THE PBGV DAY AT THE Animal Health Trust, Newmarket, on 2 December 2016 commenced with a gathering of Petits in the aptly named Small Animal Centre, where one by one they were taken into examination rooms for eye testing and measuring of height. The latter was done against a laser beam on a wall. However, it was carried out in a rather confined space where several PBGVs just didn't want to know, turned round, sat down and by and large were less than co-operative. Fortunately some owners know the height of their PBGVs and, with suspicious measurements, demanded a recount! All in all, I am concerned about how this was done as data needs to be precise for any meaningful research. Most present were used to being shown and would have been far happier on a grooming table for accurate measuring to take place. With owners' help, hopefully arrangements might be made in the future for a further confidential measuring session at some event.

It is a great pity that several who had booked were unable to go at the last minute and, with most due to take more than one dog, this had an adverse effect, bringing numbers present down to around 27. Those that realised the importance of the day and turned up despite difficulties or distance involved included Karen Powney from Lancashire, Linda Lewis from South Wales and Jean Hood from Devon. Chairman Paul Osbourne also managed a flying visit. After eye testing and measuring, he rushed away to ill-timed, urgent work. For those who could stay the day was a relaxed affair with a delicious lunch put on by the AHT and plenty of time for dog walking in the magnificent grounds of Lanwades Park.

The first of the short talks in the small lecture theatre was on the Give a Dog a Genome project, given by Cathryn Mellersh, Head of Canine Genetics. Before she started, BGVC Breed Health Co-ordinator Vivien Phillips presented Cathryn with a plaque from the Club to thank her and her team for the dedication and work over the years culminating in the final discovery of the mutant gene responsible for POAG in PBGVs. Cathryn explained that the GDG project was designed to identify genetic variants underlying inherited disorders. They were looking for mutations by insertion, deletion, substitution or inversion. She said there is a lot of variation within the canine genome. Most of that variation is neutral, some positive or advantageous but some is deleterious or disease-causing. With nearly 2.5 million "letters" of DNA, sequencing obviously takes considerable resources and time. They will be sequencing 75 breeds, including Grands, and had asked to know the three main health concerns amongst those breeds. Ninety health concerns were listed, the top one being epilepsy in 18 of those breeds.

Research Assistant and PhD student, Chris Jenkins, followed by talking about epilepsy research at the AHT and their future research plans. He outlined the types of seizure and cited statistical data gained from the RVC's Vetcompass, which shares and analyses veterinary clinical information to understand disorders and improve the welfare of companion animals. He also referred to old PBGV research completed in Denmark by C H Gulløv, N Toft, M M N Baadsager and M Berendt. Many will already know of this paper, which is on the PBGVCA website www.pbgv.org/images/PDF/Health/DanishEpilepsyStudyGullov9-2011.pdf. Research took place against a background that epilepsy was on the increase in the breed compared to the general dog population. The target population consisted of all 876 PBGV dogs registered at the Danish KC from 1 January 1999 to 31 December 2008. A mailed questionnaire was used to detect possible signs of epilepsy. The information was subsequently validated by telephone interviews of positive and possible positive responders and a negative responder control group, using an extensive questionnaire developed to detect epilepsy. Dogs evaluated as epilepsy positive after the telephone interview were offered a clinical investigation. Results showed the prevalence of epilepsy was estimated to be 8.9% (42/471) in the PBGV population. Average age of onset was 26.3 months. Sex and mode of response did not affect the prevalence, but a strong litter effect was seen. Among euthanised dogs, epilepsy was the predominant cause (6/45 = 13.3%). The conclusion was that PBGVs experience an increased risk of epilepsy characterised by a relatively early onset and dominated by focal (previously called petit mal) seizures with and without secondary generalisation. With an estimated prevalence of 8.9% and substantial clustering within litters, a genetic factor associated with epilepsy was suspected. Research is long-term, possibly 5yrs, before the possibility of developing a DNA test. Statistically the level of seizuring within the breed is 2.2 to 2.5%. Peter Marks pointed out that the AHT now holds many samples submitted from the BGVC and hoped the breed would soon be given some priority in identifying those with a variant within a single gene which causes epilepsy.

The final talk of the day was given by Cathryn Mellersh, who gave an update on POAG research. Of the PBGVs worldwide whose DNA has been tested at the AHT 41.9% are carriers. Disturbingly the frequency of POAG mutation in PBGVs and PLL in Miniature Bull Terriers, is among the highest the AHT has ever observed. As Cathryn Mellesh has written "Knowing which dogs carry the mutation and which don't (the so-called 'clear' dogs) enables breeders to make sensible choices about the dogs they mate together. All dogs can be safely bred with provided at least one of the mating pair is clear of the mutation. Breeding dogs that will never develop the condition should obviously be the priority for all conscientious breeders and the desire to eliminate a disease-associated mutation from a breed should therefore be the long-term goal. But the instinct to choose only clear dogs to breed from, as soon as a DNA test becomes available, may not always be a sensible choice. If carriers are prevented from breeding the opportunity to pass the rest of their genetic material to the next generation is also lost and the genetic diversity of the remaining population is thus reduced".

"It is also worth remembering that the disease mutation for which there is a DNA test is not the only mutation a carrier has. Every human, on average, carries about 50 recessive mutations and there is no reason to believe the average dog won't carry a similar number. So the only real difference between a clear and a carrier is the single mutation that can be tested for. Both dogs will both carry around 49 other mutations that the breeder doesn't know about and can't test for. If carriers are not bred from and clear dogs are used extensively then there is a real risk that other mutations will increase in frequency in the breed and new inherited disease(s) could emerge".

Measuring the breed was touched on, with a reference to Weill-Marchesani syndrome. In simple terms this is is a disorder of connective tissue. Connective tissue forms the body's supportive framework, providing structure and strength to the muscles, joints, organs, and skin. The major signs and symptoms of Weill-Marchesani syndrome include short stature, eye abnormalities, unusually short fingers and toes, and joint stiffness. Adult height for men with Weill-Marchesani syndrome ranges from 4ft 8ins to 5ft 6ins and for women with this condition 4ft 3ins to 5ft 2ins. Cathryn Mellersh's goal is to discover whether this syndrome in humans might be linked with the reason why PBGVs carry the mutant gene responsible for POAG if they carry the ADAMTS17 mutation making them slightly shorter than clear dogs. A few, including myself, stayed behind at the end of the talks to discuss private concerns informally with Cathryn and I know that, without exception, everyone who attended was very grateful for the time she spent with us and the enjoyable and informative day.

Linda Skerritt