Uncovering the genetic background of porcine congenital splay leg syndrome in piglets

Jaro De Kort, **Wim Gorssen**, R. Meyermans, S. Janssens and N. Buys

Session 8 “Innovative approaches to pig and poultry production”

CABG Center for Animal Breeding and Genetics

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Porcine congenital splay leg syndrome (PCS)

Three phenotypes

Prevalence <1% to 8%

Treatment is laborious

Mortality up to 50%

→ Surviving piglets recover within 7 days!

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Pathophysiology

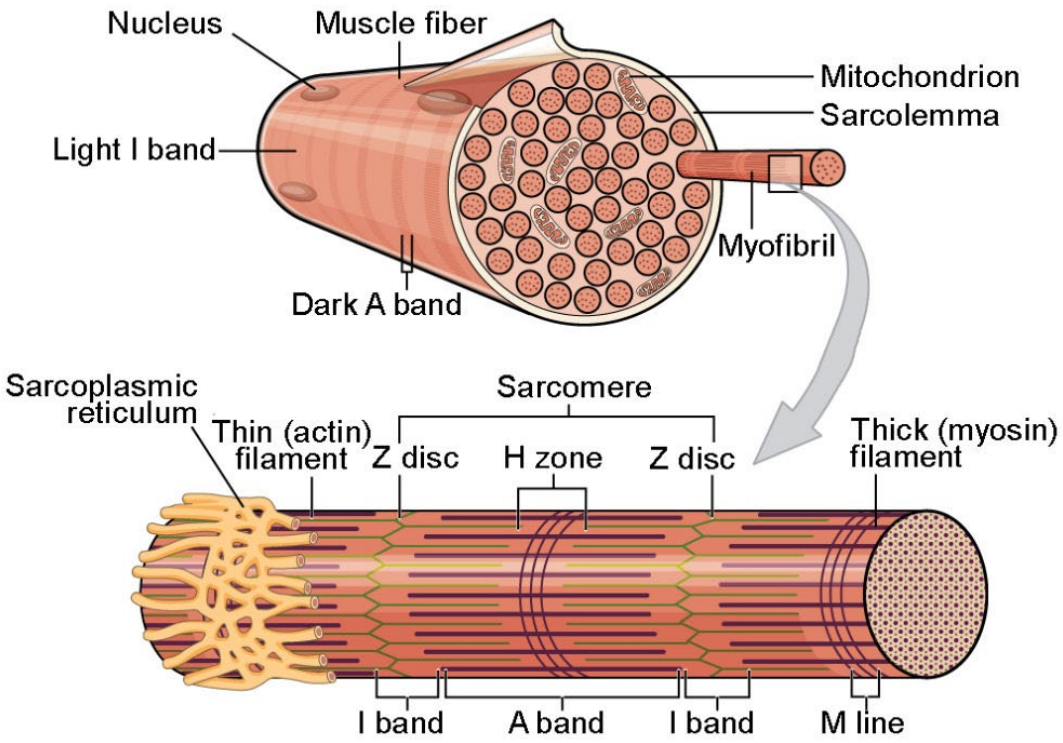
Myofibrillar hypoplasia

- reduced number of myofibrils
- small-diameter myofibrils
- distribution pattern

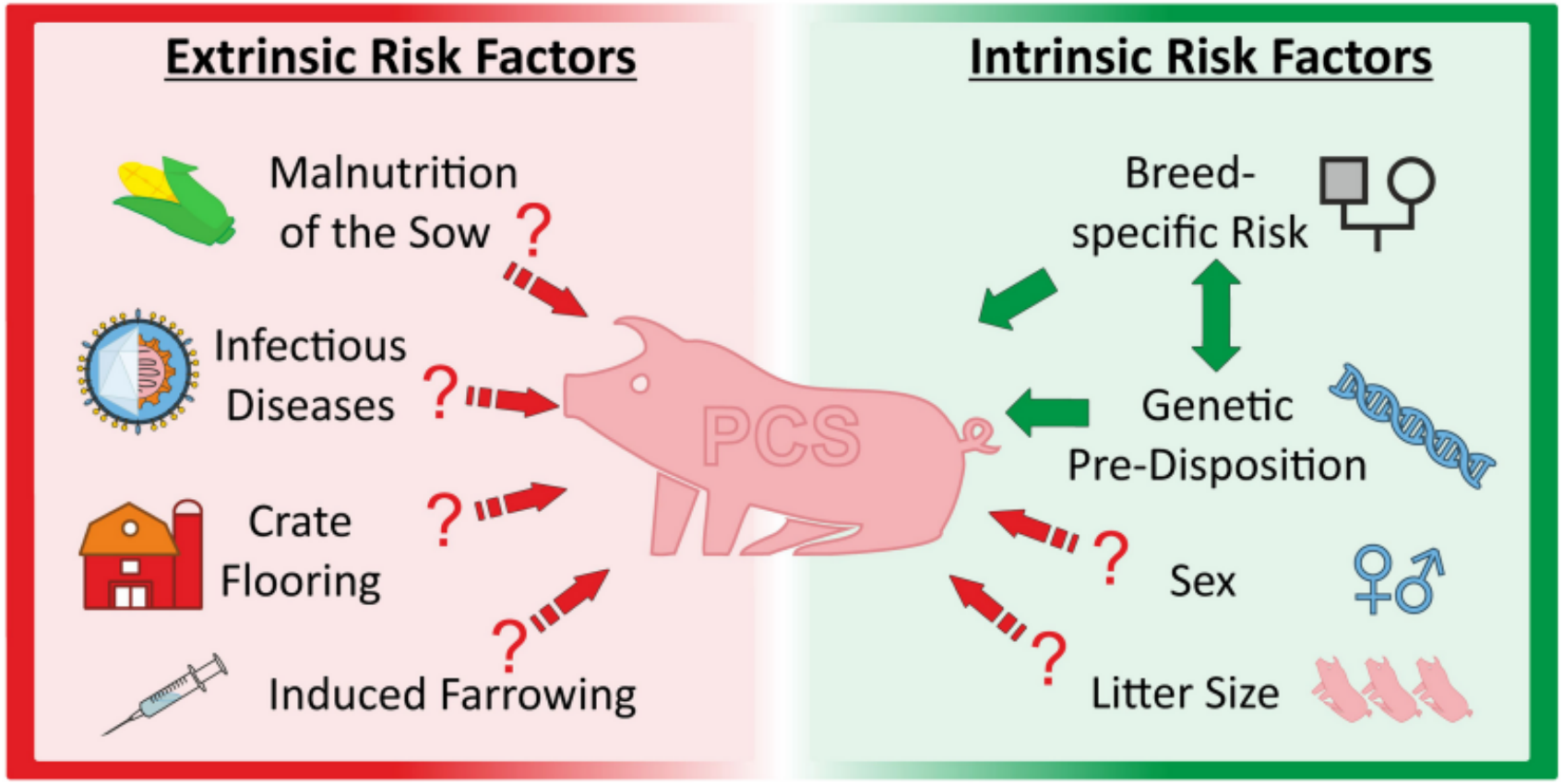
Delayed transition of type II ➔ type I

Neuromuscular differences

- Linked to diet



Etiology



Schumacher, T., Röntgen, M., & Maak, S. (2021). Congenital splay leg syndrome in piglets current knowledge and a new approach to etiology. *Frontiers in veterinary science*, *8*, 609883.

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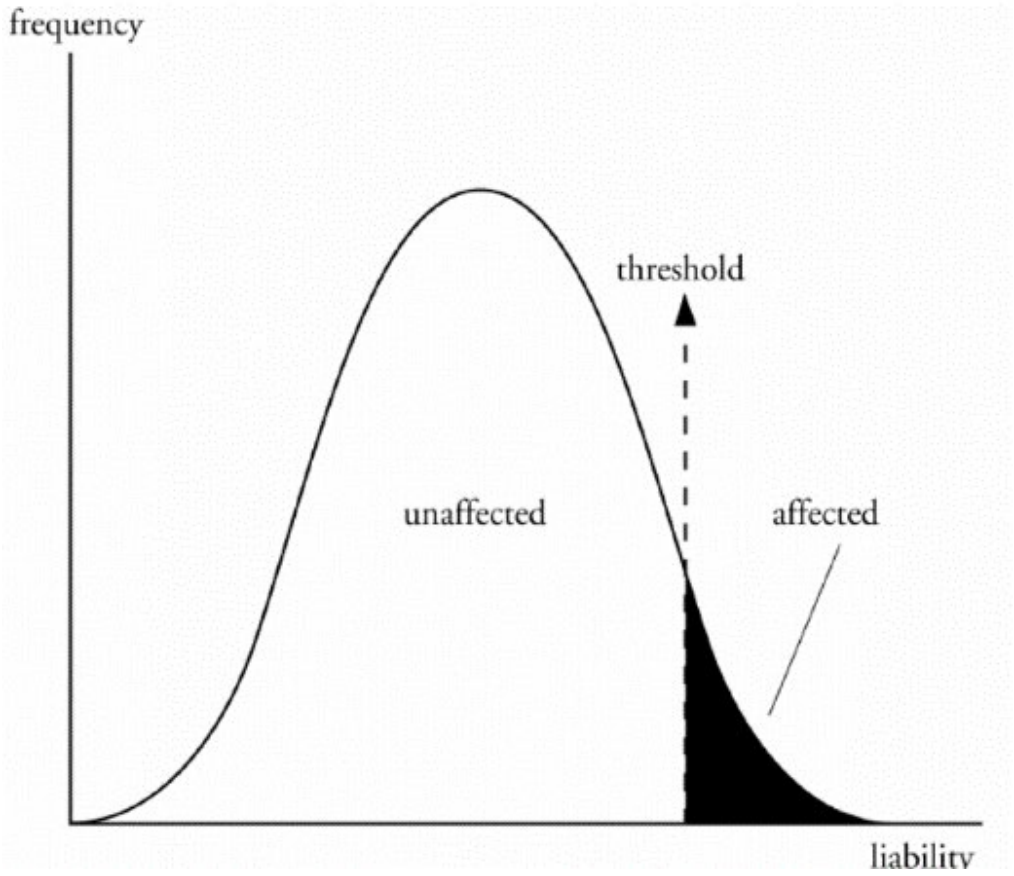
Polygenic threshold model



