# Genetic Testing for Spasticity – The Final Chapter?

By Jen Pinches

As members of a club dedicated to the Rex breeds, we know that they are unique. Devon Rex are also unique in developing a breed specific genetic disease, originally called Spasticity and now called Congenital Myasthenic Syndrome (CMS).

The condition, identified by muscle weakness, is recorded as first appearing in the UK in 1974. The immediate descendants of Kirlee, although by circumstance highly inbred, did not produce any affected kittens but pedigree research, tracing the incidence of affected and carrier cats, suggests that the gene was introduced from an early outcross or was an unfortunate mutation in a single cat.

The limited gene pool at that time meant that it wasn't long before carriers of the gene were mated together and affected kittens began to appear. The condition continued to spread and carrier cats were exported widely, gifting the defective gene along with them. By the early 1980's affected kittens had been born worldwide.

At the time, openness about health issues wasn't actively encouraged and in fact a great deal of secrecy and finger pointing surrounded the whole issue. Despite this a group worked together to research pedigrees, identify carriers, trace the source and to try and get interest from the scientific community in investigating the disease. Many breeders carried out test matings to try and ensure that their lines were clear of the gene and to a large degree that approach was successful in UK Devon Rex and over the past 30 or so years we have been fortunate, the condition appears to have been cleared from the UK genepool.

However, affected kittens have continued to appear in various parts of the world, in both Devons and Sphynx, where it was introduced from Devon outcrosses. So the gene is still out there and with the increasing numbers of imported cats being brought into the UK we risked re-introducing it to the UK genepool. Recent instances include a litter born in France only a couple of years ago, so it would be unwise to dismiss the risk as exaggerated. Recessive genes really are forever and the risk can't just be dismissed or ignored.

Helen Bryant, one of those who worked for years along with others in the UK and elsewhere, wrote this piece for the Christmas 1996 edition of the Rex Cat Club magazine I'm sure she didn't envisage that it would be another 19 years before the test she dreamed of finally became a reality! Pam Dowling, like Helen, another stalwart of the group set up to research the condition, wrote again in the summer 1998 edition of RexPress, asking for blood samples to be submitted for research. It took another 17 years for Marie Abitbol and a team at Alfort School of Veterinary Medicine in Maisons-Alfort, France to identify the gene and for a test to be made available.

So, what does the discovery of the gene and the test mean for today's breeders?

## Myopathy

### The end in sight

As the bearer of bad tidings for so many years on this subject and having suffered the subsequent flak as a result of it - you simply cannot imagine the sheer joy it gives me to announce that finally after all these years the solution is within our grasp.

For some time I have been aware that despite the valiant test-mating efforts of responsible, dedicated Devon breeders throughout the world, this was not going to be enough to eliminate the problem, but at least had bought us time till a more reliable method of identifying carriers became available, and this has now happened.

Ever since I discovered that a simple DNA blood test had been developed to identify carriers of recessive genes in humans, I have been searching for somebody somewhere in the world who was carrying out similar work in cats and in August - in America I found one who responded positively to my enquiry. This particular University Department is gene mapping the whole cat and specifically researching ten recessive conditions in cats and following a long telephone call have agreed to add myopathy to their list. This Department's brief is to locate the myopathy gene, after that the development of the blood test will be the responsibility of any country who wishes to take advantage of it. I have already established that we **do** have the facility to develop the blood test in UK.

Although at the moment I have no idea how long it will take to locate the myopathy gene and then develop the blood test or indeed what the cost is likely to be, feel sure that if mere money is all that stands between us and the elimination of this problem, we will find a way.

It is hoped more information will be available for our next AGM.

**Helen Bryant.**

Well, the GCCF is the first and so far the only registry of pedigree cats in the world to require that breeding cats are tested for CMS and the results logged with their registration.

Since the revised registration policy was approved at the Council meeting on 5th October, Devon Rex and Devon Rex Variants may only be registered in the Active Register, if they have themselves been genetically tested as normal for the CMS gene, or their parents have been tested normal, or cats on every pedigree line in previous generations have tested normal.

Cats which have not been tested, or are tested as carriers will be registered on the Non-Active Register or the Genetic Register. Any untested or carrier cats which are already registered on the Active Register will be moved to the Genetic Register.

DNA samples for testing to be submitted in support of registrations must be taken and certified by a vet and cats must be microchipped prior to testing and the number clearly shown on all documentation and the cat's own vet record.

Whilst we hope that carriers won't be found, it does mean that if any are identified, they can STILL be used in a breeding programme whilst on the Genetic Register and offspring tested clear can be registered on the Active Register. Breeders only need to test their breeding cats, if parents are clear there is no need to test kittens. The GCCF Devon Rex breeding population can be tested and proven clear within a generation.

If you do find that you have a carrier, being open and sharing that, particularly with anyone with cats of a similar pedigree can only be good for the breed. We really don't want to return to the days of whispering in corners, which was so prevalent in the 1980's and 1990's.

DNA testing, for those who haven't done it, is quick, easy and relatively inexpensive. There are a numbers of labs which offer the test but this is the process for Langford Vets at the University of Bristol. Their website is *http://www.langfordvets.co.uk/diagnostic-laboratories*, you can request swabs online and they will be posted to you with a reply paid envelope to return them.

***"Rex Cat Club members get a 20% discount by quoting a code which is available from the club secretary."***

You then need to arrange for your vet to take the swab and complete the Vet Checked Form, which has to be returned with the swab. If the cat isn't already microchipped, that will need to be done before taking the swab.

The swab is then posted off for testing and results will be emailed to you within a day or two and all you then need to do is forward the email with the results to the GCCF office, using *GCCFShorthairRegistrar@gccfcats.org* email address.

The abstract of the research paper on the CMS gene can be found here, for those interested in more of the scientific background to the condition.
*http://journals.plos.org/plosone/article?id=10.1371/journal.pone.0137019*

It has been a very long time coming but we can finally say with certainty that we can and will eradicate this disease from our breed. We should recognise and be grateful for the efforts of Helen Bryant, Pam Dowling, Sybil Drummond, Fran Tollan, Ginny Rastall, Mary Robinson and the many others who worked with them, along with the breeders who test mated, in the knowledge that they might be facing the heartbreak of producing affected kittens. We owe them all a huge debt of gratitude.