



*75th EAAP Annual Meeting,
Sep 1st – 5th 2024*



Session 48 “Horse genetics and genomics”, Sep 3 8:30-12:30
Abstract No. 2213077

Development and Genetic Architecture of White Markings in Heavy Warmblood Horses

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Introduction – White markings (WM)

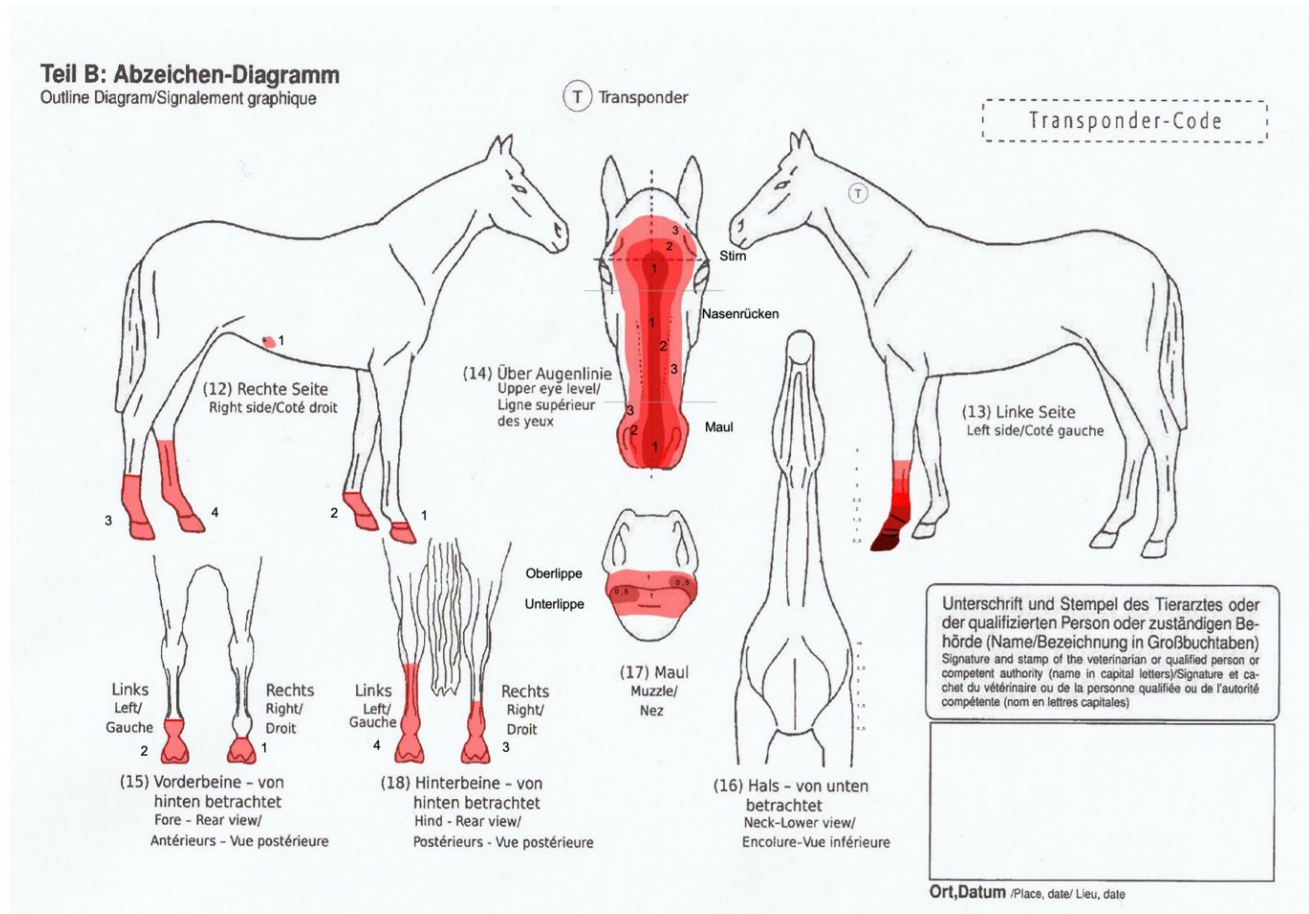
- WM are widespread in domestic horses
- WM are physiologically of high interest because of their early point of origin in embryonic development
- Extensive white markings or spotting can be negatively associated with vitality and health
- Has the proportion of white markings increased in the Saxon-Thuringian heavy warmblood (STHW) over the last 20 (or 100) years ? -> Poster session abstract no. 2212748
- Which genomic regions are relevant in this breed?



Costello *2022, EBV: 159

Material and Methods

- A quantitative WM score was derived from written WM descriptions based on Rieder et al. (2008) for STHW born between 2000 and 2023 (N = 9,519).



Costello *2022, EBV: 159

Material and Methods

- A quantitative WM score was derived from written WM descriptions based on Rieder et al. (2008) for STHW born between 2000 and 2023 (N = 9,519).
- WM on head, legs, as well as body were scored, summed up, and used as quantitative scores for head (HS), legs (LS) (for legs left/right & front/hind scores were created), and the entire animal (TS). Additionally, binary scores were created by simple transformation: if score>0 then bin=1, else bin=0
- Pedigree information was available with a mean pedigree depth of 5.83 full generations.
- 962 horses were genotyped (Illumina Equine80select), after quality control filters 758 animals had phenotypes and genotypes (~40k of 80k).
- GWAS were performed in GCTA (loco option) and BLUPF90 with year of birth and sex as fixed effects, animal as random effect.