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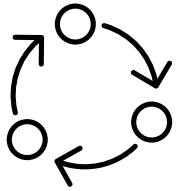
Research objectives



1. Investigate environmental factors impacting PCS

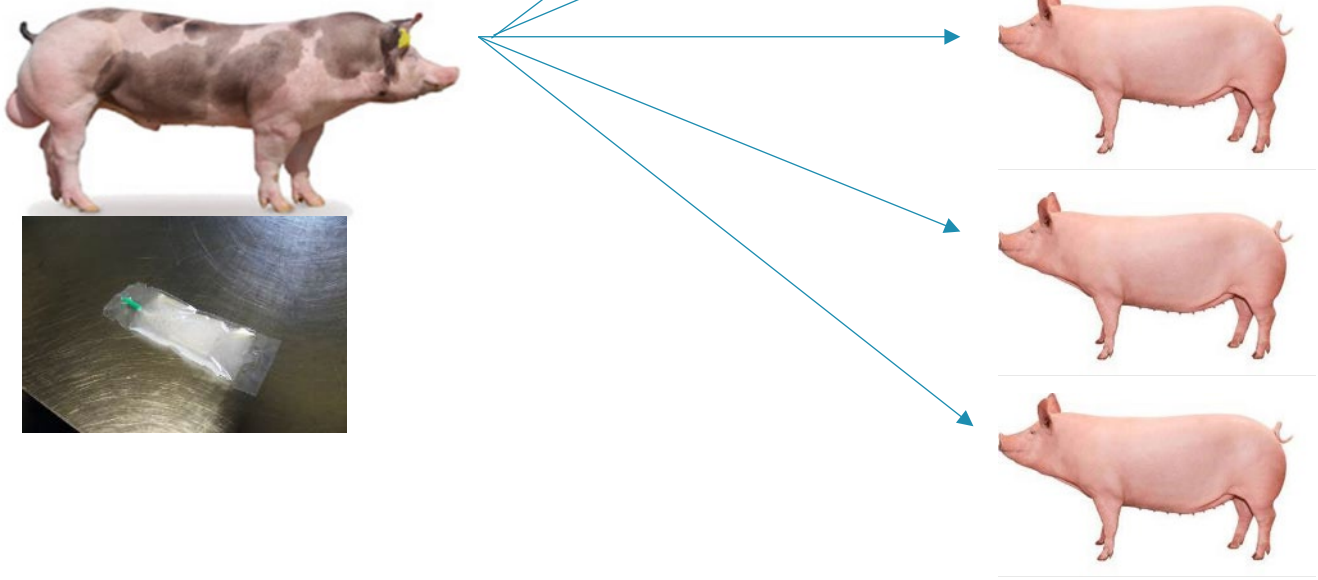
2. Estimate genetic parameters for PCS

3. Genome wide association study to uncover genetic variants

Material

Progeny testing scheme

- Commercial sows inseminated
- Piétrain sires genotyped (50K SNP chip)
- Progeny scored at litter level



Dataset

Trait	Number of records
Scored litters	12,759
Piétrain sires	2,072
Genotyped sires	1,317
Hybrid sows	3,965
Farms	3

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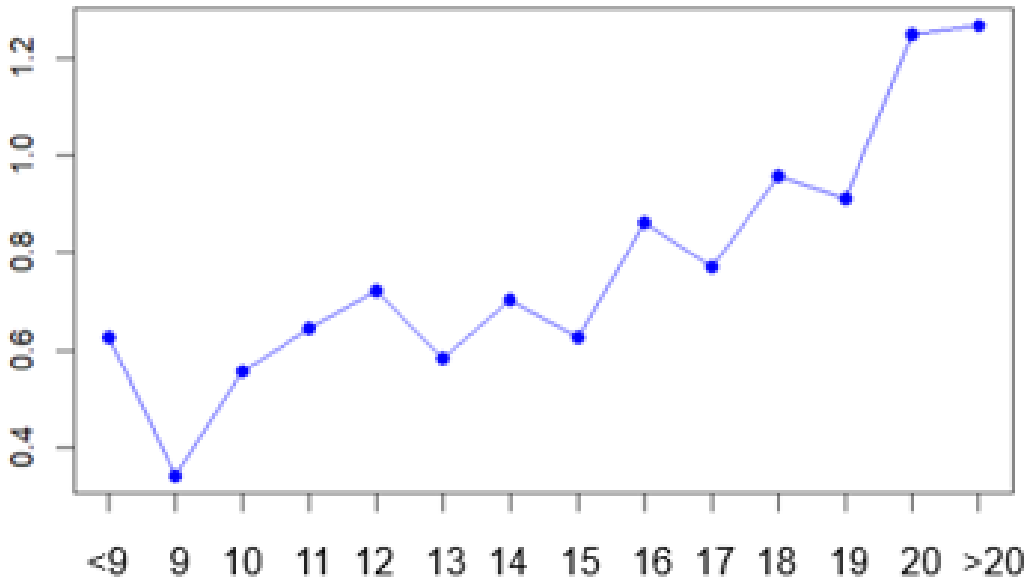
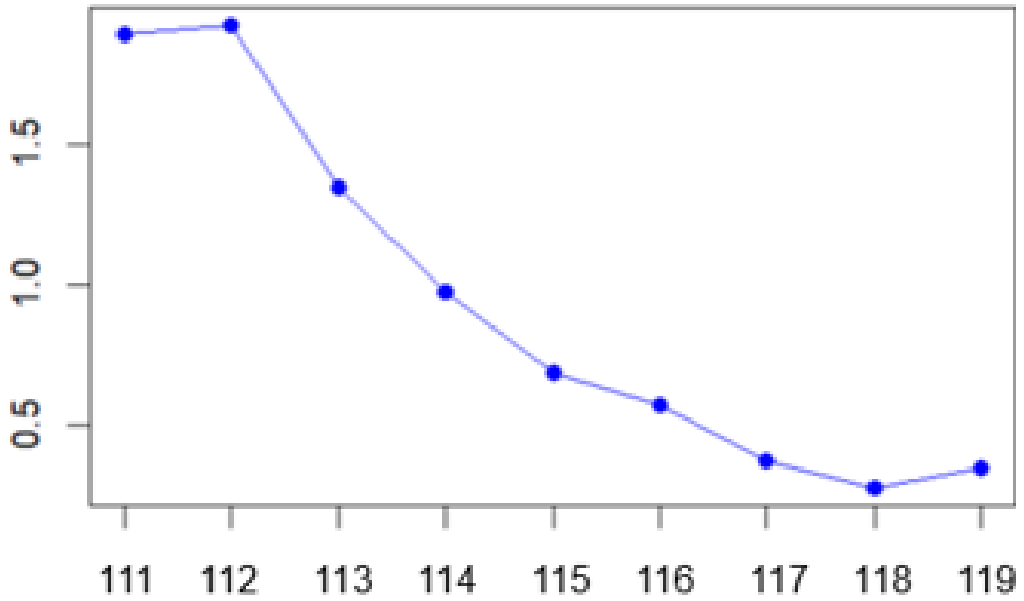
Environmental factors influencing PCS

PCS significantly associated with litter size and gestation length

PCS prevalence in %



Litter size



Gestation length (days)

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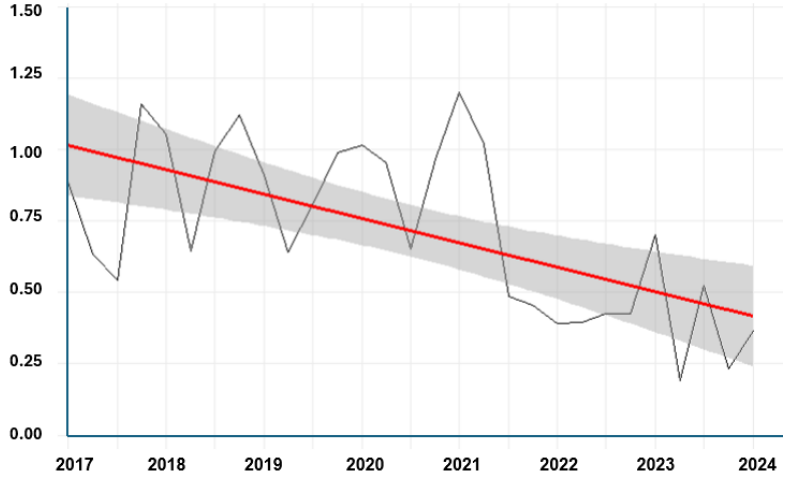
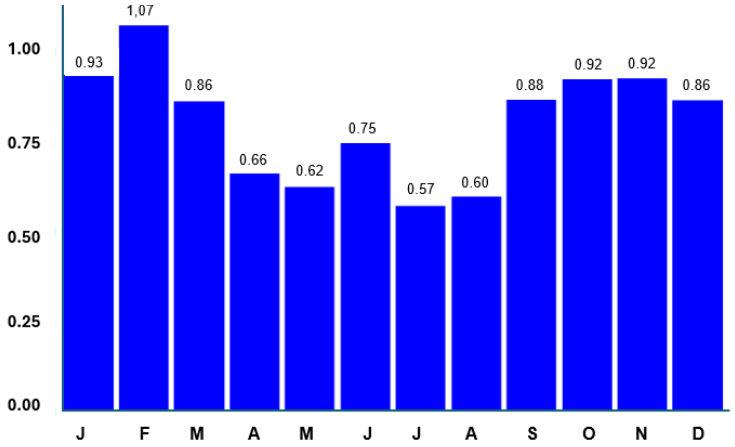
Environmental factors influencing PCS

