




www.genocan.eu



www.kchrr.com



www.ridgeback-database.com




www.vri.cz


## Ridge gene test: Its use and potential

Miroslav Hornak, Ph.D.  
Veterinary Research Institute  
Brno, Czech Republic


Rhodesian Ridgeback World Congress  
2016, June 28 - 30<sup>th</sup>, Sweden




www.genocan.eu



www.kchrr.com



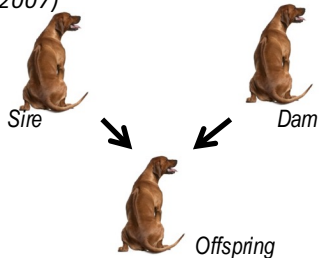
www.ridgeback-database.com



www.vri.cz

## Ridge Genetics

Ridge in Rhodesian Ridgebacks is caused by specific mutation:  
~ 133.000 DNA base pairs duplication on canine chromosome 18 = „Ridge gene“  
(Salmon Hilbertz, et al.;2007)





Sire      Dam


Offspring


Dog might have:

- 2 mutations (duplications; ridge genes) = „R/R“ (*dominant homozygote*)
- 1 mutation (duplication; ridge gene) = „R/r“ (*heterozygote*)
- no mutation (duplication; ridge gene) = „r/r“ (*wild type*)

  
[www.genocan.eu](http://www.genocan.eu)

  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)


  
[www.vri.cz](http://www.vri.cz)


## Ridge Genetics


Ridge formation is caused by presence of single mutation (duplication, ridge gene)  
= **dominant genetic trait**


Dog might be:


- ridged – R/R (=2 mutations, mutation on both chromosomes) –passes ridge gene to all puppies
- R/r (1 mutation on one chromosome, no mutation on another chromosome) – passes mutation only to 50% of puppies – *flip of coin*
- ridgeless - r/r (no mutation on both chromosomes) = passes „no mutation“ to all puppies



  
[www.genocan.eu](http://www.genocan.eu)

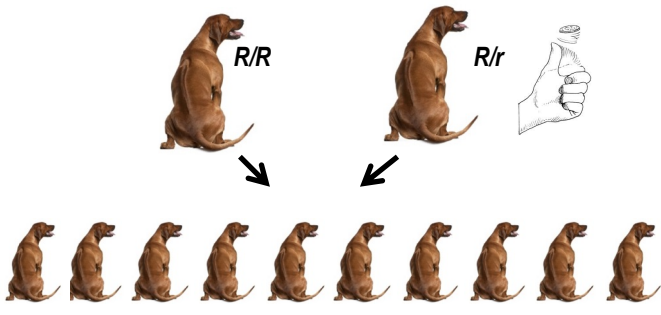
  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)

  
[www.vri.cz](http://www.vri.cz)

## Ridge Genetics

Mating combination RR x Rr





Parents


Puppies


50% R/R + 50% R/r

All puppies expected to be ridged!

  
[www.genocan.eu](http://www.genocan.eu)

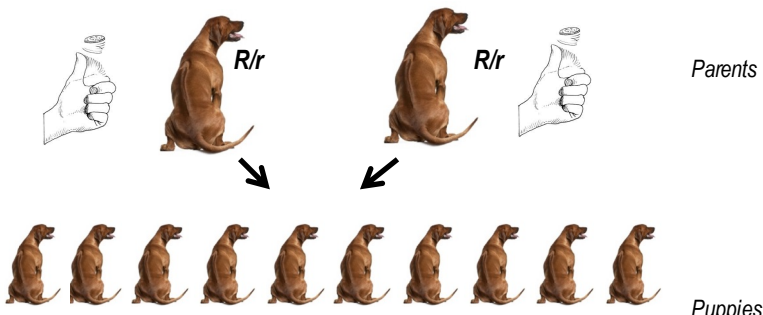
  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)

  
[www.vri.cz](http://www.vri.cz)

### Ridge Genetics

Mating combination  $Rr \times Rr$





*Parents*


*Puppies*


25%  $R/R$  + 50%  $R/r$  + 25%  $r/r$ 

25% puppies expected to be ridgeless!