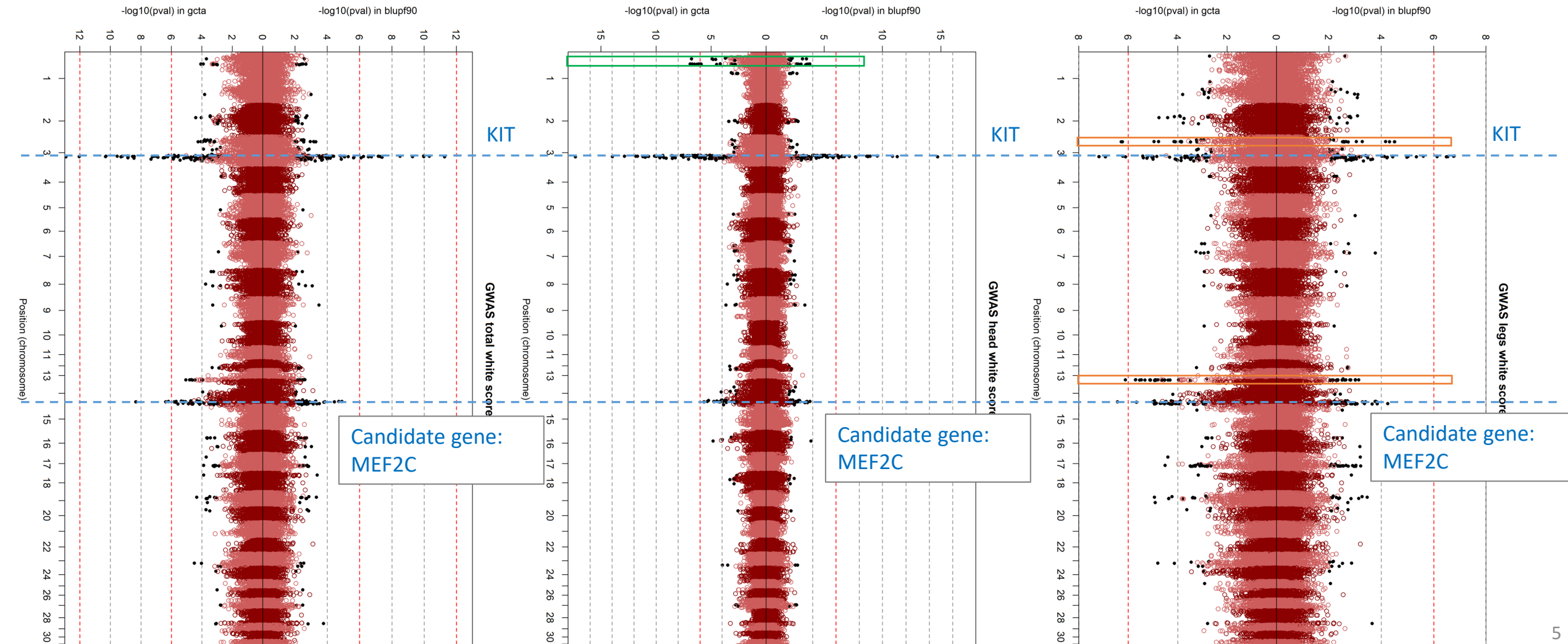
GWAS results – quantitative total, head, and leg scores

(black dots represent the intersection of the 1% strongest SNP of the two programs, left gcta, right blupf90, ref.gen. EquCab2.0)

total score ($h^2=0.71$)

head score ($h^2=0.59$)

leg score ($h^2=0.63$)



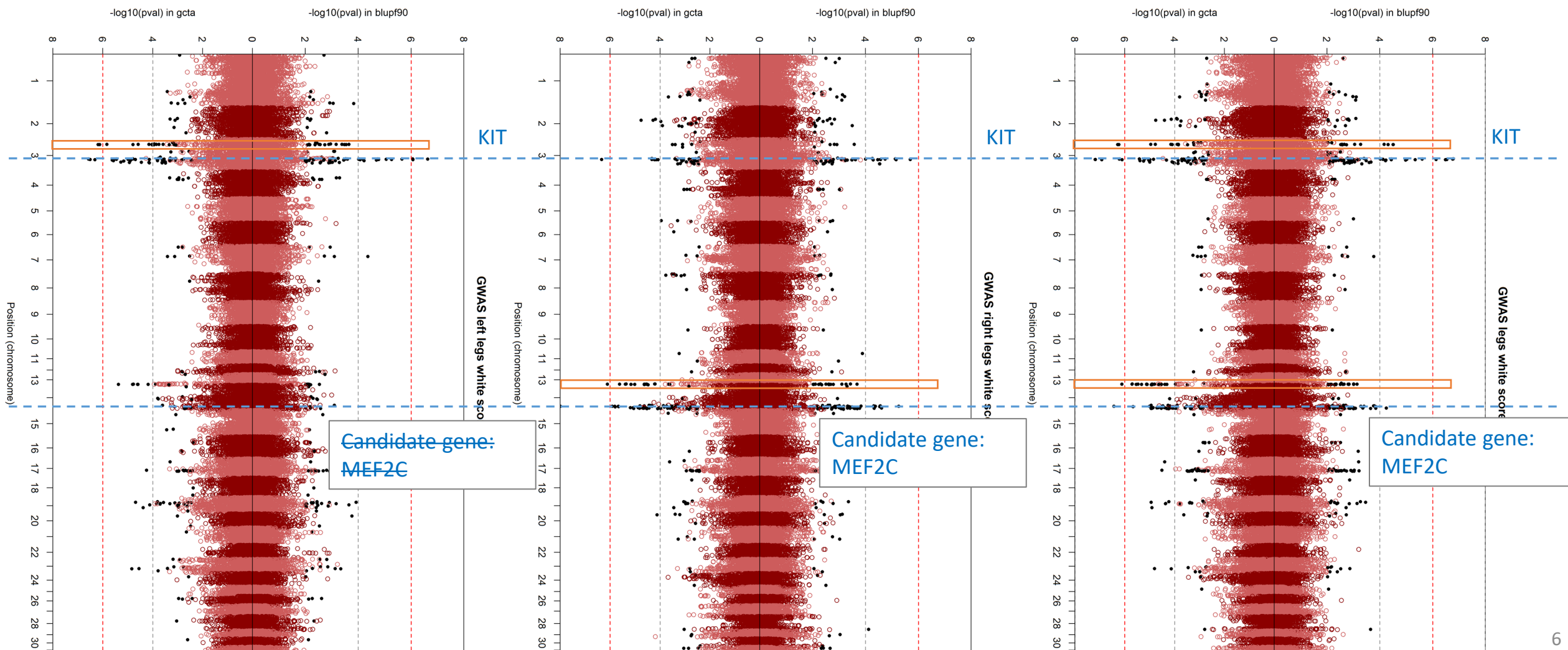
GWAS results – details on quantitative leg scores

(black dots represent the intersection of the 1% strongest SNP of the two programs, left gcta, right blupf90, ref.gen. EquCab2.0)

left leg score ($h^2=0.53$)

right leg score ($h^2=0.48$)

leg score ($h^2=0.63$)