Clear seasonal fluctuations – More PCS at first parity

PCS prevalence in %

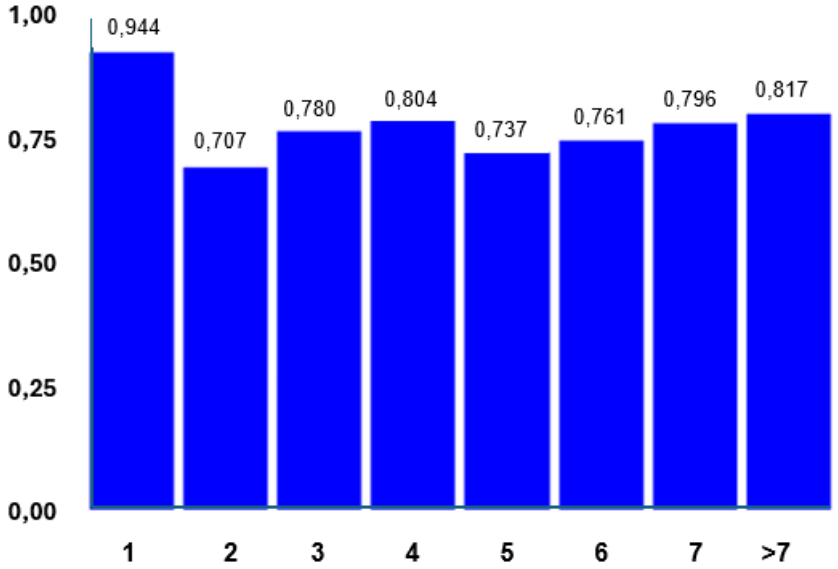


PCS prevalence in %



Month of the year

PCS prevalence in %



Sow parity

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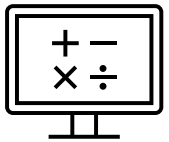
Genetic parameters animal and sire model



Heritability estimate of 11-31%

- Prospects for selection
  - Session 22: **14:45** **Validation of a Piétrain sire breeding program on crossbred piglets' vitality and congenital defects**
    W. Gorssen, C. Winters, R. Meyermans, L. Chapard, K. Hooyberghs, J. Depuydt, S. Janssens, N. Buys

- Variance explained by sow (σ_d^2) ±5 times larger than sire variance (σ_a^2)
  - Genetic and maternal effects!

	Heritability	σ_a^2	σ_d^2	σ_e^2
Animal Model	0.31	0.061	-	0.134
Sire model	0.11	0.005	0.025	0.161

Genome wide association study

Different approaches tested in PLINK and GCTA

- Case-control
- Quantitative

Biggest issues: how to set thresholds?

- Minimum amount of progeny per sire, minimum prevalence of PCS,…

Example:

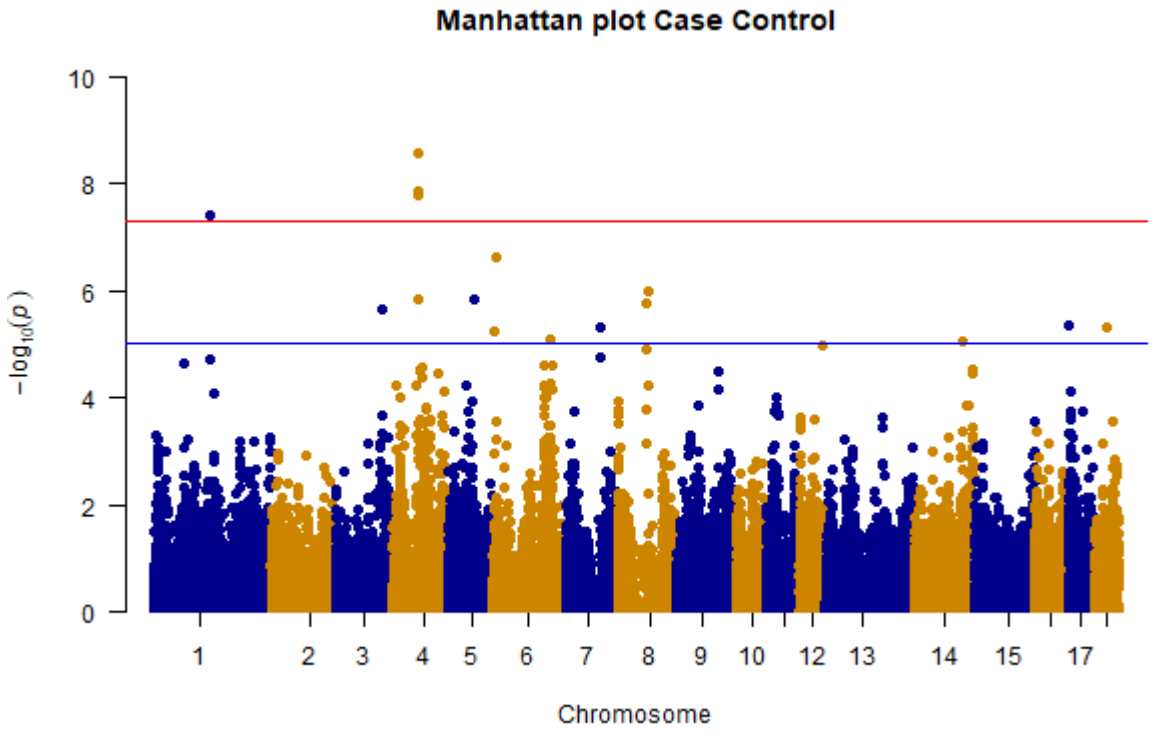
Case: 50 offspring with at least 4% prevalence: N=30

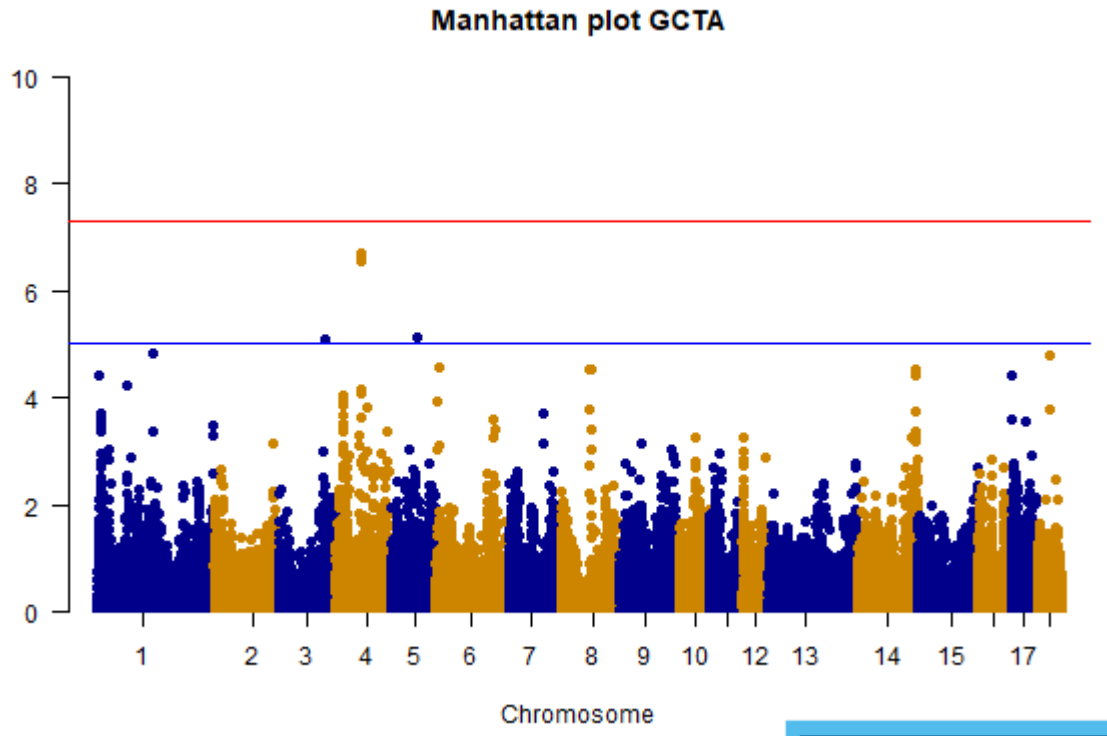
Control: 75 offspring with 0% prevalence: N=339

