Co-Chair’s Report from AGM 2017

We would like to thank the whole committee for their efforts during the past year we are working very well as a team and all of us worked hard throughout the year to consolidate the foundations of this organisation. Special thanks to Jackie for her role as secretary, Wayne for treasurer, David for member support and Matt for South Island representative.

We sadly accepted Matt’s resignation as South Island Representative and former treasurer as his change in job has left him unable to attend our monthly meetings. We thank him for the pivotal role in managing our membership database and providing member support and organizing gatherings for those in the South Island. We would like to say our thanks with a gift card to Matt, David please take it back to Christchurch & pass it onto Matt.

At this time we would like to take time to acknowledge the work of one of the initial committee members, Tom Biggar who passed away earlier in the year. Tom with the support of his wife Bronwen who is attending the meeting today, helped with setting the high standards we have on the committee today.

As Co-chair Catherine continue to enjoy her role in corresponding with members, including calling new members, keeping in touch, providing support and advice where appropriate. She manages our website, Facebook page, email & Skype accounts and keeps web directories up to date. We keep a close working relationship with health professionals and others like pharmaceutical companies who have supported us.

Currently we’re working on an article for the Dental Association and require some non-identifiable photos of people with Acromegaly jaw & dental changes to add visual representation to this highly informative article. Wayne is working on an article for the Podiatry Association. Funds raised through our fundraising activities are used for the publication and dissemination of these awareness articles.
Overall 2016/17 has been consistently busy with the following events:
We held a combined Auckland/Waikato coffee catch up at the Remuera Golf Course in November 2016 attended by 17 people. Matt arranged a catch up at their brand new RSA in Christchurch in February 2017. Both of these events were relaxed and enjoyable and provided a chance for us all to enjoy each other's company.

Clark has been busy organising fundraising with our “Get Active! for Acromegaly” on or around International Acromegaly Awareness Day on 1st November saw our members involved in walking, cycling, reading, running, kayaking, walking around Rarotonga and some just being active by going on the “give a little page” and making a donation. This activity raised approx $3000 dollars for our education awareness campaigns. We will be holding the “Get Active! For Acromegaly” again on 1st November this year or a day that suits you to be involved.

Movie night on 11th March 2017 was kindly organised by Janine Blackmore, daughter of member Barbara Stark, and Paul Harvey. This was great night out with drinks, nibbles, roast dinner and a thought provoking and inspirational movie about two groups marginalised by the government and society based on a true story. This activity raised approx $1200 dollars for our education awareness campaigns.

We have gained grants for posting from NZ Post and approx $450 from Novartis for reviewing a patient information booklet.

The committee has been very busy keeping up with the day to day management of a charitable organisation. We need more people to be participate, as this is very important for succession planning. We are also looking for keen members to help with individual projects. So please consider your skills and donate your time to make things happen at the Society.

By Catherine Chan & Clark McPhillips (co-chairs)

**AGM Educational Seminars**

**Kevin Milne - Pituitary Journey**

We were privileged to have Kevin speak to our AGM this year. He was extremely entertaining giving us a rundown on his tumours effects, and while he doesn’t have Acromegaly his symptoms were very similar to ours. He was very amiable talking to many members one to one and I hope he enjoyed lunch. We had great feedback about how enjoyable his talk was. I’m sure we’d love to have him every year but that would be asking too much as Kevin donated his time for the day at no cost to us and with a very early start for him to catch a flight from Wellington.

Once again a huge thank you Kevin.
Genetics of Acromegaly, or Doc, what’s caused my tumour & are my kids at risk?

Dr Marianne Elston (Endocrinologist, Waikato Hospital. Senior lecturer University of Auckland)

Report written by Dr Catherine Chan (co-chair)

Almost 50% of the childhood-onset acromegaly have an identifiable genetic background. AIP (Aryl hydrocarbon receptor Interacting Protein) mutations are the most common familial cause of isolated acromegaly and gigantism.

We were very privileged to have Dr Marianne Elston with us, an endocrinologist at Waikato Hospital. I found over 40 research items under her name, including the survey on acromegaly many members participated in a few years back. Her research has seen her awarded a PhD from Australia’s prestigious Kolling Institute of Medical Research in Sydney for research into the molecular basis of pituitary tumours. She won the emerging young researcher at the Kudos awards in Hamilton in 2010 for her work concerning the identification of genes involved in the development of pituitary tumours.