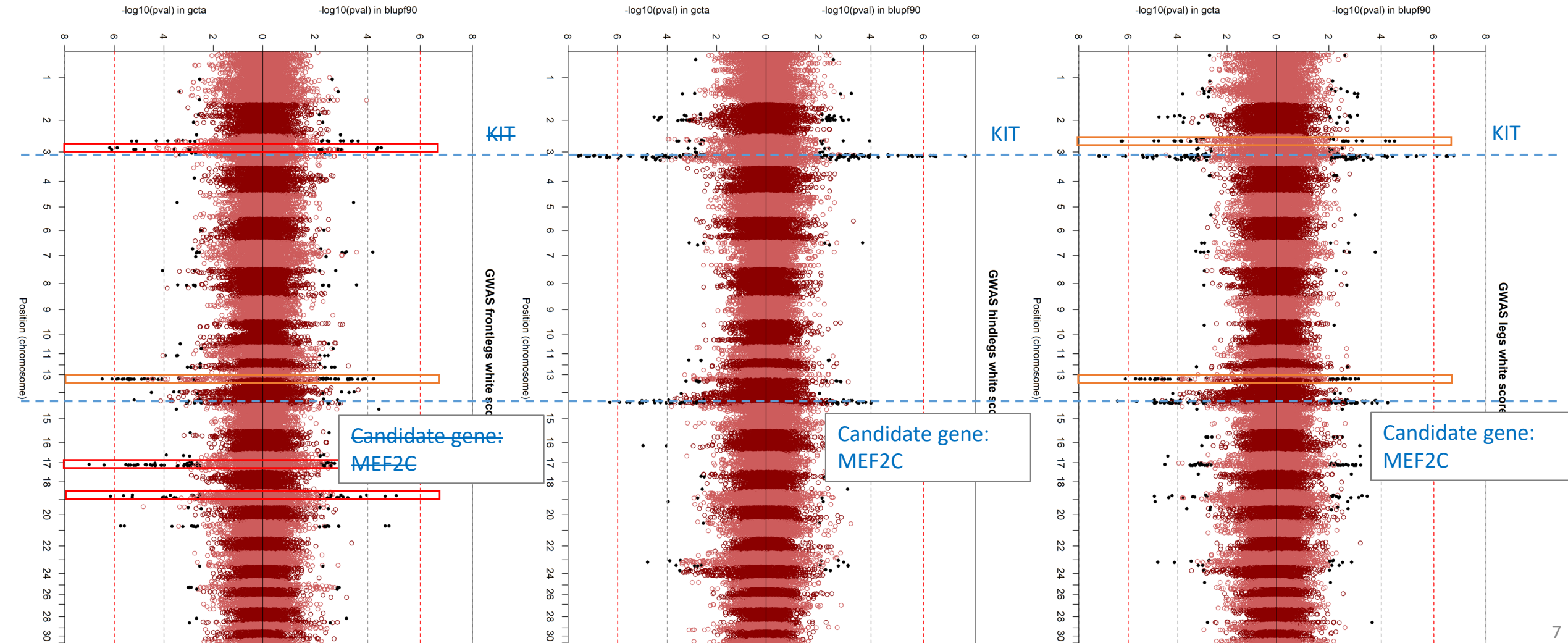
GWAS results – details on quantitative leg scores

(black dots represent the intersection of the 1% strongest SNP of the two programs, left gcta, right blupf90, ref.gen. EquCab2.0)

front leg score ($h^2=0.56$)

hind leg score ($h^2=0.48$)

leg score ($h^2=0.63$)



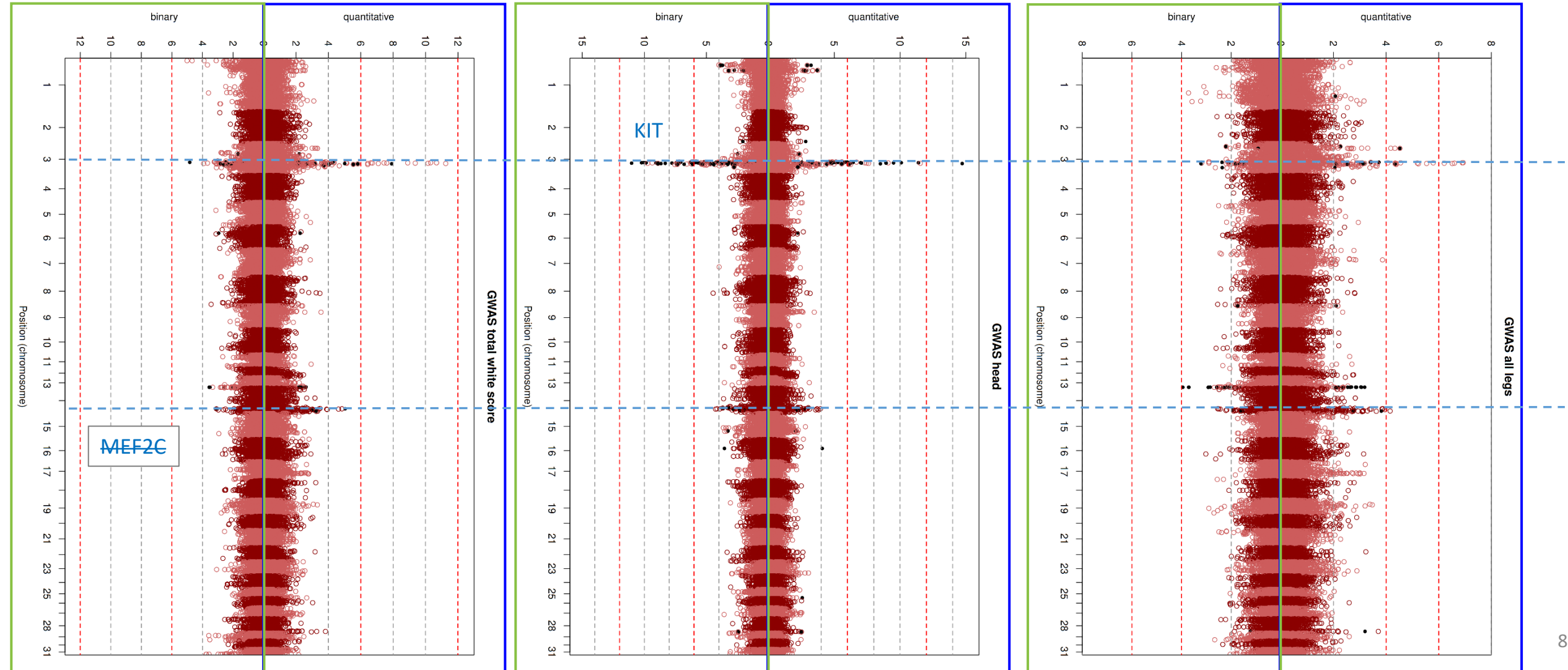
GWAS results – binary scoring versus quantitative scoring