Case-control analysis gives putative signals

Strongest signal on SSC4

Effect decreases after correction for GRM and 4 first PCS in GCTA





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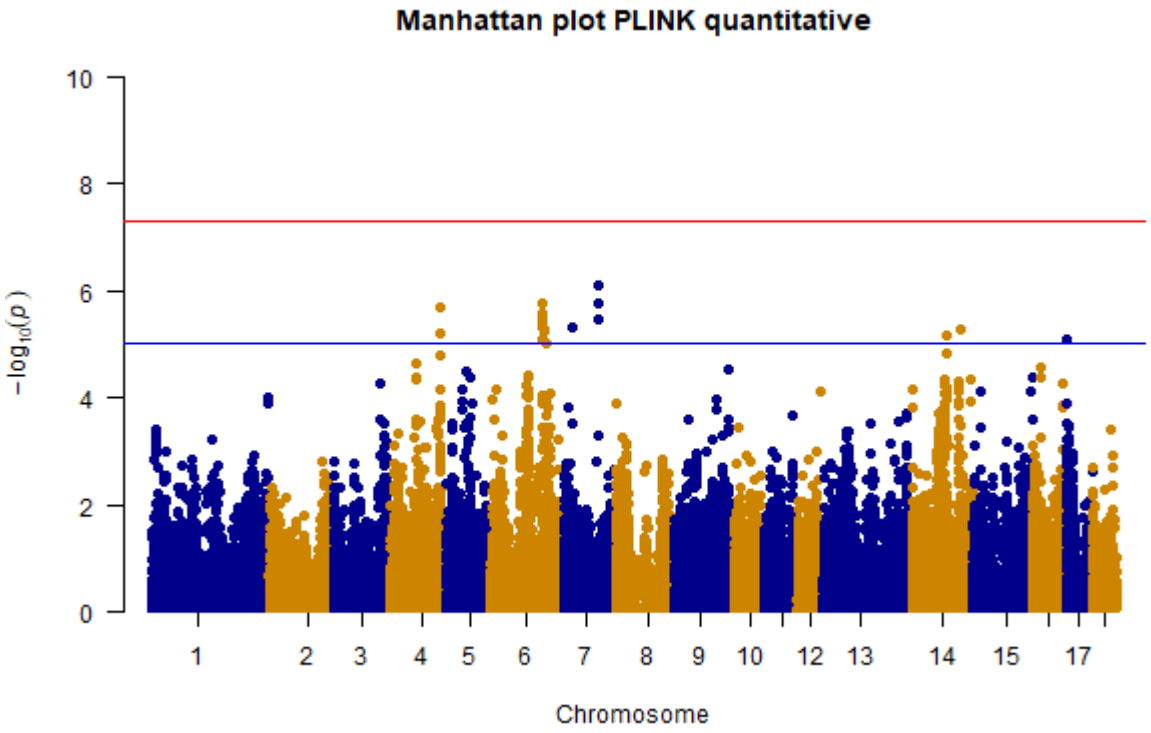
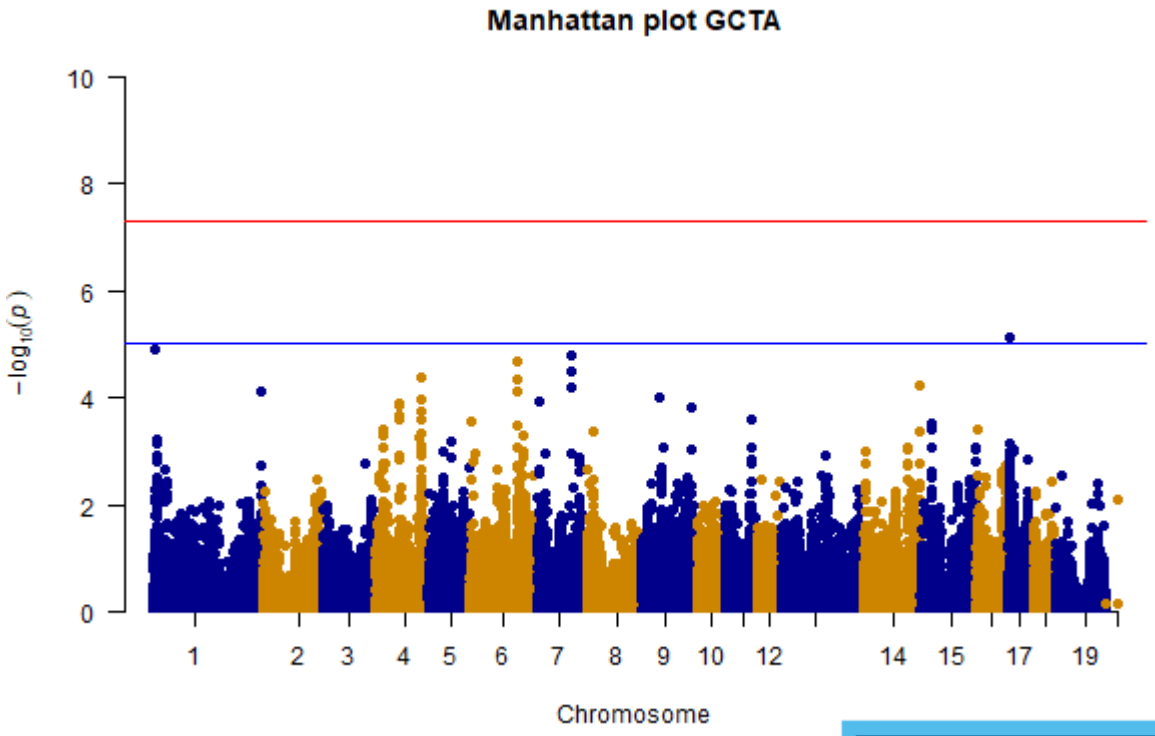
Quantitative analysis gives less significant results

Previous signal on SSC4 just below threshold





Take-home message

PCS has an underlying genetic basis, although environmental factors also have a substantial impact

- Litter size, parity and gestation length
- Seasonality: more PCS in winter and autumn
- Heritability of 11-31%

Genome wide association study gives several associated genetic regions

- Region on SSC4 as potential candidate gene

More research needed to confirm this and increase power:

- Genobreed project will start 01/10/2024
- PhD-student Jaro will present future results himself in Innsbruck ☺

wim.gorssen@kuleuven.be – jaro.dekort@student.kuleuven.be

Acknowledgments

- Data providers

- Funding

Research Foundation Flanders
Opening new horizons

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wim.gorssen@kuleuven.be – jaro.dekort@student.kuleuven.be

Thank you for your attention!

wim.gorssen@kuleuven.be
jaro.dekort@student.kuleuven.be

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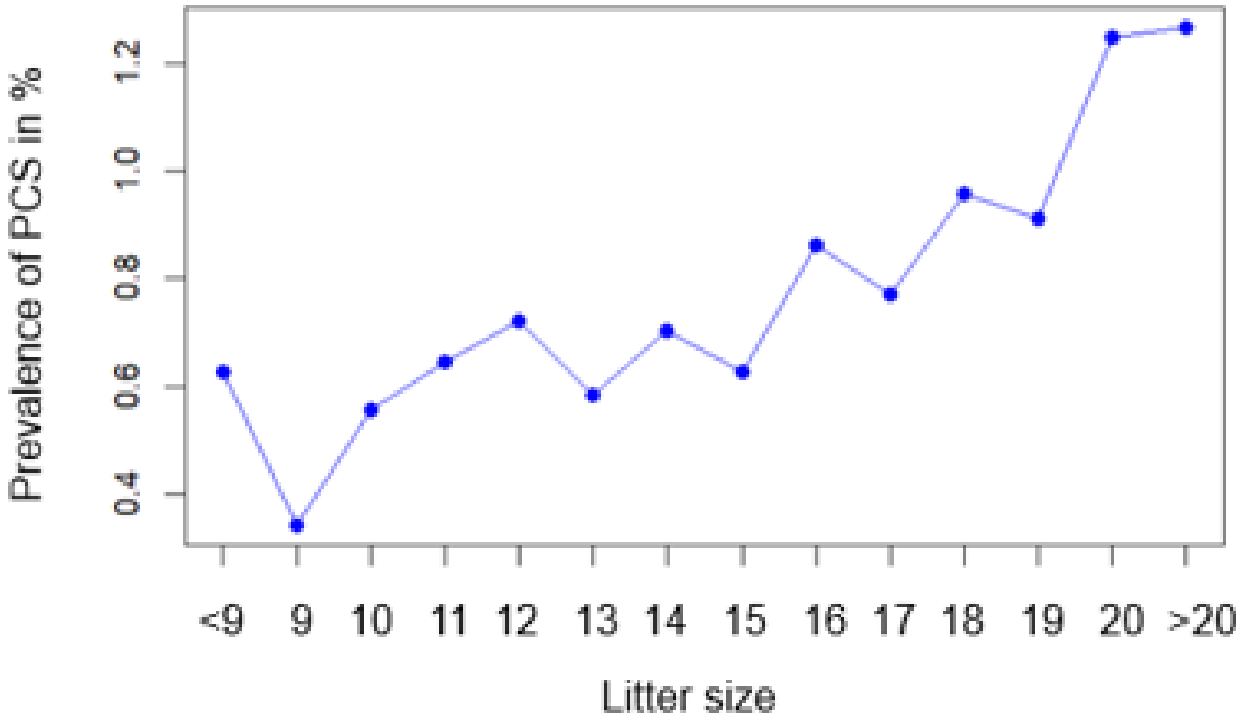
1. Invloed omgeving en maternale factoren