We started off with a brief overview of pituitary tumours and acromegaly. Pituitary is commonly known as the master gland or as described by Sir Walter Langdon-Brown “leader of the endocrine orchestra” since it affects almost every organ in the human body. The anterior lobe makes up approximately 80% of the gland.

Pituitary tumours are very common, in autopsy studies 14.4% of people had a pituitary tumour, 22.5% in radiological studies (Ezzat et al. 2004). Clinically significant tumours occur in approximately 1 in 1000 people (Daly et al. 2006).

Pituitary tumours can cause problems by mass effects & hormonal dysfunction. As the pituitary tumour grows it can push onto the optic nerves and affect vision resulting in gradual reduction of visual fields. The tumour can also push the normal pituitary gland causing a loss of normal pituitary hormones. In acromegaly, the pituitary tumour makes too much growth hormone.

Hypopituitarism is a term to describe the loss of one or more pituitary hormones. Some pituitary
hormones are more important than others. For example ACTH controls cortisol which is essential for life. Growth hormone even though it is not essential for life, is important for muscle, bone & general well being in adults.

Other very rare non-pituitary cause of acromegaly include neuroendocrine tumours elsewhere in the body producing too much growth factor.

The cause of pituitary tumours remain largely unknown, with familial syndromes (result directly from gene defects inherited from a parent) accounting for less than 5% of those with acromegaly. Most pituitary tumours are sporadic (meaning random, spontaneously occurring) with no known inherited genetic basis. Over the past decade there have been many advances in the field of genetics.

Tumours may develop from two common mechanisms. Firstly from the loss of a tumour suppressing gene (genes that help prevent tumours forming), or secondly due to overactivity of a proto-oncogene (genes that help cause cancer).

We moved onto the most important question “Doc, could my kids get this?” There are around 6 genes that we know of involved in Familial pituitary tumours. We will go through these below.

MEN1 - mutations in the tumour suppressor gene MEN1 causes multiple endocrine neoplasia type 1 (MEN1). This is rare occurring in 1/10,000 to 1/100,000 people, or around 1.2% of those with acromegaly. Think about MEN1 if someone has a pituitary tumour and another feature such as high calcium. It is commonly known as the 3 “P”s disease:

- Parathyroid disease - almost all by approx age 50. Results in high calcium, which can presents with kidney stones, bone aches, fatigue, depression etc.
- Pituitary tumours in approx 30-40%
- Pancreas - neuroendocrine tumours approx 30-75%

MEN4 presents with similar features to MEN1 but is due to a different gene mutation - CDKN1B. It is extremely rare with only 9 patients with pituitary tumours & CDKN1B reported worldwide, of these 5 had acromegaly. If someone presents with MEN1 features but no MEN1 gene mutation is found, then consider testing for a CDKN1B mutation.

AIP mutations were first described in 2006 and stands for Aryl hydrocarbon receptor Interacting Protein. This is the most common familial cause of acromegaly and gigantism. People with AIP mutations often present at a young age e.g. <30yrs and with large tumours. AIP mutations are found in up to 33% of those presenting younger than 18 yrs old, 13% in those <30 yrs, and just 3%
when assessing patients of all ages with acromegaly. Luckily not everyone who carries this mutation develops pituitary disease.

The famous Irish giants have been found to have the AIP mutation. One of the most well known was Charles Byrne (1761-1783) and he was over 2.3m tall, and he became a celebrity in England. Unfortunately he died age just 22 probably due to complications from the then undiscovered acromegaly. Byrne was living in London contemporaneous with the pre-eminent surgeon and anatomist of the time John Hunter. Hunter had a reputation for collecting unusual specimens for his private museum, and as Byrne's health deteriorated he feared that Hunter wanted his body for dissection (a fate reserved at that time for executed criminals) and probable display. Byrne had made express arrangements with friends that when he died his body would be sealed in a lead coffin and buried at sea. But his burial wishes were thwarted and his worst fears realised when John Hunter arranged for Byrne's cadaver to be snatched on its way to sea. Hunter then reduced Byrne’s corpse to its skeleton and four years later put Byrne's skeleton on display in his Hunterian Museum, where it still resides and now located in the Royal College of Surgeons, London. (http://surgicat.rcseng.ac.uk/Details/collect/4123)

Carney Complex is an autosomal dominant condition most commonly caused by mutation in the PRKAR1A gene, which may be a tumour suppressing gene. People with this condition gets spotty skin pigmentation, heart issues and endocrine hyperactivity. They develop acromegaly due to pituitary hyperplasia and overactivity rather than pituitary tumours. There are about 500 cases reported worldwide.