(black dots represent the intersection of the 1% strongest SNP of the two trait definitions, left binary, right quantitative, both with blupf90, ref.gen. EquCab2.0)

total score

head score

leg score



GWAS results – known and newly detected chromosomal regions

ECA	Position bp (Reference genome EquCab 2.0 (3.0))	Traits (white markings)	Known QTL in horses from Animal QTL Database	Candidate genes with connection to coat colour/pigmentation
1	23354753-24571245 (23524813-24740241)	Head	None	
1	39945367-42102289 (40284267-42443246)	Head	Jumping test scores	<i>PTEN</i>
3	18822505-20064543 (19515452-20663374)	All legs, left legs	Osteochondrosis dissecans, navicular bone morphology, white markings	<i>CDH1</i>
3	71145683-87549031 (72731870-89673744)	Total score, all legs, head, hindlegs, left legs, right legs, front legs	Many QTL for white markings (Berber, Franches-Montagne, Spanish horse, Chinese Horse, Noric, Thoroughbred x Jeju, and other) also: female fertility, insect bite hypersensitivity, bone fracture, withers height in ponies	<i>KIT-gene</i> (with W-alleles)
13	38415075-41648568 (39651109-42855398)	Total score, all legs, frontlegs, right legs, left legs	Female fertility	<i>CLUAP1</i>
14	72321492-80034178 (72338680-80056488)	Total score, head, all legs, hindlegs, right legs	Osteochondrosis dissecans	<i>MEF2C</i>

GWAS results – known and newly detected chromosomal regions

ECA	Position bp (Reference genome EquCab 2.0 (3.0))	Traits (white markings)	Known QTL in horses from Animal QTL Database	Candidate genes with connection to coat colour/pigmentation
3	49000259-54723285 (50381340-56195886)	Front legs	None	<i>MAPK10</i>
17	49434295-51671934 (49314284-51550288)	Front legs	QTL for gaitedness (Standardbred) and QTL withers height	<i>EDNRB</i>
19	20040552 (22415959)	Front legs	None	<i>SOX2</i>

Conclusion



- For the STHW the *KIT* gene on ECA3 was identified as the main gene responsible for the expression of WM for the total as well as head and leg scores.
- In addition, another candidate gene (*MEF2C*) on ECA14 which seems to be related to the WM on these parts of the body was found.
- Two regions on ECA1 also seem to be responsible for WM on the head. However, they are not associated with WM of the legs.

Conclusion

- However, in the leg score subanalyses (front/hind, right/left) **neither the KIT nor the candidate gene could be found for WM on front legs** but additional regions on ECA3, ECA17, and ECA19 were identified that could be exclusively responsible for the front leg WM.
- A region on ECA13 seems to be related to the WM of the front, left, and right, but not for hindlegs.
- The evaluation of the summed scores as a binary trait does not seem to have any advantages, but rather disadvantages. Both the number of chromosomal regions found and the intensity of the p-values decreased.
- The definition of the trait plays an important role in the search for relevant genomic regions
- The evaluation of the individual legs (quantitative/binary) is still planned.