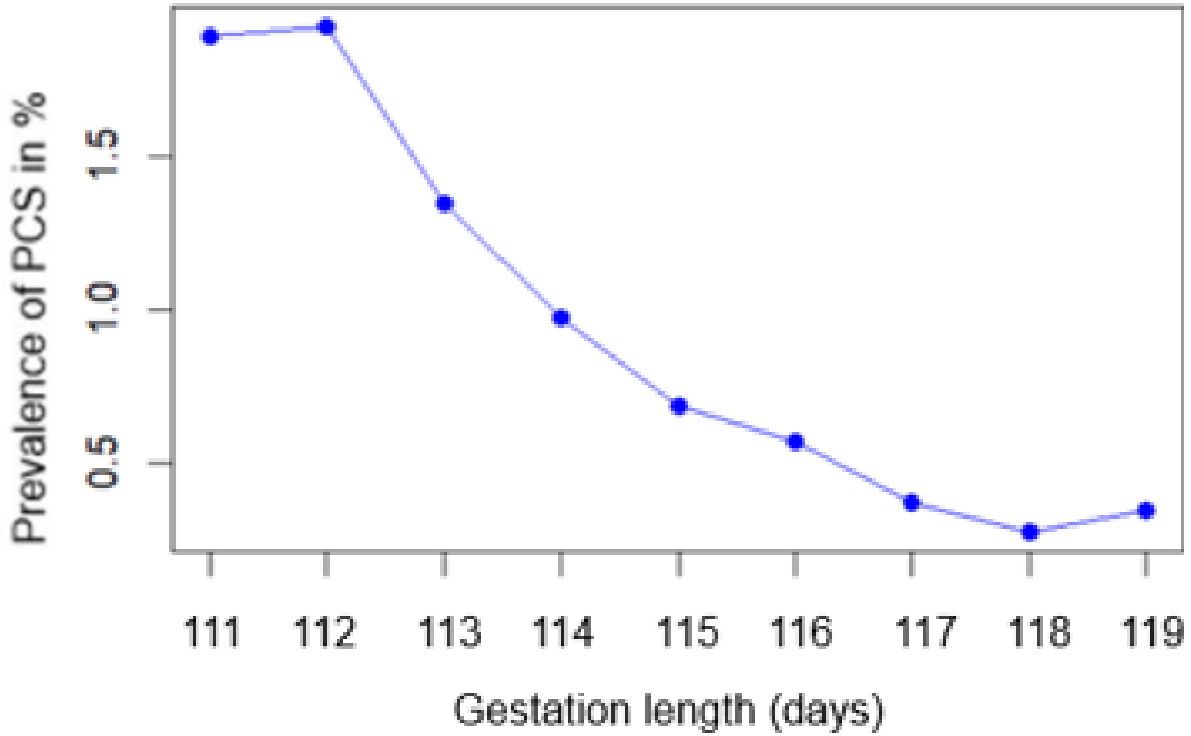
Worpgrootte



	Effect	p-waarde
SR	+0,02	2e-16***
MR	+0,01	5e-6***

1. Invloed omgeving en maternale factoren

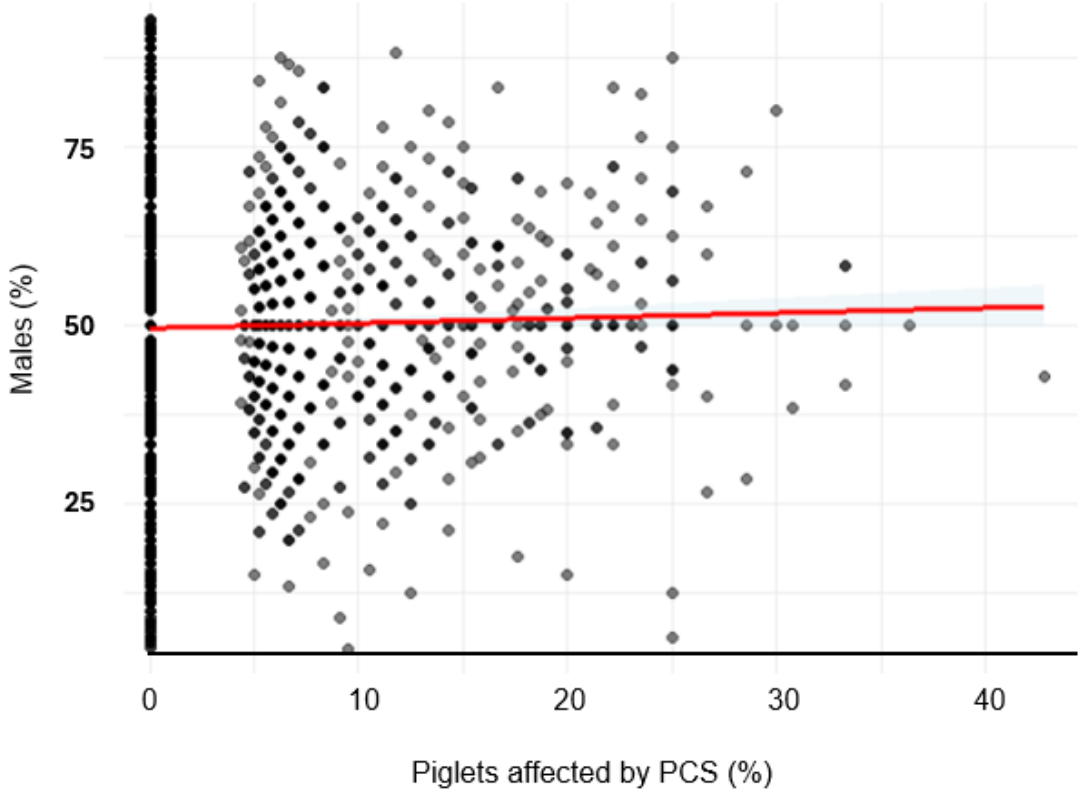
Drachtduur



		Effect	p-waarde
SR	d	-1,28	3e-4***
	d^2	+0,01	5e-4***
MR	d	-1,21	0,003**
	d^2	+0,01	0,004**

