Syringomyelia in Cavalier King Charles spaniels

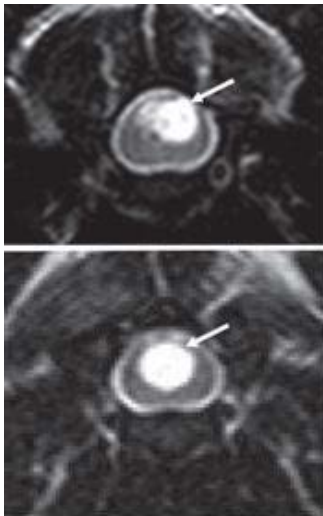## New phenotype for the genetic evaluation

K. Wijnrocx, W. Eggelmeijer, L.W.L. van Bruggen , N. Buys, S. Janssens, P.J.J. Mandigers
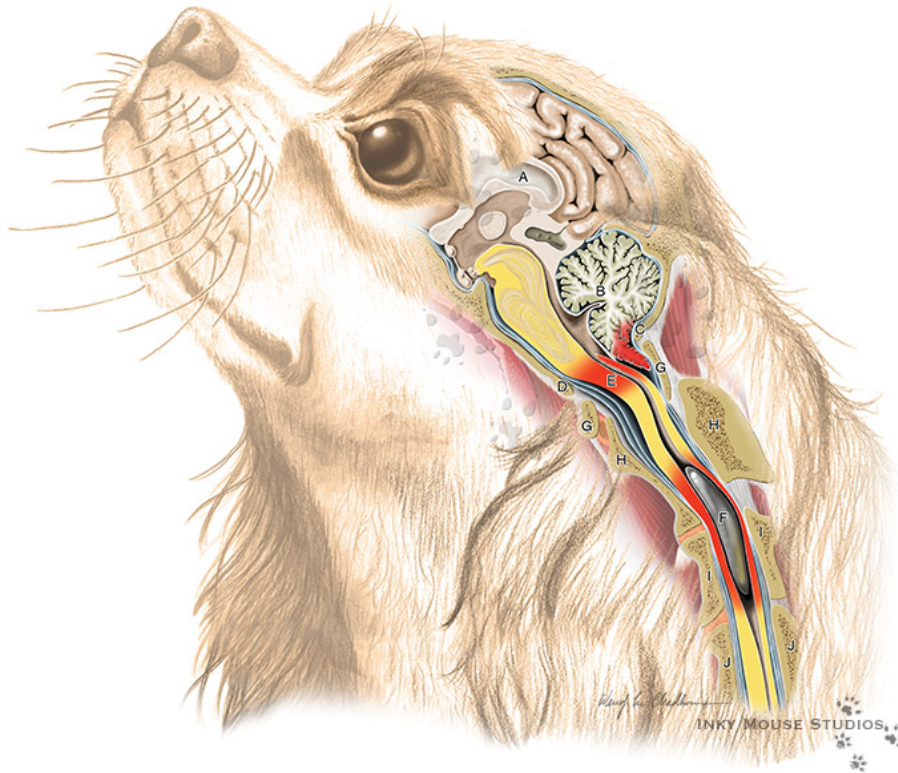
Syringomyelia

Fluid-filled cavities (syrinxes) formation within spinal cord

➔ Obstruction cerebrospinal fluid

➔ Neurological damage

➔ Clinical signs

➔ Frequent in brachycephalic toy breeds

Chiari-like malformation

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- Possible relation of SM with chiari-like malformation (CM)
  - Protrusion of cerebellum through the foramen magnum
  - Related to miniaturization and brachycephaly

How to asses SM?

- MRI scan of brain and spinal cord
  - o BVA/KC scheme based on threshold of 2 mm

SM GRADE	AGE (YEARS)	BREED TO
0a (normal)	over 5	any
0b (normal)	3-5	SM grade 0a, 0b, 0c, 1a
0c (normal)	1-3	SM grade 0a, 0b, 1a
1a (CCD)	over 5	any
1b (CCD)	3-5	SM grade 0a, 1a
1c (CCD)	1-3	SM grade 0a, 1a
2a (SM)	over 5	SM grade 0a, 1a
2b (SM)	3-5	SM grade 0a, 1a
2c (SM)	1-3	Do not breed

How to asses CM?

Chiari-like malformation (CM)

- **Grade 0**—No Chiari malformation

- **Grade 1**—Cerebellum indented (not rounded)

- **Grade 2**—Cerebellum impacted into, or herniated through the opening at the rear of the skull (the foramen magnum).

