8. PRA
Update August 2010

Historical news: Gene mutation causing PRA found!

-- 4th August: the scientific publication of Mrs. Regina Kropatsch (University Bochum, Germany) named 'Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene' is online available. Please read the full publication [here](#).

-- 11th August: the scientific publication of Dr. Acland (Cornell University, Ithaca, US) named 'An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9' is also [online](#) and confirms the Bochum findings.

It is exactly the same gene mutation which causes PRA in Glens. As expected and now proved there is only one kind of PRA in Glens worldwide.

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From 2006 up to now we reported periodically about this project. Due to the fact that we promised Prof. Epplen and his team to keep their findings confidential until the scientific publication has been released we could not inform you in detail about the last very successful proceedings of the Bochum project. Now that Bochum has published their results on August 4th we could add the last chapter of the success story below.

Final episode

First success
In the mid of April 2010 the Bochum-team found a gene mutation which showed a significant relationship with the eye disease gPRA. All Glens diagnosed as affected showed without any exception the same gene defect.
The defect itself was very complicated and not comparable to the gene defect at e.g. Schapendoes. They found out that a part of the gene was missing but at that time they didn't know where the missing part was beginning and where it was ending. So they were able to identify only affected dogs, not carriers.
From that moment on the research work was concentrated on this gene disorder. And with success.

Second success
In the beginning of May 2010 they were able to define the gene defect and they developed a gene test for it. This direct gene test made it possible to identify affected, carriers and normal Glens. This test is used to screen almost all 171 DNA-samples they had in their project. The results were very plausible and more important they didn't show any contradictions. Overall the result of this population was 28,8% homozygous for this defect (already diagnosed as PRA affected or will develop PRA sooner or later), 44,1% carrier and 27,1% normal. Fortunately this is not representative for the whole breed because they researched pedigrees with a large number of affected Glens.
After some consultation we did send in additional blood samples of some less related, affected Glens and their test results were like expected. With high probability it can be said that we are
talking about only one kind of PRA worldwide (incl. the Glens in the US because they are very related to the European Glens). So the second success was born.

On Friday the 13th of May they informed us officially about this great news and invited us to come to Bochum for further explanation and discussion. They asked us to keep the given information strictly confidential until Regina had finished the scientific proof of what they found. So on Tuesday the 18th of May, we visited Bochum and prof. Epplen informed us about their success. We discussed the test results and the age an eye specialist is able to diagnose PRA. Therefore we prepared some homework and we found that, based on our database information, the average age (worldwide) is 6 years with a standard deviation of 2 years, so this means that the majority of the Glens can be diagnosed for PRA between 4 and 8 years. In practice this will be a bit earlier because some Glens already showed signs of blindness before they were tested. The moment that they actually get blind as a result of PRA is very different, some very early, some many years later.

Time for a champagne toast to celebrate this joyous moment.

This visit was a great moment for us. Last 3 years Bochum did an excellent job, the marker was found, a test was developed! This was absolutely the most important event in Glen history. We wished to cry this success from the roofs…… but we promised to wait.

Still they had to provide scientific evidence that the found gene disorder will cause the eye disease PRA. The theory is that the found gene defect always leads to a lack of a certain protein production which subsequently results in PRA. They estimated to spend another few months to realize this final milestone.

On the 17th of June Optigen (US) announced to have a PRA test, named crd3, available for the Glens. Although there was no scientific publication one could conclude that Dr. Acland and his team had found the marker as well. What a coincidence. After having contacting Prof. Epplen we
decided to publish on the 18th of June the availability of a PRA-test in Bochum as well.

Meanwhile the Bochum research team went on finding evidence; they performed a detailed microscopic examination of an eye of a Glen and Prof. Epplen reported begin July having achieved major results and Mrs. Kropatsch was about finishing her publication.

**Third and final success**

On the 4th of August the scientific publication of Mrs. Regina Kropatsch named 'Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene' was online available. Details of this publication can be found [here](#).

One week later, 11th of August, the scientific publication of Dr. Acland (Cornell University, US) named 'An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9' came also online available. Details of this publication can be found [here](#).

This publication confirmed that the gene mutation, as published by Mrs. Kropatsch, is exactly the same. According to Prof. Epplen both publications can be seen as very complementary to each other because of the different way of research. As expected and now proved there is only one kind of PRA in Glens worldwide.

Who in the world could ever predict such an unique and successful ending of two fully independent research projects.

First of all we have to thank both research teams for achieving this fantastic result.

Special thanks go to Mrs. Regina Kropatsch, Dr. Gabriele Dekomien and Prof. Dr. Jörg Epplen of the Bochum University; it was a pleasure to cooperate with them and to support their project needs.

We highly appreciated the warm receptions as we were visiting them. Furthermore we have to thank the G.K.F. (Gesellschaft zur Förderung Kynologischer Forschung) for providing financial budget to run this project. Without their support this project could not have taken place.

Besides that we very much appreciated the contribution of all these 171 European Glen owners by sending in blood samples of their Glens, some did this up to 4 times! Of course thanks to the KFT Germany, EFG England, EFG Finland, the Dutch Association for the Irish Glen of Imaal Terriërs, some private companies and all individual people who contributed the project with a financial gift.

Last but not least we'd like to thank our partners from the International Bochum team, Mrs. Stefanie Blänkner and Mrs. Jean Rogers, for the excellent cooperation.

Applause to all of you!

Huub and Theo