SDHx (Succinate dehydrogenase) mutations are associated with phaeochromocytomas and paragangliomas, which are adrenaline & noradrenaline-secreting tumours. In very rare cases pituitary tumours may occur including acromegaly.
GPR101 gene duplications have been linked to X-linked acrogigantism (XLAG). A form of infant-onset gigantism, some cases were sporadic and some were inherited. There have been only 31 patients reported worldwide, causing very early onset gigantism age <5yrs, and usually starts at age 1.

McCune-Albright syndrome are caused by mutation in GNAS gene. Features include cafe-au-lait spots, bone and endocrine issues (including acromeglay, cushings, overactive thyroid).

Neurofibromatosis type 1 have been rarely associated with acromegaly, it is unclear if this is just coincidence or an increased risk.

Bearing in mind 95% of acromegaly occurs sporadically and are not inherited, almost 50% of the childhood-onset cases have an identifiable genetic background. Dr Elston went on to discuss the current guidelines at Waikato Hospital for genetic testing. She suggests patients who fulfill the following criteria be offered tested (criteria & funding varies depending on DHB):

- All patients with gigantism - underlying genetic cause can be identified in up to 50%
- Acromegaly if onset age <30 years old, especially if macroadenomas
- Family history of acromegaly - given the rarity of acromegaly this suggests possible inherited genetic cause
- If clinical features suggestive of possible syndrome e.g. high calcium caused by hyperparathyroidism, and an associated neuroendocrine tumour

Genetic testing is a new field and many patients may find this daunting, therefore testing has to be discussed on a case by case. Advantages of testing includes knowing to look for other associated conditions, earlier disease pick up in family members, and reassurance for family who don't carry the mutation. There are also disadvantages such as anxiety for the patient & family member if a mutation is found; testing may identify mutations that we are still uncertain if they are disease causing. Also there may be insurance implications in the future so discussion with your Endocrinologist and referral to genetics service is recommended if considering testing.

For more info go to “The genetics background of acromegaly” by M. Gaelha et al. Published in journal Pituitary Feb 2017. [https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5334425/](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5334425/)

Member’s Story

Yvonne kindly shared her journey with us at the AGM, thank you!

My name is Yvonne and I am 76 years old. At the end of 2001 I was diagnosed with Acromegaly when I had a mild stroke. I had the tumour on my pituitary gland removed in February 2002.

The acromegaly affected my hands and feet, so I have numbness in my feet due to the nerves being squashed by the extra growth in my feet. I have enlarged hands and this has affected the bones in my hands and fingers, and I now have arthritis in both my fingers and knuckles on both hands. The worse part about having acromegaly is that my tongue continued to grow. This took
many years but it enlarged to the point I couldn’t shut my mouth properly and it pushed my teeth out to right angles. It got to the point that I was self-conscious of my large tongue and felt embarrassed when people commented on it.

I had been going to the same dentist since 1978 and at no time did he pick up that my teeth were moving out of position due to the pressure of my tongue on them. I mentioned at the last AGM meeting for the Acromegaly Society that dentists should be made aware of what to look for when dealing with patients on a regular basis. This should start with the dental students at Otago University.

My dentist retired earlier in 2016 and the new dentist told me that something could be done to reduce my tongue size to help me with eating and to help my self-esteem. So on the 23rd of May 2016 I had surgery to reduce my tongue. I was meant to be in the Intensive Care Unit for two days, but due to complications I was on full life support for sixteen days. That was a very scary time for my family and at one point they didn’t think I was going to pull through.

The specialist told me that I had the biggest tongue of anyone in New Zealand. It took a long time to get over the surgery, but my family was amazed at how different I looked, and for the first time in many years I could smile properly and they could see my teeth.

My family helped me so much with my recovery, especially my husband, making special meals each day that had to be pureed so I could eat them. I have now made headway with my recovery and I am able to eat foods that I have struggled with in the past.

I am still having ongoing issues, like tingling and nerve pain in my tongue which I hope will correct itself soon. I now look forward to getting on with my life without having the label of having the largest tongue in New Zealand.

Social Events for the year

- Combined Auckland/Waikato catch up to be held at Remuera Golf Club restaurant. Date: 1st or 2nd Saturday of November (To be confirmed)
- Christchurch & Wellington coffee catch ups - date/venue to be confirmed
- AGM 2018: 1st Saturday of May 2018 (5th May 2018) Venue TBC

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