1. Invloed omgeving en maternale factoren

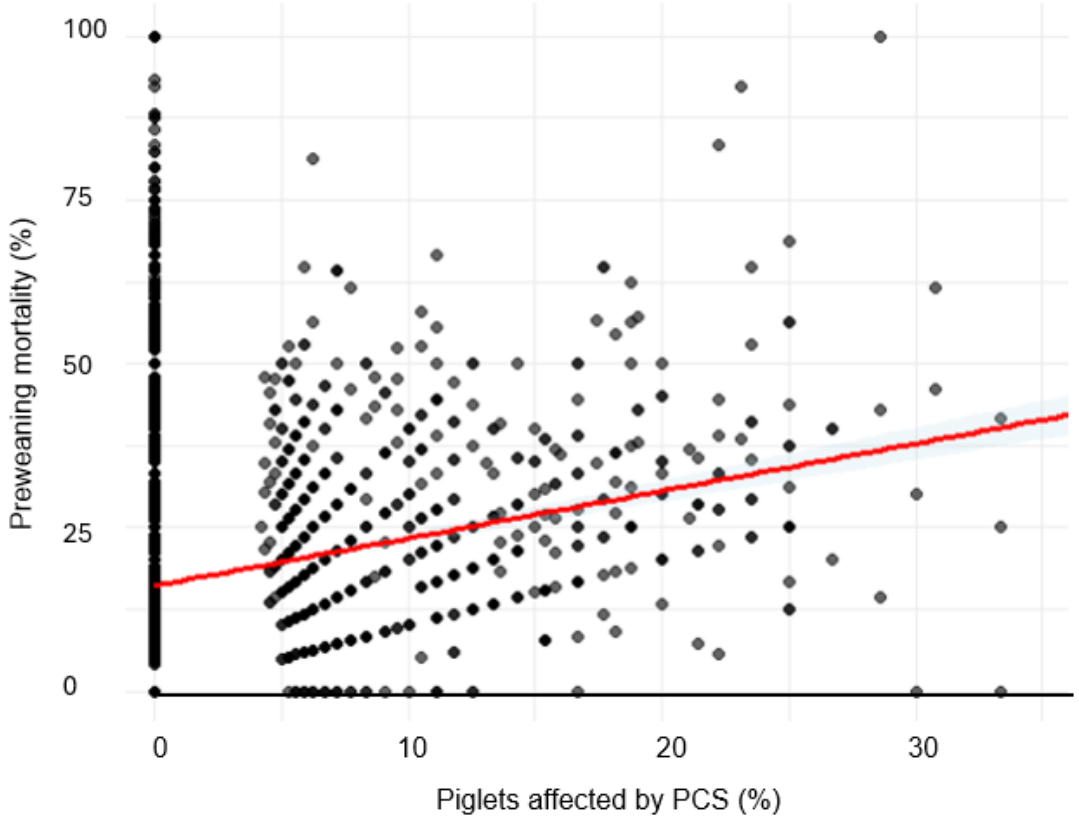
Geslacht



	Effect	p-waarde
SR	+0,00	0,08
MR	+0,00	0,45

1. Invloed omgeving en maternale factoren

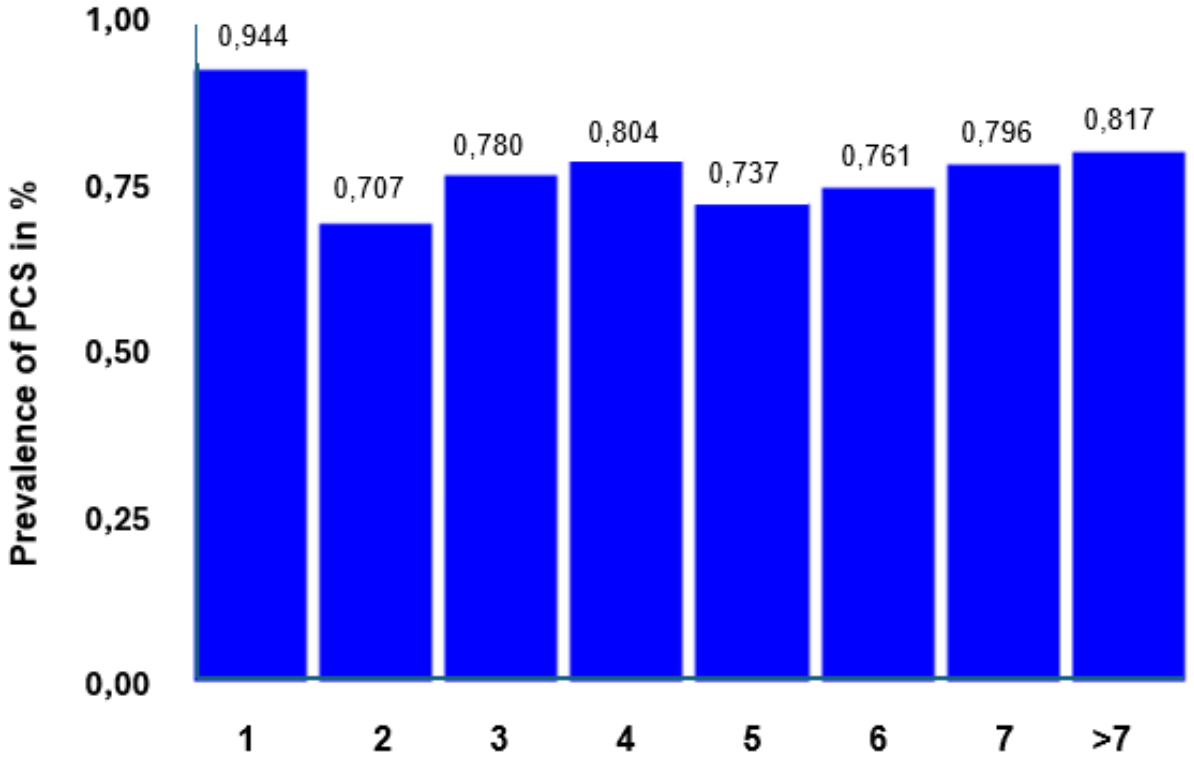
Sterfte voor spenen



	Effect	p-waarde
SR	+0,01	2e-16***
MR	+0,01	2e-16***

1. Invloed omgeving en maternale factoren

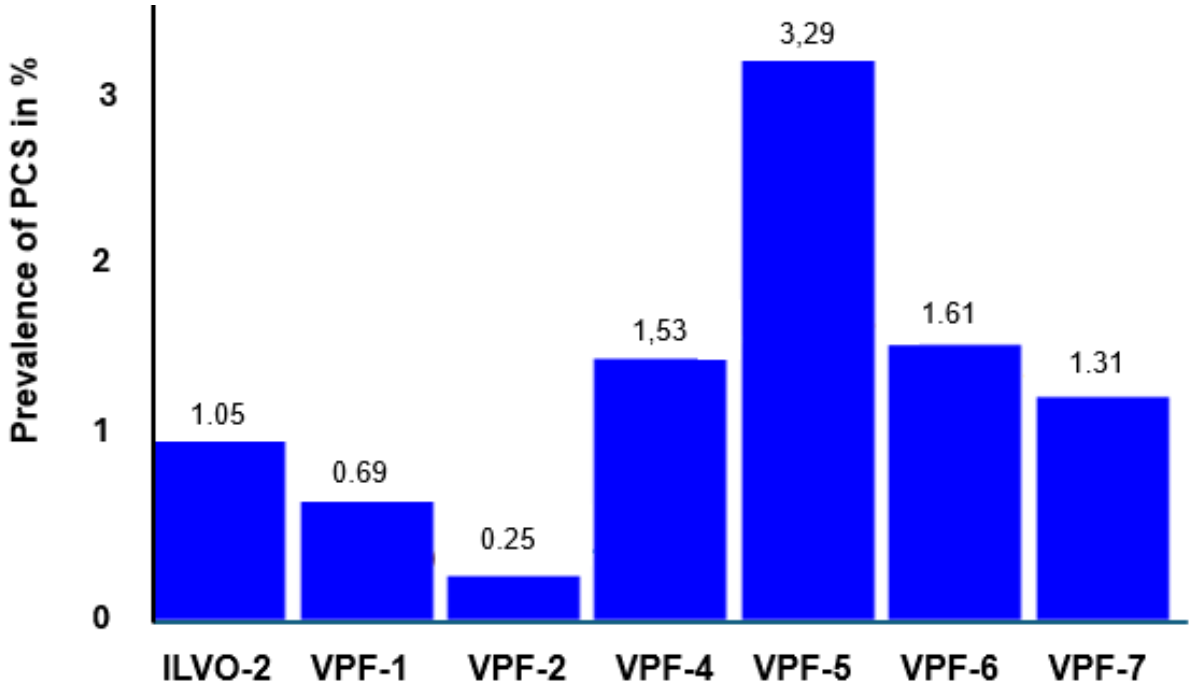
Pariteit



	SR		MR	
	Effect	p	Effect	p
C1	Ref	-	Ref	-
C2	-0.04	0.01 *	-0.02	0.29
C3	-0.02	0.21	-0.05	0.009**
C4	-0.01	0.43	-0.04	0.02*
C5	-0.02	0.15	-0.08	1.5e-5***
C6	-0.02	0.21	-0.07	1.1e-4***
C7	-0.02	0.33	-0.06	0.002**
C8	-0.00	0.92	-0.02	0.35
C9	-0.04	0.07	-0.08	0.002**

1. Invloed omgeving en maternale factoren

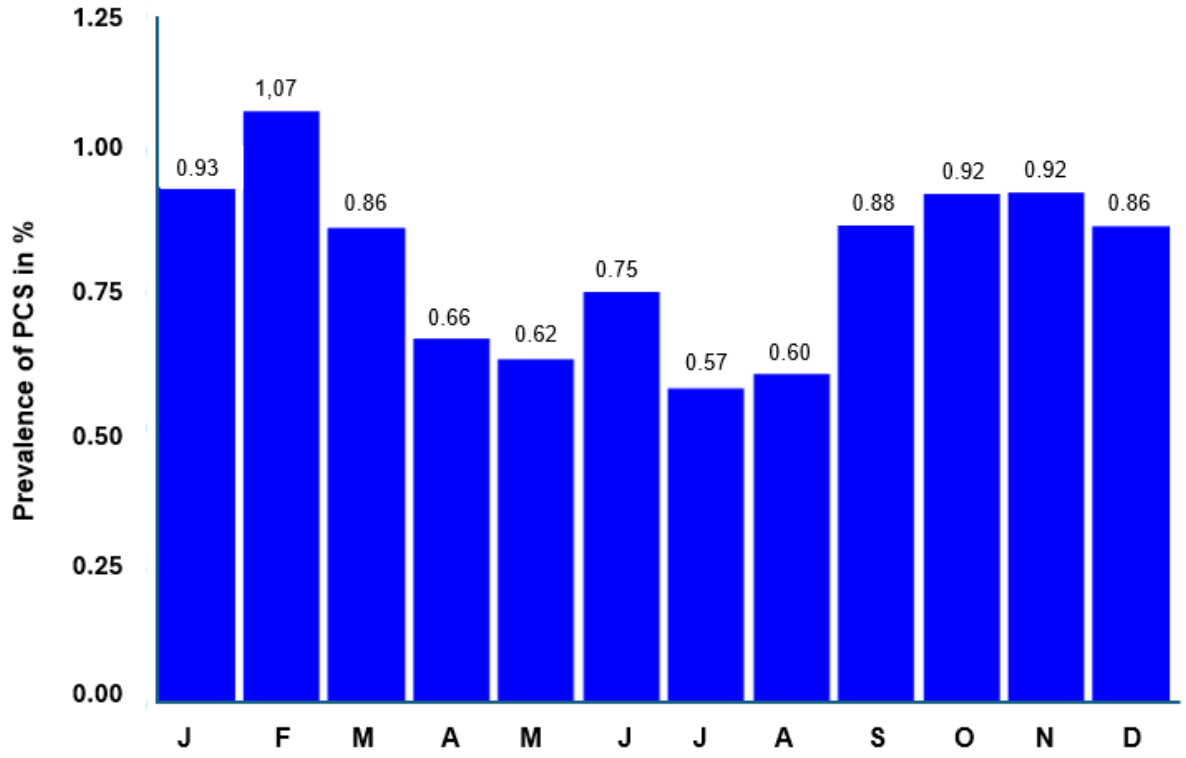
Bedrijf



	SR		MR	
	Effect	p	Effect	p
VPF1	Ref	-	Ref	-
ILVO2	+0.07	1.8e-10***	-	-
VPF2	-0.06	2,8e-7***	-0.14	2e-16***
VPF4	+0.14	4.9e-11***	+0.08	1.9e-4***
VPF5	+0.27	2.0e-11***	+0.33	6.7e-16***
VPF6	+0.13	1.1e-9***	+0.09	2.6e-5***
VPF7	+0.07	0.003**	+0.09	4.0e-4***