Efino *2017, EBV: 150



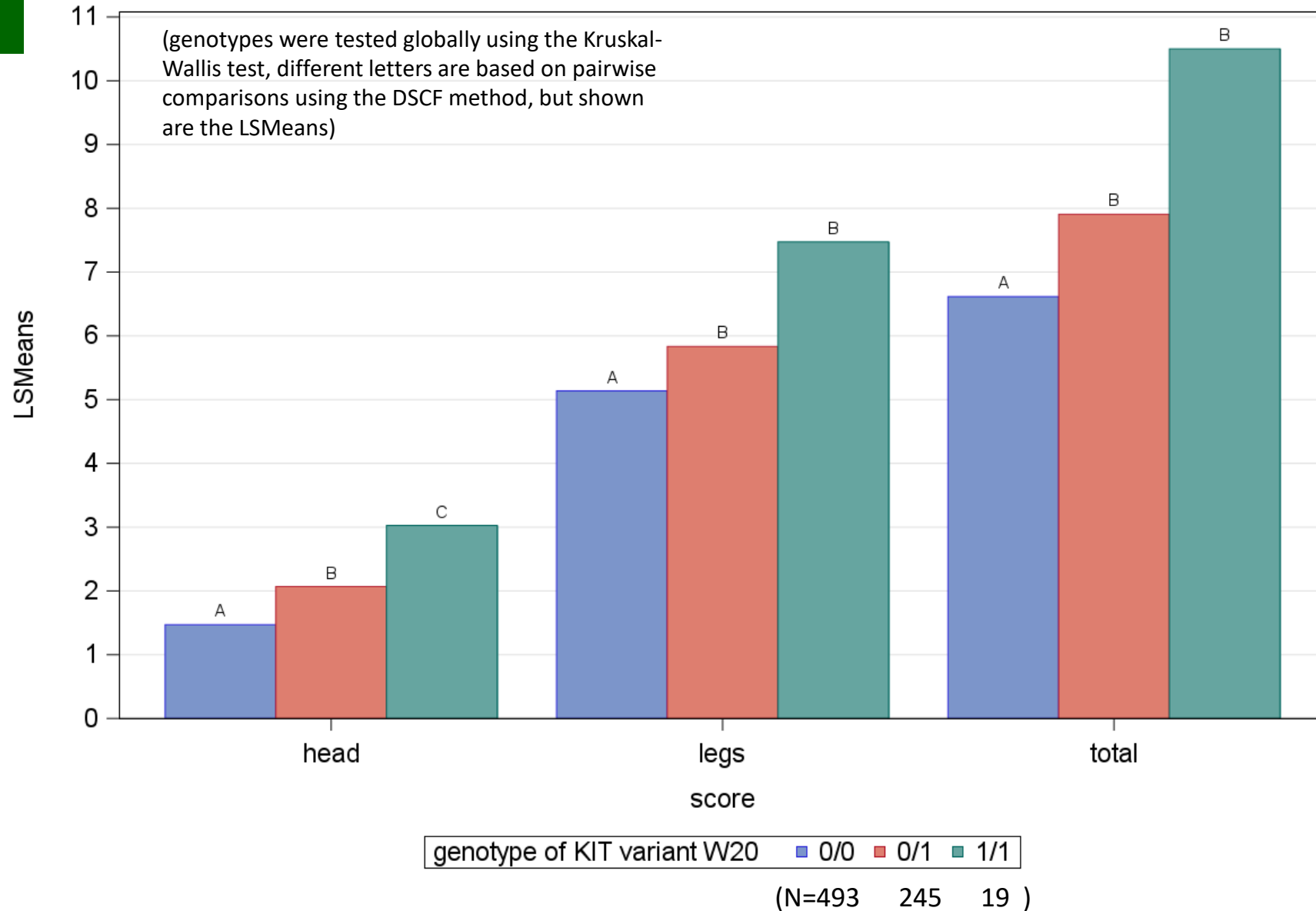
Friesius *2017, EBV: 63

Missed the poster session #221278 ? ->

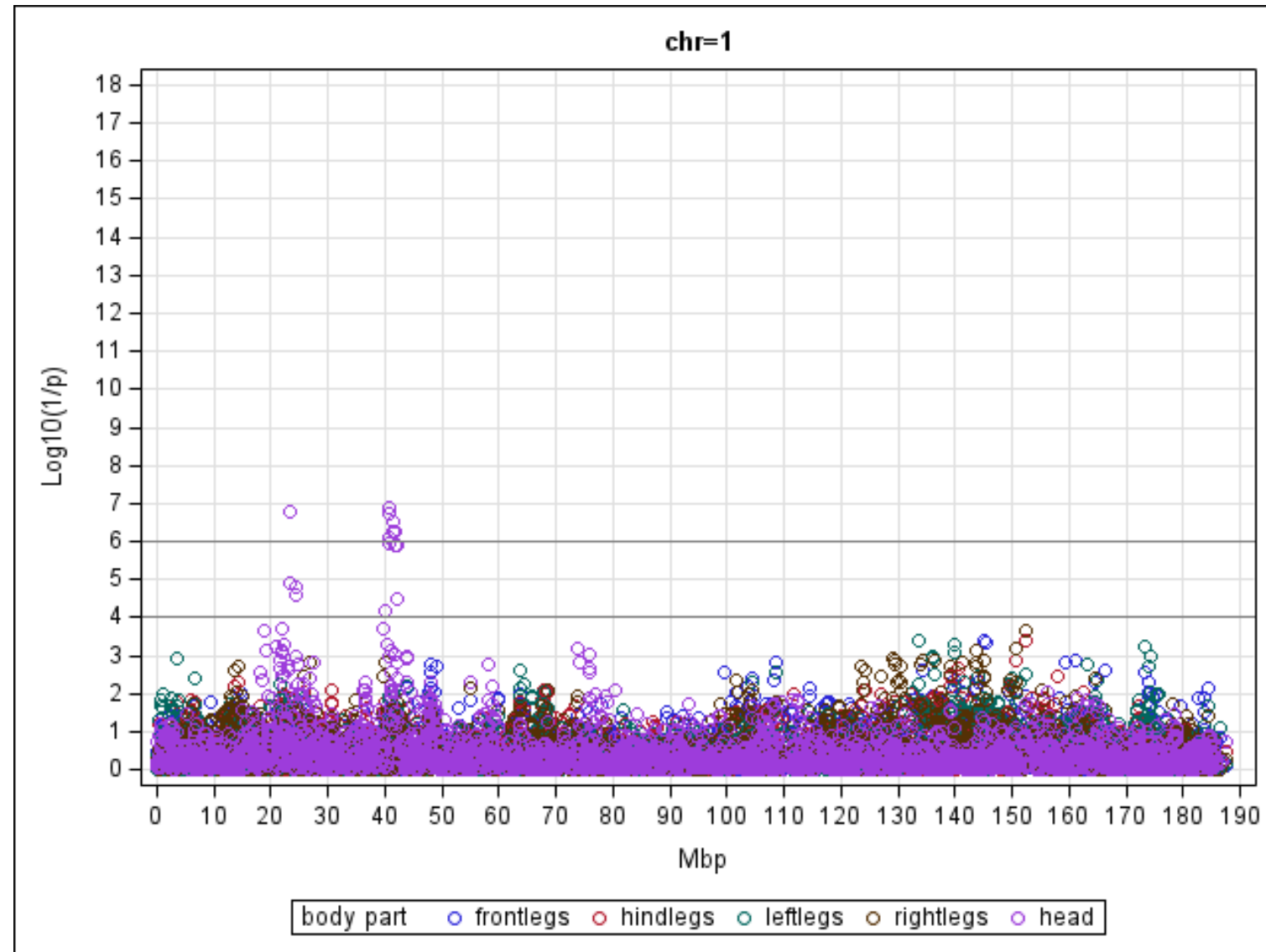


Thank you for your attention!