Prevalence of SM

- Prevalence high in small toy breeds
  - American Brussels Griffon population: 52%
  - UK Brussels Griffon population: 46%
  - Cavalier King Charles spaniel: up to 70% (different populations)

Heritability of SM

- One previous study found a moderate heritability
  - 384 CKCS in UK
  - Presence/absence of SM (binary trait)
  - $h^2 = 0.37$

Lewis et al., (2010). Vet. Journal, 183: 345-347

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Our study

- Total of 1194 Dutch and Belgian Cavaliers measured
- 223 dogs scanned multiple times
- Screening period 2004-2016

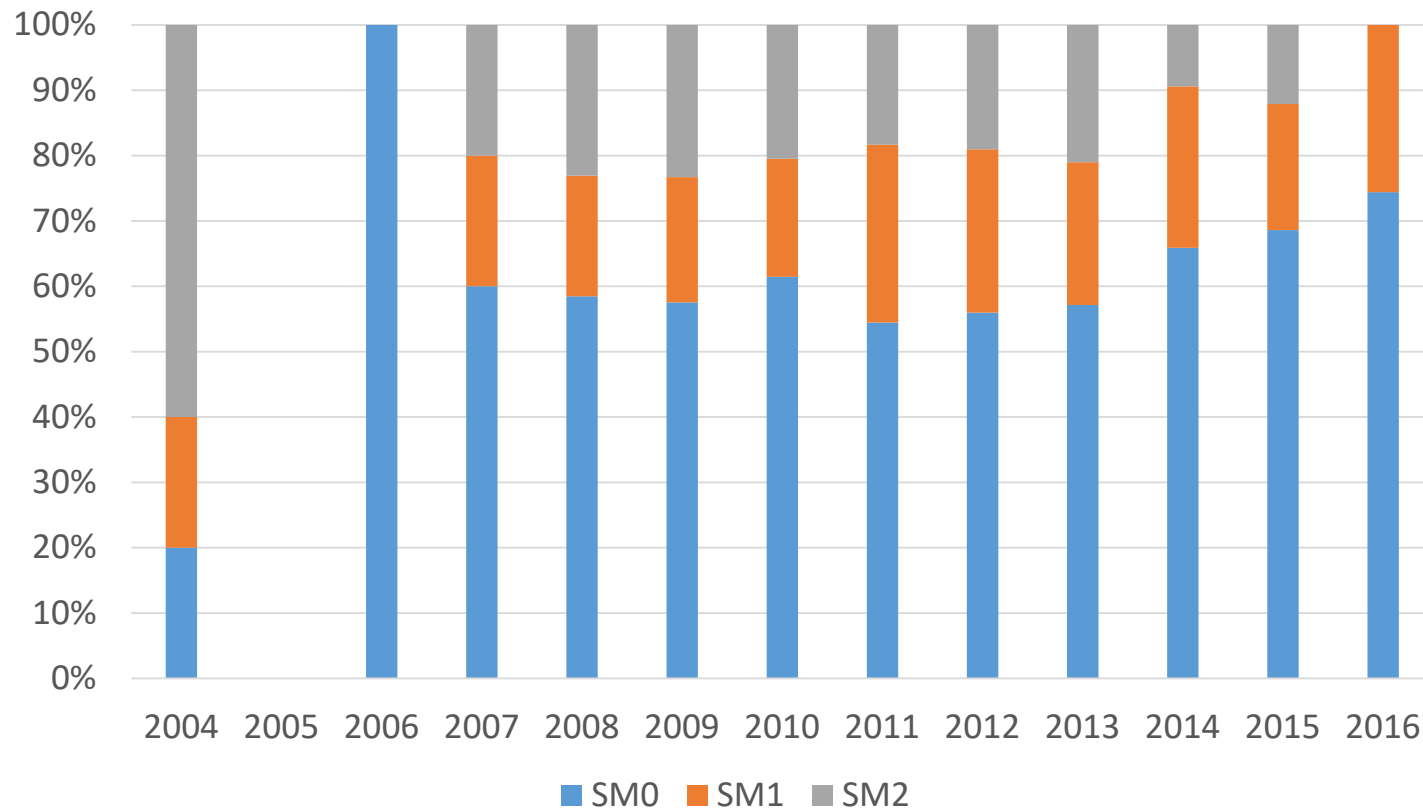➔ Re-evaluation of all MRI-scans → search for new phenotype

