

Evaluation of genetic variants proposed as myopathy risk factors in relation to sport performance in riding horses

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Background & motivation

- wide variation of equine myopathies: frequency (overall, within breeds or breed groups), type, age at onset and contexts of clinical manifestation
→ discussions about **muscle integrity myopathy (MIM) = "PSSM2"**
- six genetic variants proposed as risk factors for MIM
- relation to sport performance in riding horses?
- coverage of important breeding goal traits by the spectrum of estimated breeding values (EBV) for sport performance

➤ increasing knowledge about MIM in riding horses

Data

- Oldenburg studbooks OL (mainly dressage) and OS (mainly jumping):
> 11.000 broodmares with EBV from the routine national genetic evaluation for riding horses (FN-ZWS 2023)
- genotype data (MIM test result) for 557 Warmblood riding horses
 - owner-motivated testing
 - including all six MIM variants
→ P2, P3, P4, Px, P8, K1
- subset of Oldenburg broodmares with EBV and MIM test results: 219 mares
 - age 3 to 24 years
 - OL (N=159) > OS (N=60)
 - EBV for traits relating to performance in dressage (N=8) and show jumping (N=6)

Data distribution

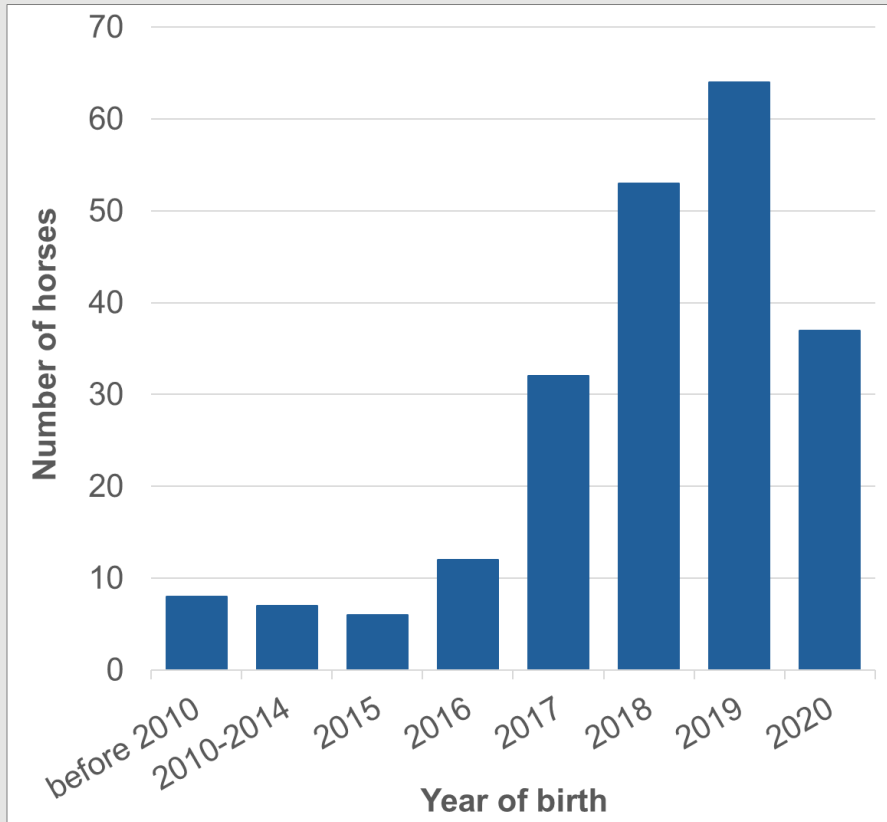


Fig. 1: Distribution of the 219 mares with MIM test result and EBV by year of birth

EBV	Active broodmares with EBV and MIM test result (N=219)				
	N	mean	std	min	max
EBV-D young horses	219	116,12	20,53	67	161
EBV-D performance test	219	115,16	19,84	67	158
EBV-D walk (performance test)	219	112,13	16,22	73	143
EBV-D trot (performance test)	219	114,93	20,29	65	157
EBV-D canter (performance test)	219	111,75	16,05	71	152
EBV-D rideability (performance test)	219	111,89	16,14	73	149
EBV-D young horse competition	219	114,42	18,13	74	153
EBV-D sport competition	132	120,94	16,96	76	158
EBV-J young horses	212	91,21	19,65	57	139
EBV-J performance test	212	89,01	22,13	57	140
EBV-J free jumping (performance test)	213	86,78	24,32	56	143
EBV-J jumping under saddle (performance test)	194	93,87	16,25	67	131
EBV-J young horse competition	196	96,39	13,09	70	130
EBV-J sport competition	86	104,48	17,58	59	134

Tab. 1: Distribution of performance related EBV (D dressage, J show jumping) from FN-ZWS 2023 in the 219 mares with MIM test result

