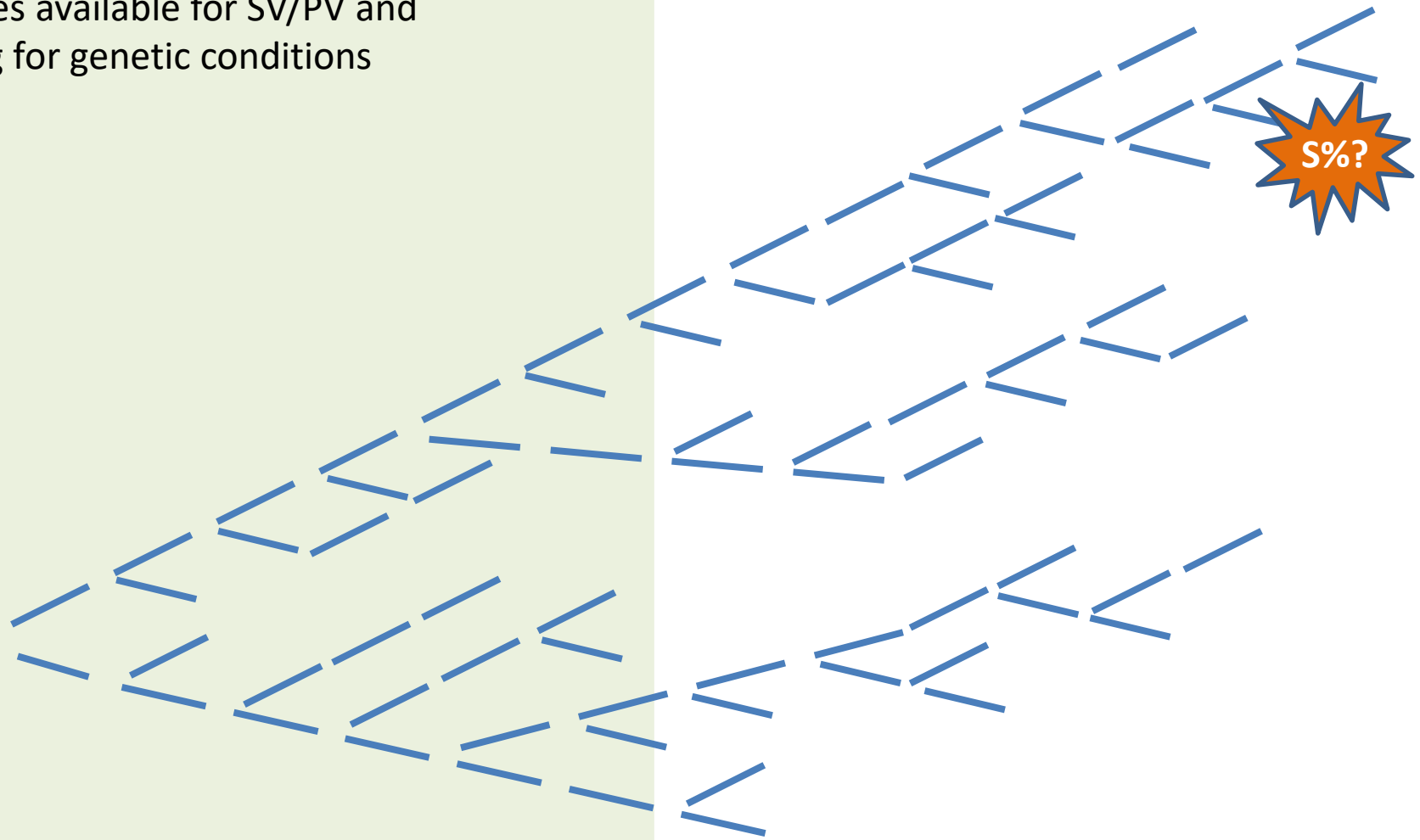


# GeneProb probability results depend on:

- Correctness of pedigree records:
  - Errors in pedigree contribute to errors in probability calculations
  - but GeneProb can identify potential pedigree errors
- Correctness of genetic test condition results:
  - Correct result for correct animal?
  - but GeneProb can identify potential recording errors
- Depth (knowledge) of pedigree
  - Less verification of pedigree as we move deeper into pedigree
  - Low/no test results among historic population
- Frequency of testing for genetic conditions
  - Influences knowledge about patterns of inheritance

**Assume:**

Services available for SV/PV and testing for genetic conditions



## Truncating pedigree: $\geq$ DDS20%

Cohort	Count	Corr	AvS%		Max change	
			Full	Trunc	Down	Up
1990-99	2,189	0.499	0.220	0.089	-0.948	0.824
2000-09	10,472	0.955	0.309	0.295	-0.834	0.326
2010-19	13,130	0.996	0.298	0.298	-0.227	0.232
2020+	197	0.999	0.239	0.238	-0.010	0.005

Full GP analysis: all animals

Truncated GP analysis: calving years 1990+

# Questions