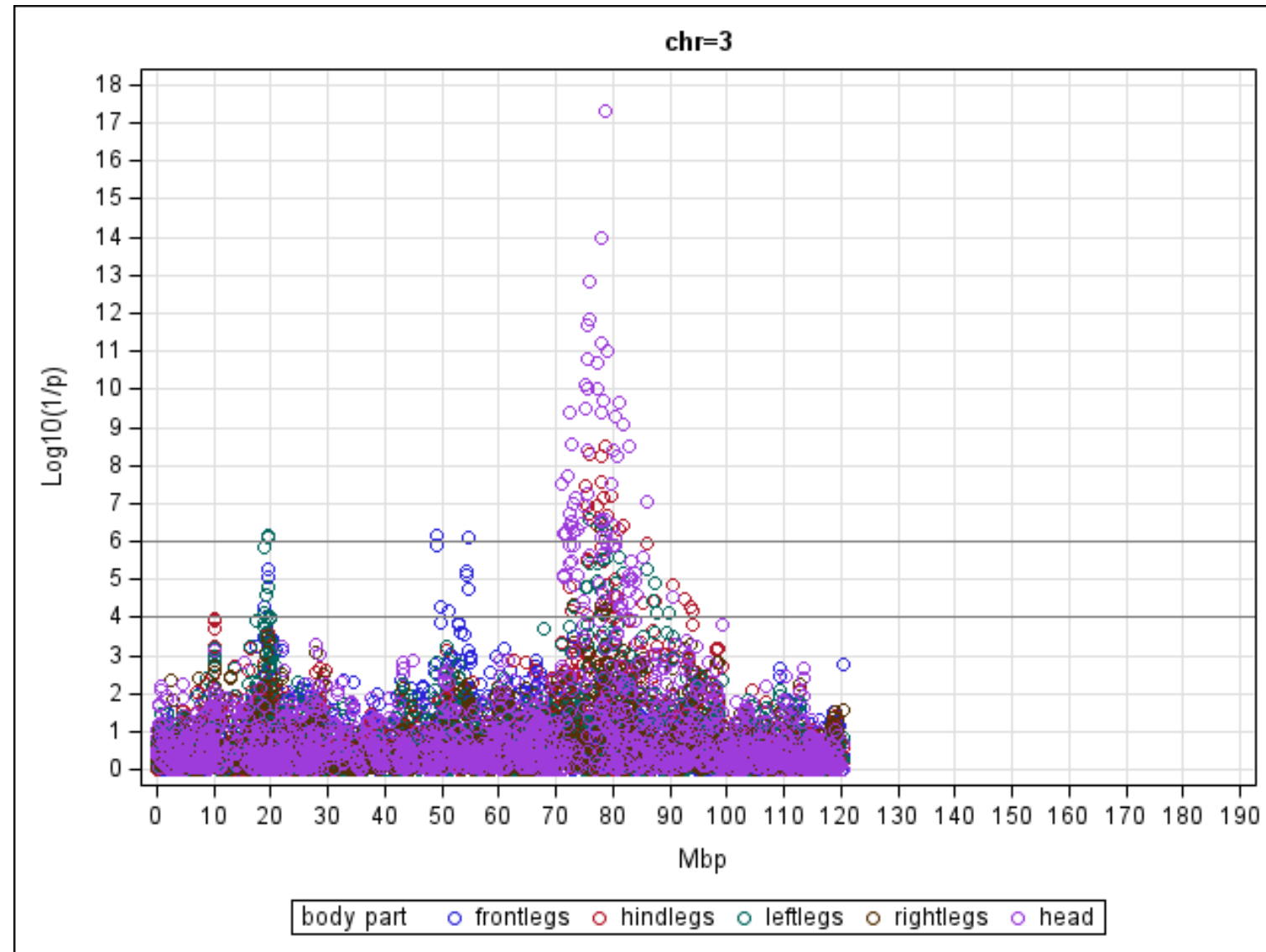
Relationship between KIT variant W20 genotypes and white markings



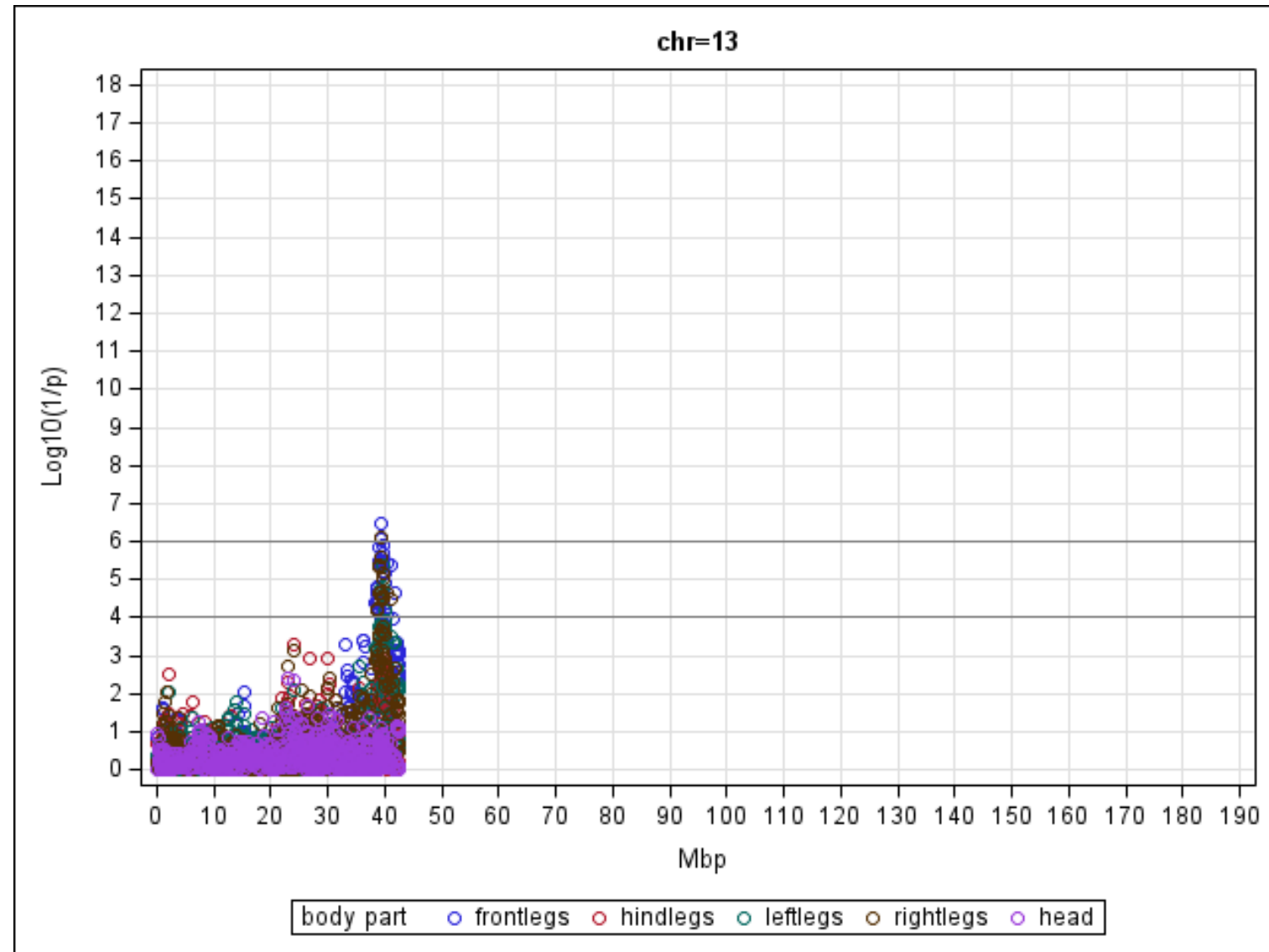
Chr 1 (reference genome EquCab 2.0)