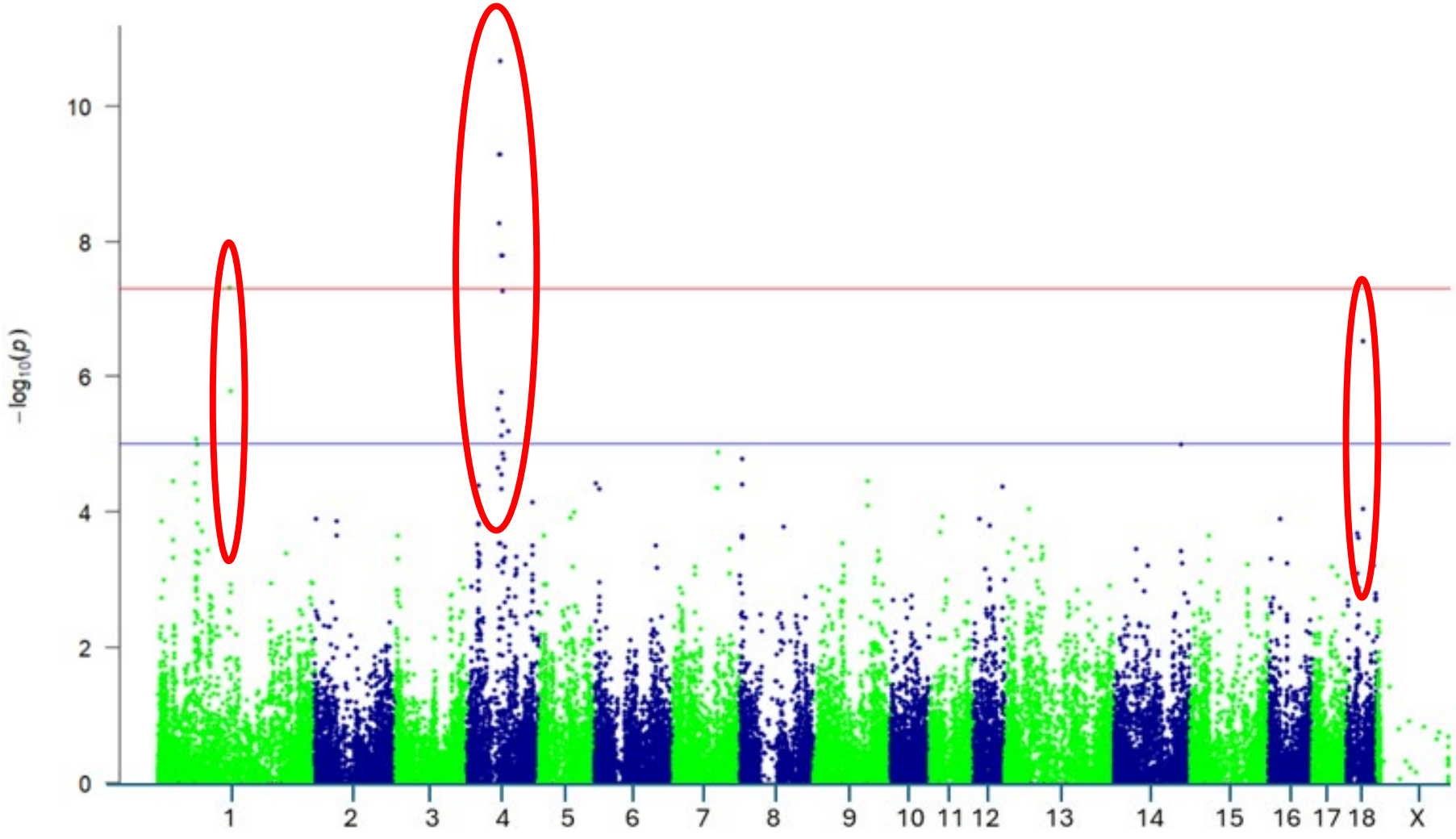
1. Invloed omgeving en maternale factoren

Seizoen



	SR		MR	
	Effect	p	Effect	p
Winter	Ref	-	Ref	-
Spring	-0.03	2e-3**	-0.03	0.03*
Summer	-0.05	3e-5***	-0.03	0.02*
Autumn	-0.00	0.41	+0.01	0.62