Data distribution II

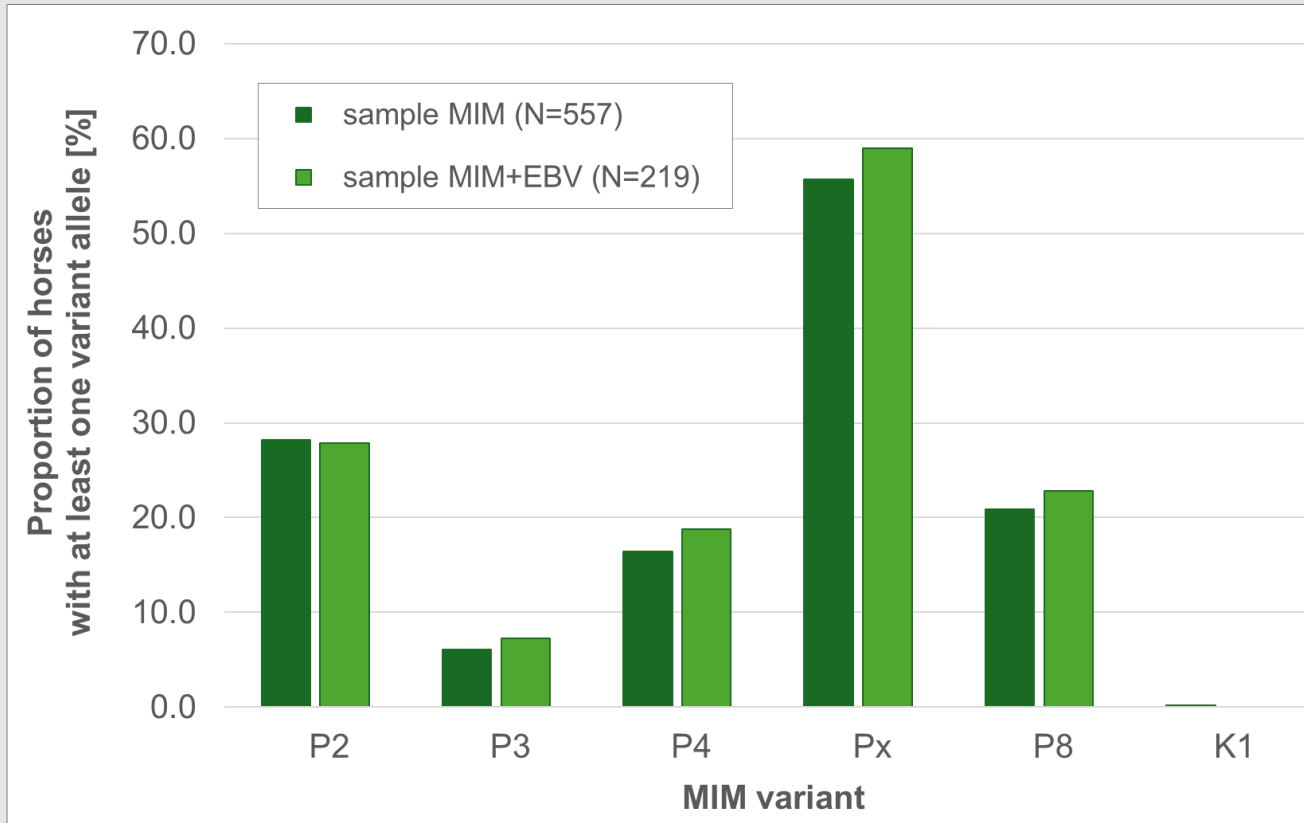


Fig. 2: Prevalence of MIM variants among all 557 horses with MIM test results and among the 219 mares with MIM test result and EBV

- similar proportions of horses with one or two MIM variant alleles in the whole sample (N=557) and the subset of mares with EBV
- MIM variants occurring in > 10% of horses considered for further analyses: 4 of 6 (P2, P4, Px, P8)

Analyses

- possible relationship between MIM variant status and genetic performance potential
- general linear models (SAS software, version 9.4) with fixed effects:
 - year of birth (<2017, 2017-2018, 2019-2023)
 - discipline focus in breeding (studbook; OL, OS)
 - MIM variant status → separate for P2, P4, Px, P8
 - at least one variant allele (Y/N)
 - number of variant alleles (0, 1, 2)

$$y_{ijkl} = \mu + \text{Studbook}_i + \text{BYear}_j + \text{MIMstatus}_k + e_{ijkl}$$

- multiple analyses of variance performed variant by variant for 14 performance traits (EBV)

