Syringomyelia in Cavalier King Charles spaniels

New phenotype for the genetic evaluation

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Syringomyelia

Fluid-filled cavities (syrinxes) formation within spinal cord

- Obstruction cerebrospinal fluid
- Neurological damage
- Clinical signs

- Frequent in brachycephalic toy breeds
Chiari-like malformation

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

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

<table>
<thead>
<tr>
<th>SM Grade</th>
<th>Age (Years)</th>
<th>Breed To</th>
</tr>
</thead>
<tbody>
<tr>
<td>0a (normal)</td>
<td>over 5</td>
<td>any</td>
</tr>
<tr>
<td>0b (normal)</td>
<td>3-5</td>
<td>SM grade 0a, 0b, 0c, 1a</td>
</tr>
<tr>
<td>0c (normal)</td>
<td>1-3</td>
<td>SM grade 0a, 0b, 1a</td>
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<tr>
<td>1a (CCD)</td>
<td>over 5</td>
<td>any</td>
</tr>
<tr>
<td>1b (CCD)</td>
<td>3-5</td>
<td>SM grade 0a, 1a</td>
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<tr>
<td>1c (CCD)</td>
<td>1-3</td>
<td>SM grade 0a, 1a</td>
</tr>
<tr>
<td>2a (SM)</td>
<td>over 5</td>
<td>SM grade 0a, 1a</td>
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<tr>
<td>2b (SM)</td>
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<td>SM grade 0a, 1a</td>
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<tr>
<td>2c (SM)</td>
<td>1-3</td>
<td>Do not breed</td>
</tr>
</tbody>
</table>
How to assess 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
  o 384 CKCS in UK
  o Presence/absence of SM (binary trait)
  o $h^2 = 0.37$

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
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
Heritability of syrinx width

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

• Estimates:

<table>
<thead>
<tr>
<th>BVA/KC scheme</th>
<th>Width of syrinx</th>
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<tbody>
<tr>
<td>$h^2$</td>
<td>0.13</td>
</tr>
<tr>
<td></td>
<td>0.30</td>
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</tbody>
</table>
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
  o More informative phenotyping

• First step towards EBVs as selection criterium
Thank you!

Questions?

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