Our study

- Extra measures of SM compared to BVA/KC scheme:
  - o Measuring the exact syrinx width in mm
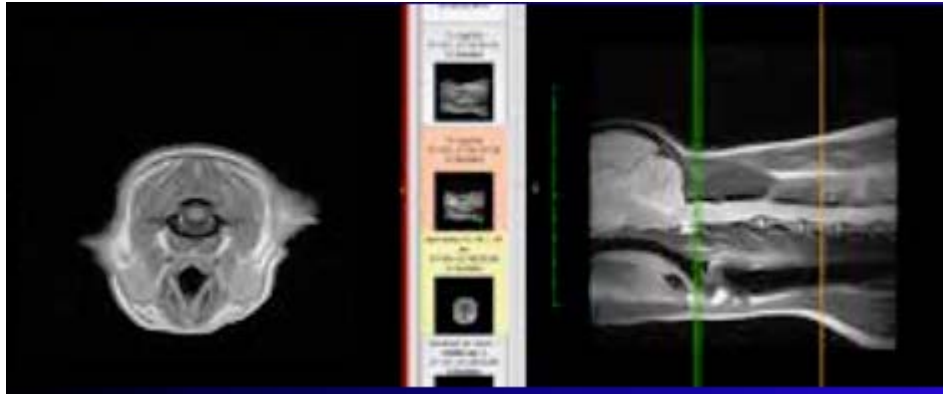
  - o Shape of the syrinx
  - o Age
  - o Animal clinic
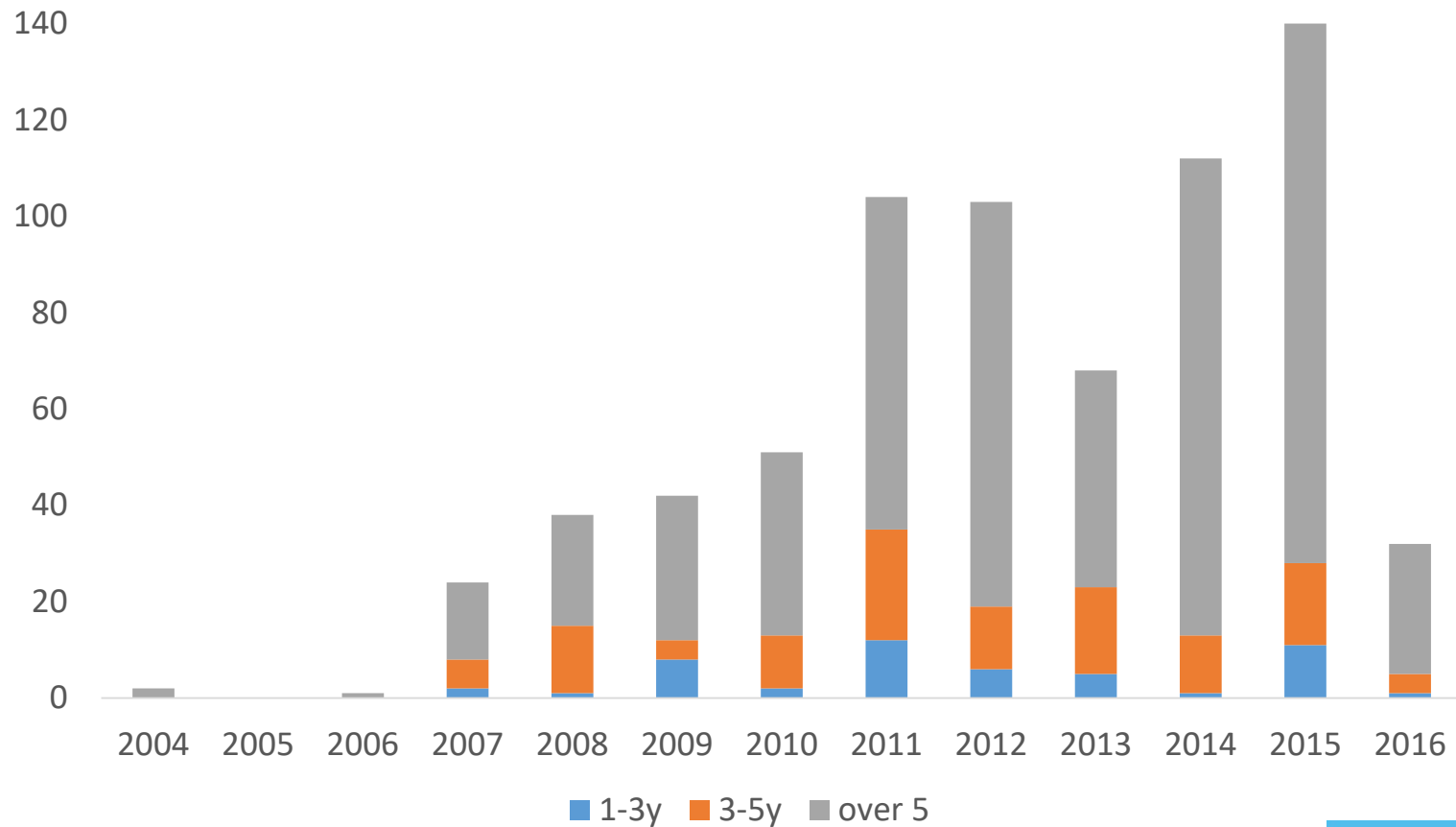- Extra measures of CM: shape of the cerebellum