Results: MIM variants vs. performance related EBV

EBV	MIM variant P2		MIM variant P4		MIM variant Px		MIM variant P8	
	N	P value	N	P value	N	P value	N	P value
EBV-D young horses	219	0.080 +	218	0.126 n.s.	217	0.916 n.s.	219	0.236 n.s.
EBV-D performance test	219	0.086 +	218	0.223 n.s.	217	0.826 n.s.	219	0.327 n.s.
EBV-D walk (performance test)	219	0.426 n.s.	218	0.791 n.s.	217	0.618 n.s.	219	0.041 *
EBV-D trot (performance test)	219	0.080 +	218	0.155 n.s.	217	0.918 n.s.	219	0.385 n.s.
EBV-D canter (performance test)	219	0.065 +	218	0.095 +	217	0.562 n.s.	219	0.784 n.s.
EBV-D rideability (performance test)	219	0.068 +	218	0.270 n.s.	217	0.865 n.s.	219	0.392 n.s.
EBV-D young horse competition	219	0.072 +	218	0.056 +	217	0.558 n.s.	219	0.124 n.s.
EBV-D sport competition	132	0.155 n.s.	131	0.798 n.s.	130	0.683 n.s.	132	0.687 n.s.
EBV-J young horses	212	0.701 n.s.	211	0.188 n.s.	210	0.475 n.s.	212	0.605 n.s.
EBV-J performance test	212	0.987 n.s.	211	0.459 n.s.	210	0.464 n.s.	212	0.452 n.s.
EBV-J free jumping (performance test)	213	0.655 n.s.	212	0.814 n.s.	211	0.603 n.s.	213	0.378 n.s.
EBV-J jumping under saddle (performance test)	194	0.918 n.s.	193	0.234 n.s.	192	0.639 n.s.	194	0.667 n.s.
EBV-J young horse competition	196	0.610 n.s.	195	0.029 *	194	0.706 n.s.	196	0.802 n.s.
EBV-J sport competition	86	0.572 n.s.	86	0.337 n.s.	85	0.492 n.s.	86	0.489 n.s.

Tab. 2: Results of multiple analyses of variance for performance related EBV (D dressage, J show jumping) with regard to the MIM variant status (binary) and referring to 219 mares with MIM test result and EBV – P values, with P<0.1 highlighted

Results: estimates for MIM variant status

EBV	MIM variant status (binary)	MIM variant P2	MIM variant P4	MIM variant Px	MIM variant P8
		LSM (95% CL)	LSM (95% CL)	LSM (95% CL)	LSM (95% CL)
EBV-D young horses	0	106.3 (103.8-108.8)	(n.s.)	(n.s.)	(n.s.)
	1	110.0 (106.2-113.8)			
EBV-D performance test	0	105.9 (103.5-108.4)	(n.s.)	(n.s.)	(n.s.)
	1	109.5 (105.7-113.2)			
EBV-D walk (performance test)	0	(n.s.)	(n.s.)	(n.s.)	104.8 (102.9-106.6)
	1				108.3 (105.1-111.5)
EBV-D trot (performance test)	0	105.3 (103.0-107.6)	(n.s.)	(n.s.)	(n.s.)
	1	108.7 (105.2-112.5)			
EBV-D canter (performance test)	0	105.1 (102.7-107.5)	105.6 (103.4-107.8)	(n.s.)	(n.s.)
	1	108.9 (105.2-112.5)	109.5 (104.9-114.1)		
EBV-D rideability (performance test)	0	104.3 (102.2-106.4)	(n.s.)	(n.s.)	(n.s.)
	1	107.5 (104.3-110.7)			
EBV-D young horse competition	0	105.6 (103.4-107.8)	105.9 (103.9-108.0)	(n.s.)	(n.s.)
	1	108.9 (105.6-112.2)	110.0 (105.2-114.1)		
EBV-J young horse competition	0	(n.s.)	99.6 (98.3-100.9)	(n.s.)	(n.s.)
	1		102.8 (100.0-105.7)		

Tab. 3: Results of multiple analyses of variance for performance related EBV (D dressage, J show jumping) with regard to the MIM variant status (binary) and referring to 219 mares with MIM test result and EBV – Least Square Means (LSM) for P<0.1

Summary and conclusions |

- significant discipline differences (P2, P4, P8, Px; all EBV / indices)
- no indications of associations of MIM variants with unfavorable performance potential
- some indications of favorable performance potential regarding young horse classes in carriers of at least one variant allele (P2, P4, P8; up to 6 EBV / indices)
 - P2: EBV-D young horses, EBV-D performance test (index; trot, canter, rideability), EBV-D young horse competition
 - P4: EBV-D canter (performance test), EBV-D young horse competition, EBV-J young horse competition,
 - P8: EBV-D walk (performance test)
- advantages as possible explanation for high frequency of variant alleles in the population
- special role of Px: most frequent, apparently unrelated with genetic performance potential
- **further research needed to verify these findings**

Summary and conclusions II

- ...
- **further research needed to verify these findings**
 - update (August 2024): extended dataset
 - genotype data (MIM test result) for ~~557~~ 1,362 Warmblood riding horses
 - subset of Oldenburg broodmares with EBV and MIM test results: ~~219~~ 692 mares
 - MIM variants vs. performance-related EBV: consistent results (pattern confirmed)
 - statistical power ↑
 - significant associations ↑ (former tendencies / P2 and n.s. / P4)
 - favorable performance potential in dressage in carriers of at least one variant allele (P2: 5+1 of 8 EBV-D, P4: 7 of 8 EBV-D; LSM differences 2.8 - 4.6)



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Thank you!



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