  
[www.genocan.eu](http://www.genocan.eu)

  
[www.kchrr.com](http://www.kchrr.com)


  
[www.ridgeback-database.com](http://www.ridgeback-database.com)


  
[www.vri.cz](http://www.vri.cz)

### Ridge gene test


Based on physical appearance not possible to distinguish between:  
 $RR$  – 2 ridge genes and  $Rr$  – 1 ridge gene

On several breeders' requests  
- **development of genetic test for ridge disposition** (= copy number of ridge genes)  
Veterinary Research Institute, Brno, Czech Republic  
from 8/2014







www.genocan.eu



www.kchrr.com



www.ridgeback-database.com




www.vri.cz

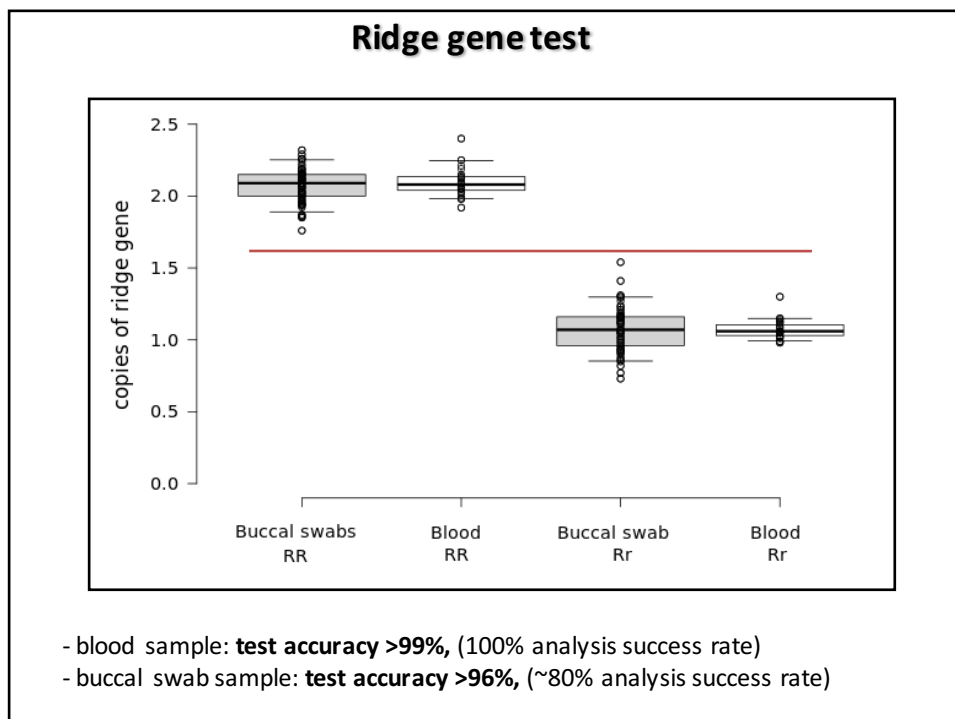
## Ridge gene test

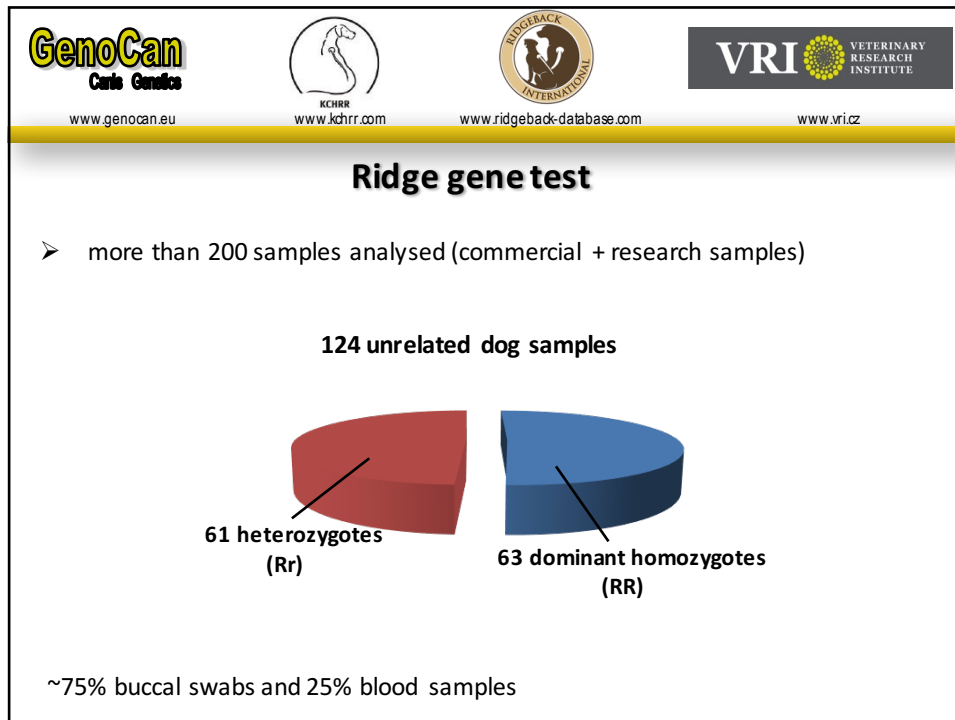
- molecular genetic test for copy number of ridge gene = detection of duplications  
**result** = no ridge gene – rr; 1 ridge gene – Rr; 2 ridge genes – RR
- **Quantitative fluorescent PCR test** based on threshold values

Dog sample:

- blood or buccal swab samples







GenoCan  
Canine Genetics  
www.genocan.eu

KCHRR  
www.kchrr.com

RIDGEBACK  
INTERNATIONAL  
www.ridgeback-database.com


VRI  
VETERINARY  
RESEARCH  
INSTITUTE  
www.vri.cz

### Ridge gene test – surprising finding


- Some sires or dams with 2 ridge genes (RR) produced ridgeless puppies!