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12 years of screening: results

- SM prevalence over years (based on BVA/KC scheme)

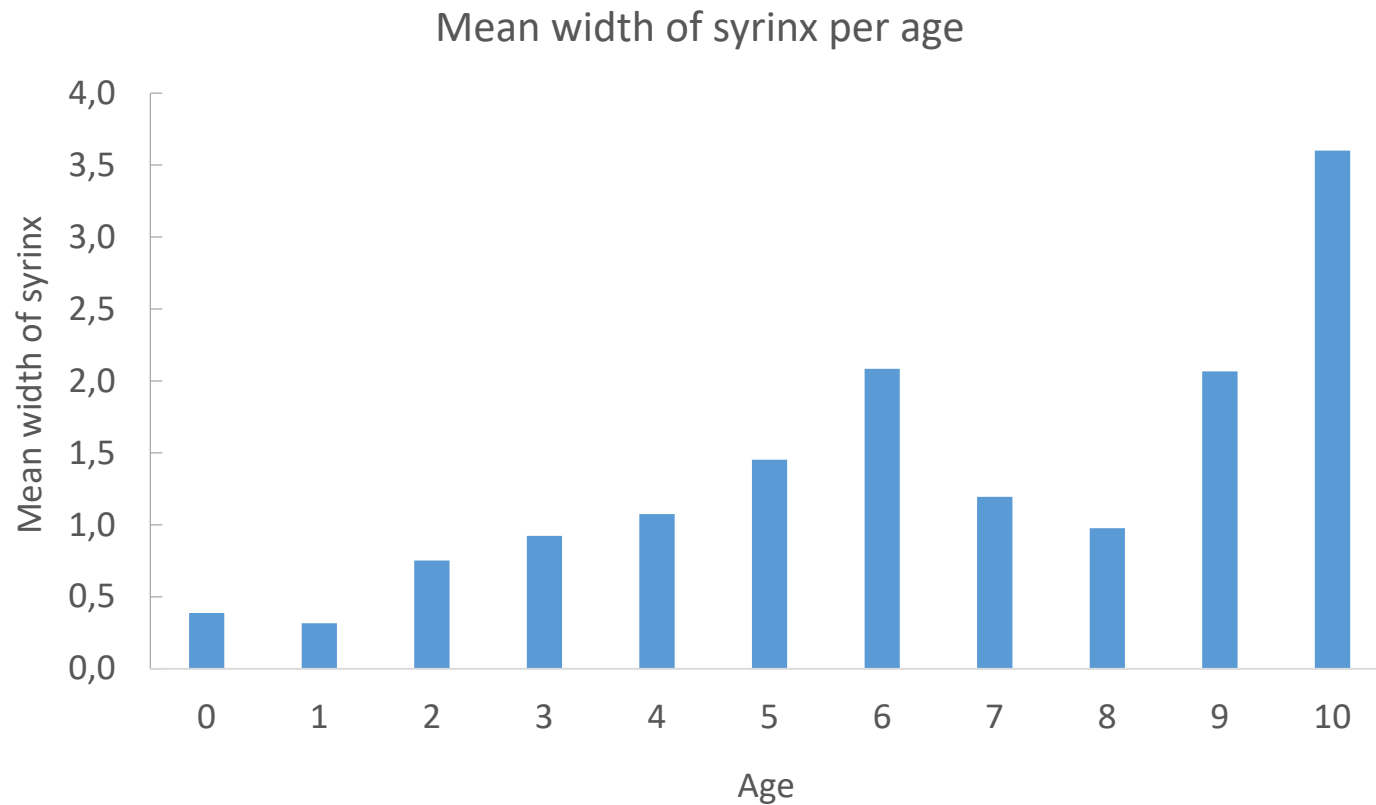# 12 years of screening: results

Eg. Score SM0 for different age classes

12 years of screening: results

- Age effect on SM

Mean width of syrinx per age

Heritability of syrinx width

- GLM models to assess non-genetic factors (using SAS)
- Non-genetic factors considered:
  - Age at scanning
  - Animal clinic * year of evaluation

- Estimates:

	BVA/KC scheme	Width of syrinx
h^2	0.13	0.30

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12 years of screening: results

- CM prevalence in entire period

  - CM0: 0 dogs
  - CM1: 15 dogs
  - CM2: 1179 dogs

  ➔ Characteristic of the breed!

Conclusions

- SM phenotype as a continuous trait looks promising

- Clear age effect of SM was shown: older dogs have higher scores

- High heritability of width of syrinx compared to low heritability of BVA/KC scores
  - More informative phenotyping

- First step towards EBVs as selection criterium

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Thank you!

Questions?

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