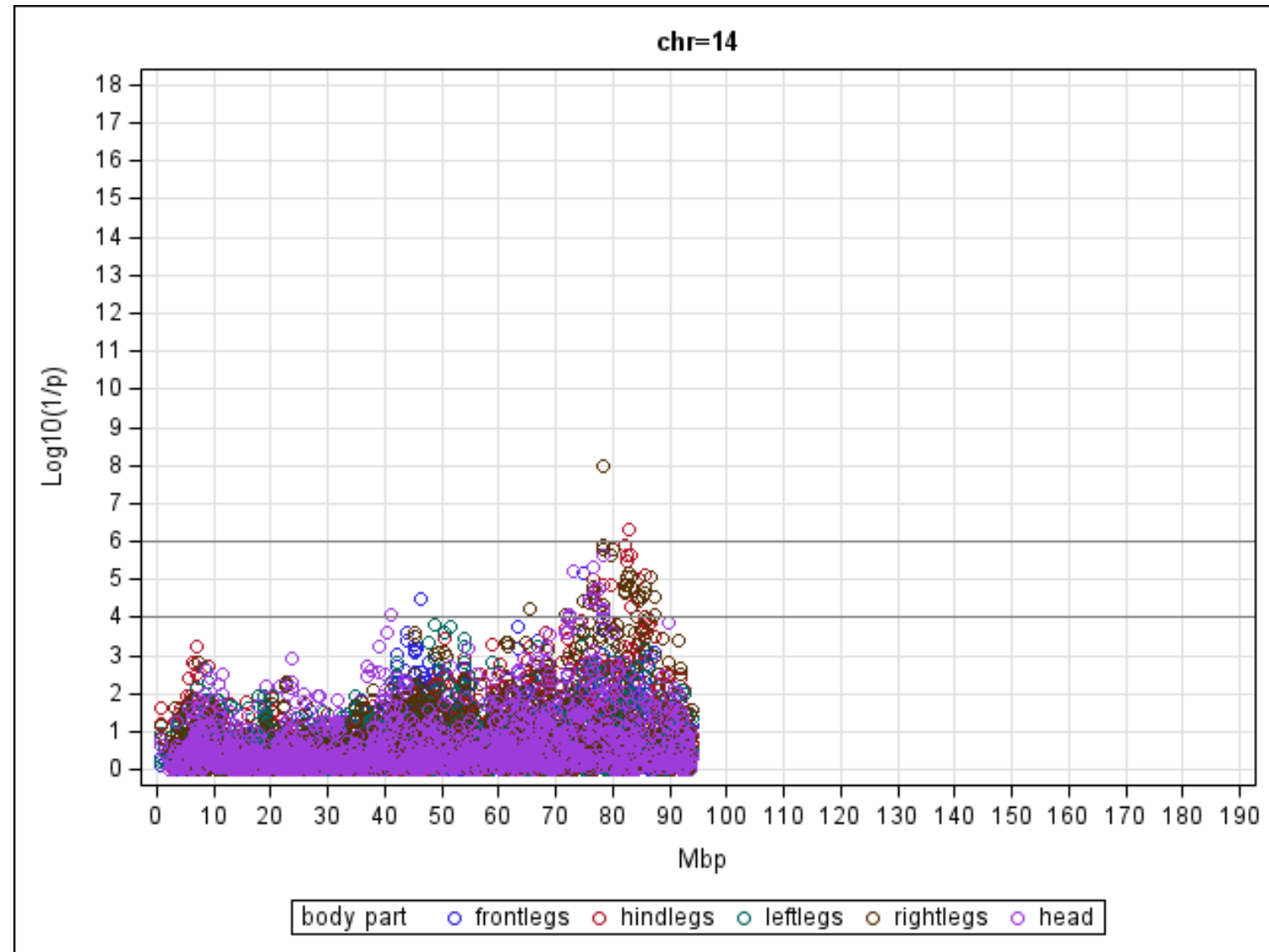
Chr 3 (reference genome EquCab 2.0)



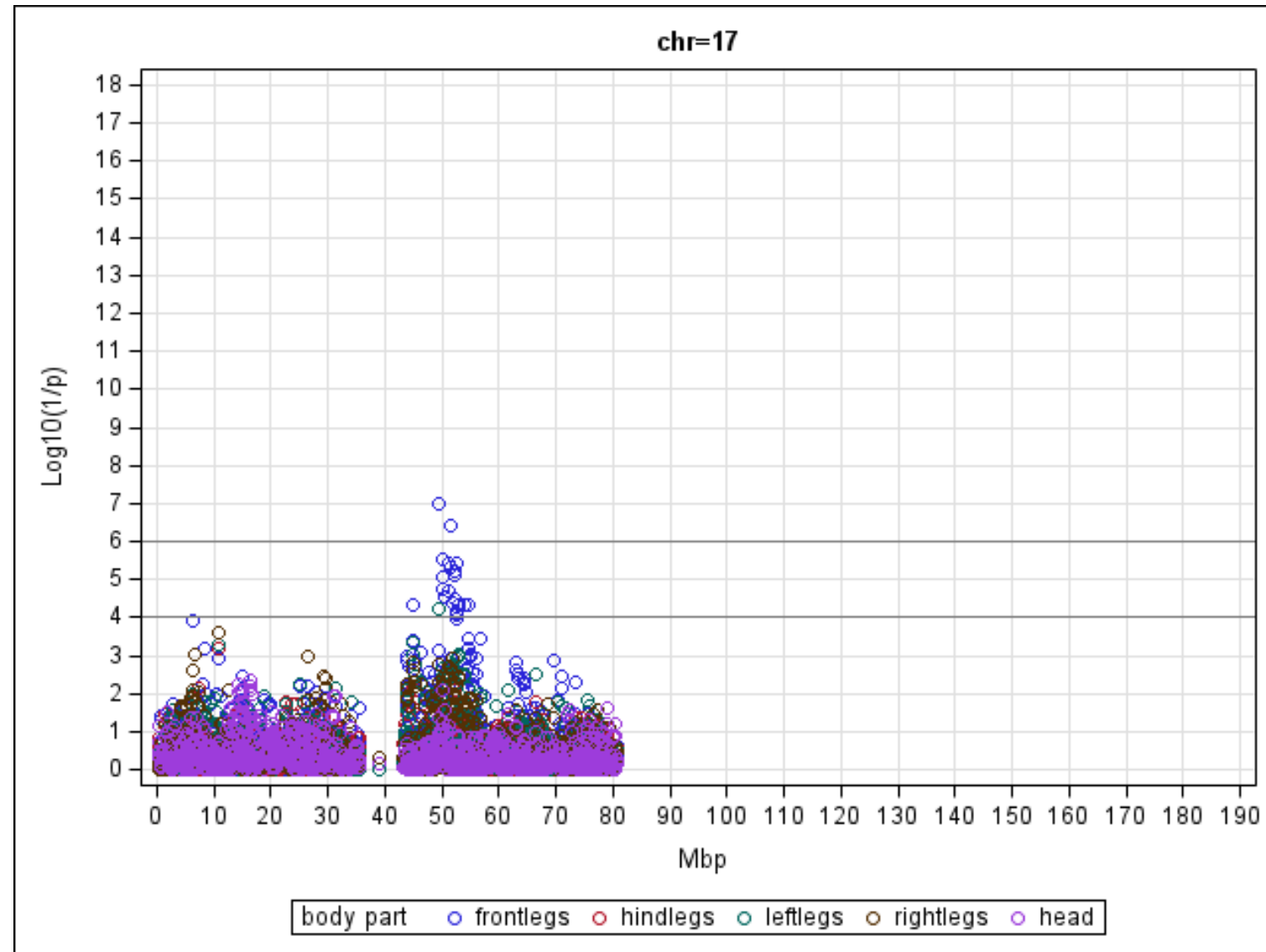
Chr 13 (reference genome EquCab 2.0)