Example 1:  
A stud dog (RR) produced 15 litters  
- in 14 litters all puppies ridged  
- in 1 litter two ridgeless puppies  
Stud dog retested – RR confirmed  
both ridgeless puppies tested – both Rr


Example 2:  
A dam (RR) produced 16 puppies in 1 litter  
- 15 puppies ridged, but 1 was ridgeless  
Dam retested – RR confirmed  
ridgeless puppy tested – Rr!




We genetically confirmed 4 (Rr) ridgeless animals

  
[www.genocan.eu](http://www.genocan.eu)

  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)

  
[www.vri.cz](http://www.vri.cz)

### Ridge gene test – surprising finding

- Dataset analysis (in collaboration with Stephanie Muller – founder of Ridgeback International Database)


RR x Rr mating (confirmed by ridge gene test)


- in total 229 puppies born
- 9 puppies ridgeless


- RR x Rr mating produces ridgeless puppies in 3.9% (9/229)
- RR x RR mating never produced ridgeless puppy


**Ridge gene might be silenced (suppressed) occasionally!**

**Ridge genetics is not governed by simple dominant inheritance, but rather incomplete penetrance model!**

  
[www.genocan.eu](http://www.genocan.eu)

  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)

  
[www.vri.cz](http://www.vri.cz)

### Ridge gene test – research samples


- Focus on Dermoid sinus and ridge abnormalities


Dermoid sinus – ridge gene is predisposing genetic factor


Dominant homozygotes (RR) ~5x increased risk for DS  
(Salmon Hilbertz, et al.;2007)


- In our laboratory we confirmed occurrence of DS predominantly in dominant homozygotes (RR)

**We request samples of dogs with DS for research**

  
[www.genocan.eu](http://www.genocan.eu)


  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)


  
[www.vri.cz](http://www.vri.cz)


### Ridge gene test – research samples


- ridge abnormalities
  - short (partial) ridge
  - multiple crowns,
  - offset crowns
  
- We tested several Ridgebacks with multiple crowns – all homozygous (RR)




**We request samples of dogs with ridge abnormalities**

  
[www.genocan.eu](http://www.genocan.eu)

  
[www.kchrr.com](http://www.kchrr.com)

  
[www.ridgeback-database.com](http://www.ridgeback-database.com)

  
[www.vri.cz](http://www.vri.cz)


### Summary I


Genetics in Rhodesian ridgeback breeding			
Parents (Sire x Dam)	Puppies		
	ridged	ridgeless	risk of Dermoid sinus
RR x RR	100%	0%	increased
RR x Rr or Rr x RR	>90%	<10%	normal
Rr x Rr	75%	25%	normal / low
RR x rr or rr x RR	>90%	<10%	low
rr x rr	0%	100%	very low


RR – dominant homozygote (2 ridge genes), RR puppy is always ridged


Rr – heterozygote (1 ridge gene), Rr puppy is in 90% ridged, in approx. 10% ridgeless (ridge gene is suppressed)

rr – ridgeless (no ridge gene)

  
 www.genocan.eu

  
 www.kchrr.com

  
 www.ridgeback-database.com

  
 www.vri.cz

## Summary II

- We developed and validated sensitive ridge gene test for commercial and research use (more info at [www.genocan.eu](http://www.genocan.eu))
- The ridge gene (133.4 kb duplication) predispose to ridge formation with incomplete penetrance
  - ridgeless dogs occasionally carry „silenced“ ridge gene (Rr)
  - approximately 8% of heterozygotes (Rr) might be ridgeless (deduced from RR x Rr mating)
- Dominant homozygotes (RR) are predisposed to DS occurrence and very likely to some ridge abnormalities (multiple crowns)

  
 www.genocan.eu

  
 www.kchrr.com

  
 www.ridgeback-database.com

  
 www.vri.cz

## Acknowledgements:

- Nicolette Salmon Hillbertz *et al.*,  
Upsala University, Sweden



- Czech Rhodesian Ridgebacks Breeding Club (KCHRR)  
(Sarka Stusakova – Head of advisers to breed)

  
 KCHRR

- Ridgeback International (Stephanie Muller)

  
 RIDGEBACK INTERNATIONAL