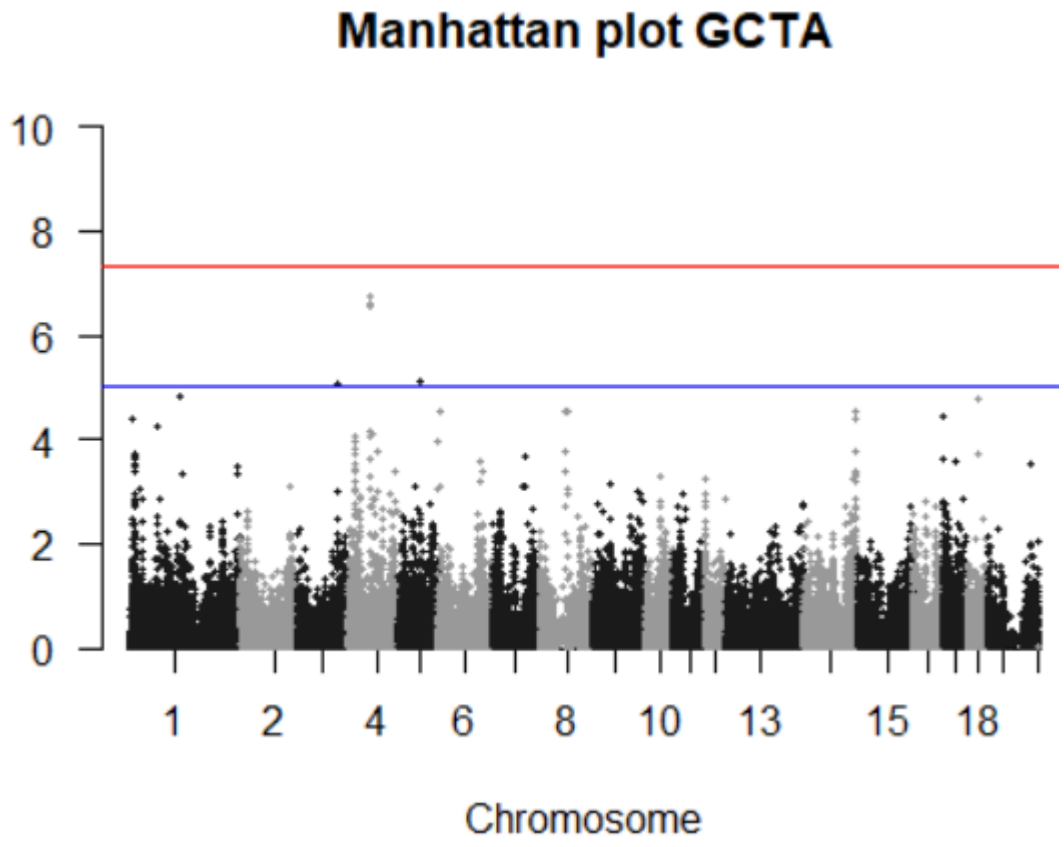
Case-control approach PLINK

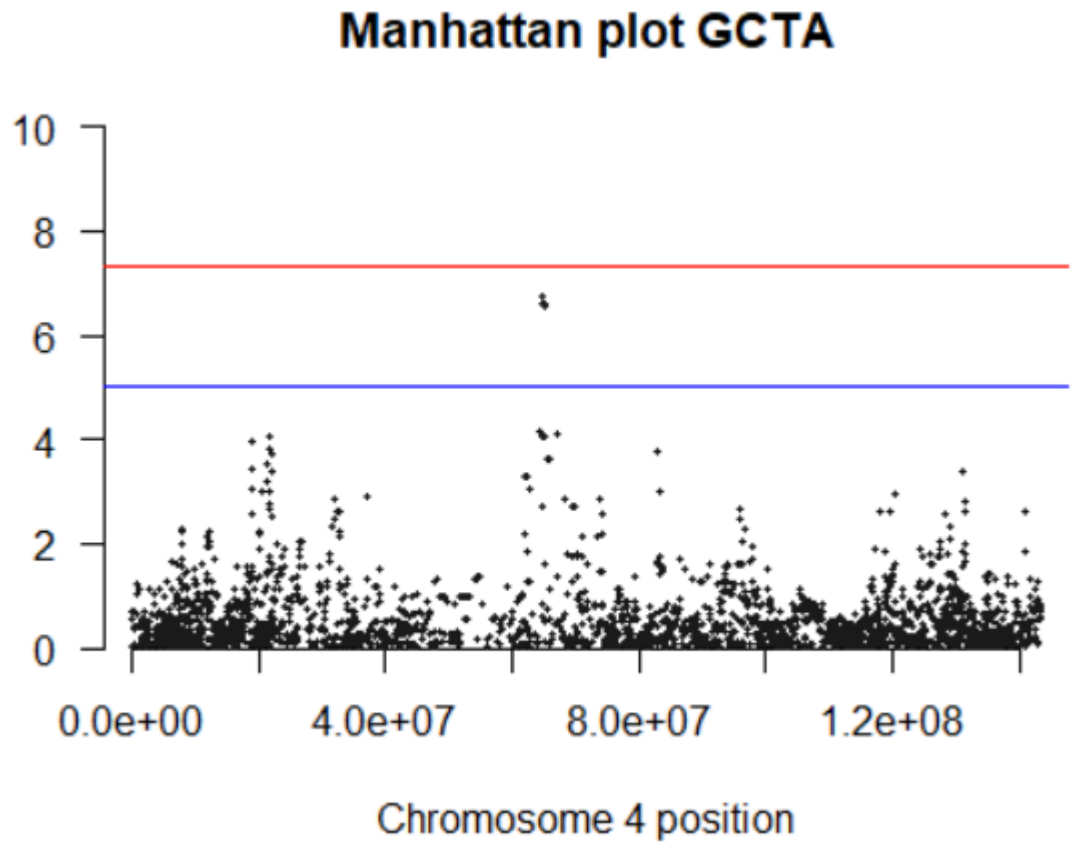


Case-control GCTA

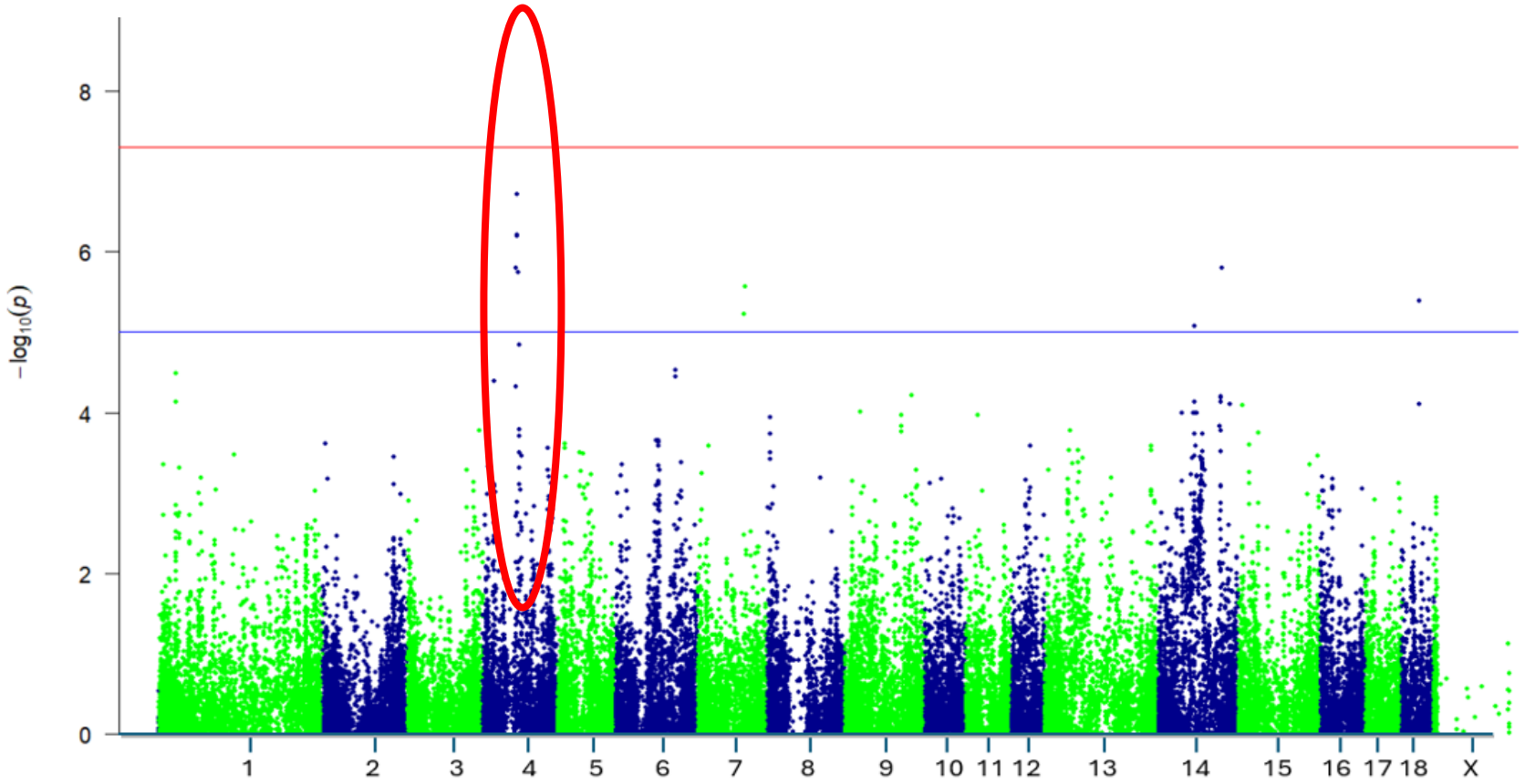
including GRM and correction of first 4 PCs

Same region in SSC4 pops up, just below threshold



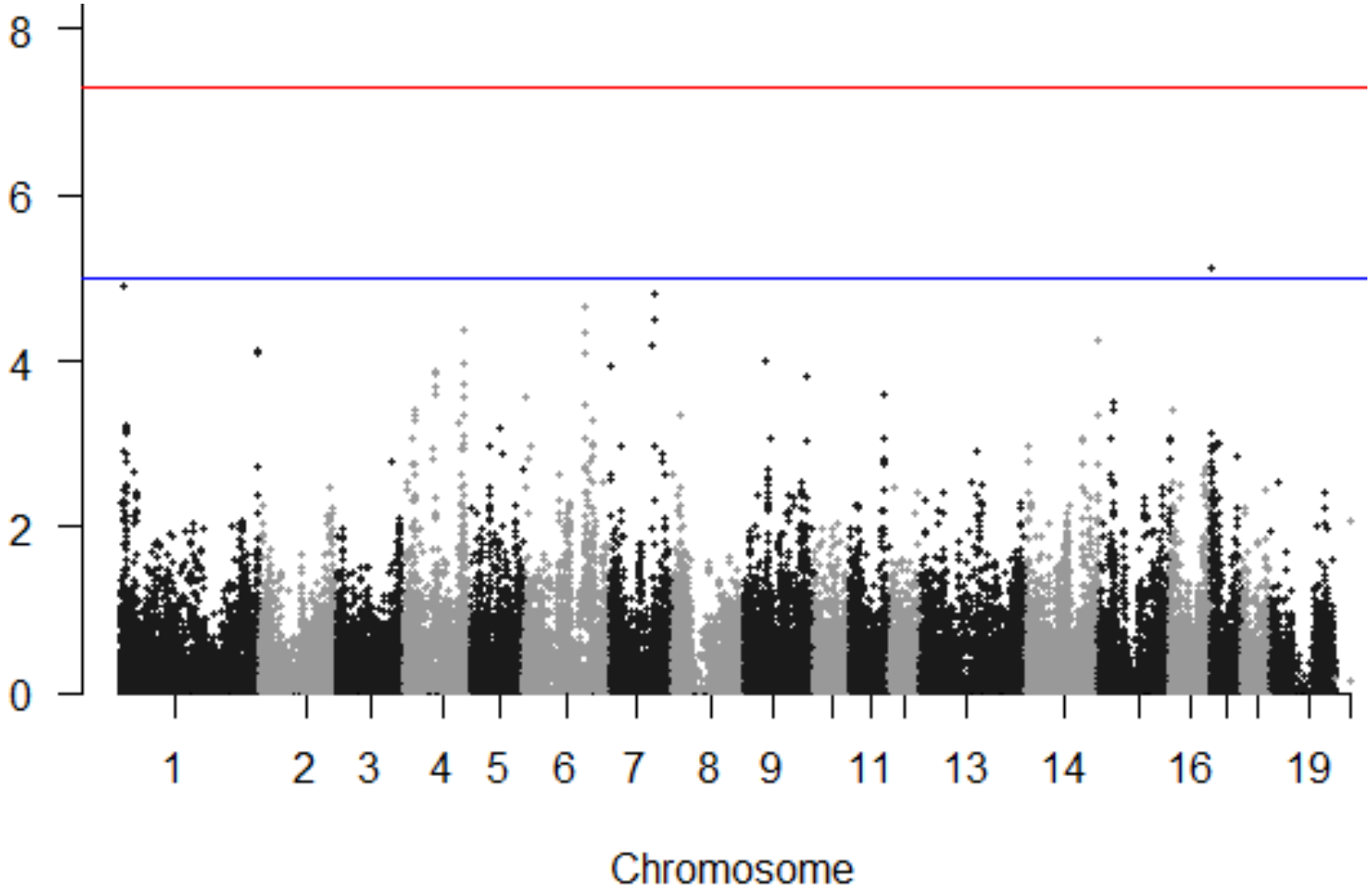


Quantitative GWAS in PLINK



Quantitative GWAS in GCTA

Region on SSC4 below threshold



The 75th EAAP Annual Meeting
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