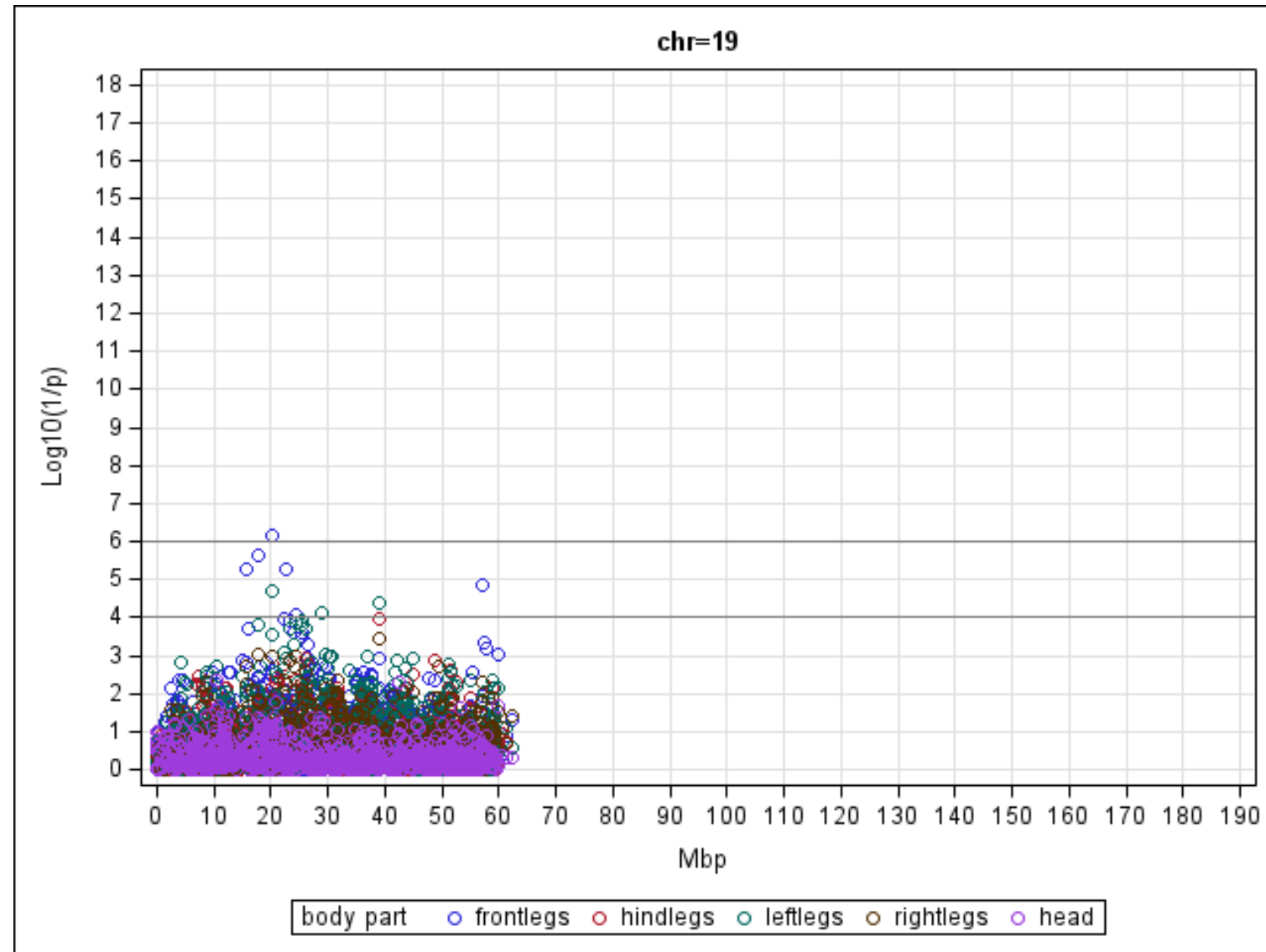
Chr 14 (reference genome EquCab 2.0)



Chr 17 (reference genome EquCab 2.0)



Chr 19 (reference genome EquCab 2.0)





#white fl #white hl	0	1	2	sum
0	959 (92%)	64(6%)	18(2%)	1041 (11%)
1	1646 (77%)	359 (17%)	118 (6%)	2123 (22%)
2	3573 (56%)	1794 (28%)	988 (16%)	6355 (67%)
sum	6178 (65%)	2217 (23%)	1124 (12%)	9519

~2/3 have 2 white hind legs and 11 % have 0 white hind legs

12% have 2 white front legs and ~2/3 have 0 white front legs

~50% have 2 white hind legs & 0 white front legs