King Edward Referrals News

Christmas is just around the corner and 2011 appears over almost before it really started—passed in a blur of patients, talks and conferences (for next years' talks and conferences see page 3).

Business management tutoring has paid off and the practice's turnover is up—so I'll be sticking around. And sticking to the current business format ie seeing only small animal medicine referrals. Obviously, your support had a huge part to play in the practice's growth: thank you for your referrals in these interesting economic times! I value them and the trust you and your clients place in me very much. Any feedback you wish to give (positive or negative) is always very welcome.

This month's newsletter is short—just one interesting case I hope you find as fascinating as I did. So without more wittering:

Happy Christmas!

King Edward Referrals opening times during the Festive Season

Dec 16-18: Closed
Dec 19-23: normal working hours ie 8 am to 5 pm
Dec 24–26: Closed
Dec 27-30: normal working house ie 8 am to 5 pm
Dec 31–Jan 2: Closed

Summary:
open on work days, closed on public holidays

Case study no 8: L-2 WHO???

History: Dr Sharon Bouwer from Kragga Kamma Veterinary Hospital referred Roxie, a 5 year old Staffie because of 2 episodes of bizarre behaviour. The first, in Feb this year, lasted for a day. The 2nd current episode had been going on for 5 days at the time she presented to me. She was pacing, panting, appeared disorientated, would not respond to commands and attacked inanimate objects and other dogs. In addition, she had waxing and waning episodes of subtle weakness and / or mild ataxia since January. She is an only dog, is fully vaccinated and has minimal contact with other animals. She eats only kibbled dog food. Clinical and neurological examination revealed no deficits apart from altered mentation.

Question 1: Where would you localize the problem to?

Question 2: How would you investigate Roxie's case further?

Index

Answer 1: There is only altered mentation. This dog either has an encephalopathy / cerebral disease

Answer 2: Sharon's haematology, serum biochemistry and a urine analysis had gone a long way to eliminate the common metabolic encephalopathies (hepatic encephalopathy, uraemic encephalopathy, hypocalcaemia, hypoglycaemia etc.)

The weirder encephalopathies are usually associated with storage diseases or inborn errors of metabolism. Most present at a young age, but some can present in mature dogs.

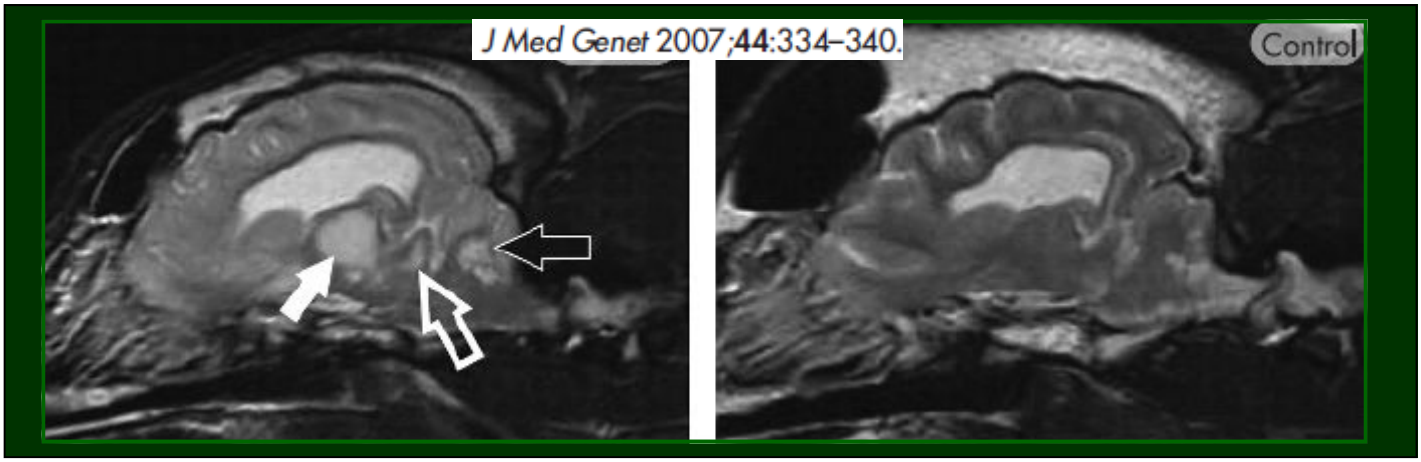
Once extra-cranial causes have been eliminated, intra-cranial disease is investigated with MRI and a CSF tap.

Thus my DD list was:

- Brain tumour
- tapeworms (very rare)
- toxoplasma / neospora (very slow progression)
- ceroid lipofuchsinosisn (one of those storage diseases)
- L-2 hydroxygutaric aciduria (L-2 HGA)

I remembered going to a lecture given by an old classmate from OP, Jacques Penderis (who is now head of the neurology unit at Glasgow vet school). He talked about this weird thing in Staffies—this L2-HGA. So I went and dug out his notes and papers. **And if you didn't know about it: don't worry. Neither did a couple of other specialists I spoke to!**

An MRI scan excluded a brain tumour and neurocysticercosis. There were no dilated ventricles / cerebral cortical atrophy that could point to ceroid lipofuchsinosis and no meningeal contrast uptake that would point to a meningitis. The MRI **was** consistent with L-2 HGA—though the lesions were a bit more subtle than the ones below from Jacques' paper: Basically you're looking at the bright areas (on T2 weighted sequences) - compare to control on the right.



A urine analysis run at UCT that specifically measured levels of organic acids in Roxi's sample confirmed that she had huge levels of L-HGA in her urine.

I found out later that Inqabatec (www.inqababiotec.co.za) already has a PCR test set up for this condition.

Now here's the kicker: according to various websites, **prevalence** of the gene causing L-2 hydroxyglutaric aciduria (L-2 HGA) **may be as high at 15-30% in the Staffie population**! The disease has also been reported in about 100 humans and in 1 West Highland White Terrier.

To learn about L-2 HGA, please turn the page

L-2 Hydroxyglutaric aciduria

People and animals affected by L-2 HGA have a mutation in the L-2 hydroxyglutrate dehydrogenase gene, resulting in a malfunctioning enzyme, accumulation of L-2 HGA (its substrate) and a deficiency of 2 ketoglutarate (its product). We're not sure what 2 ketoglutarate's function is, but we know that the enzyme is found on the mitochondrial membrane and that high levels are expressed in the brain, testes and muscle. In addition to being involved in energy metabolism in some way, the enzyme also appears to influence the function of L-glutamate, one of the neurotransmitter substances.

Presenting complaints in Staffies have included:

- sudden onset seizures
- Loss of obedience training, staring at walls, chewing on walls and stairs
- Ataxia
- head tremors
- Muscle stiffness and decreased exercise tolerance
- Untrainable with acute episodes of dementia ie bizarre behaviour

Take home:

Several owners of dogs described in the first paper thought their dogs were normal!

Onset of signs can be insidious

You won't find this problem unless you look

We should encourage breeders to test

Diagnostic options:

Genetic test (Inqabatec): R 450 (less if send multiple samples)

Urine screen for organic acids: R 1200

MRI scan: R 3500-4000

Dates to remember for 2012

February 2012: PECG meeting start up again—exact dates TBA
February 11-12: SC-SAVA minicongress, Oubaai, George
February 12-16: SA Equine Veterinary Association congress, Oubaai, George
May 4-5: EC-SAVA annual minicongress, venue TBA (East London direction)
May 6: IVPD practice management meeting, same venue
Early August: SAVA congress, venue: CSIR conference centre, Pretoria

Happy Christmas!