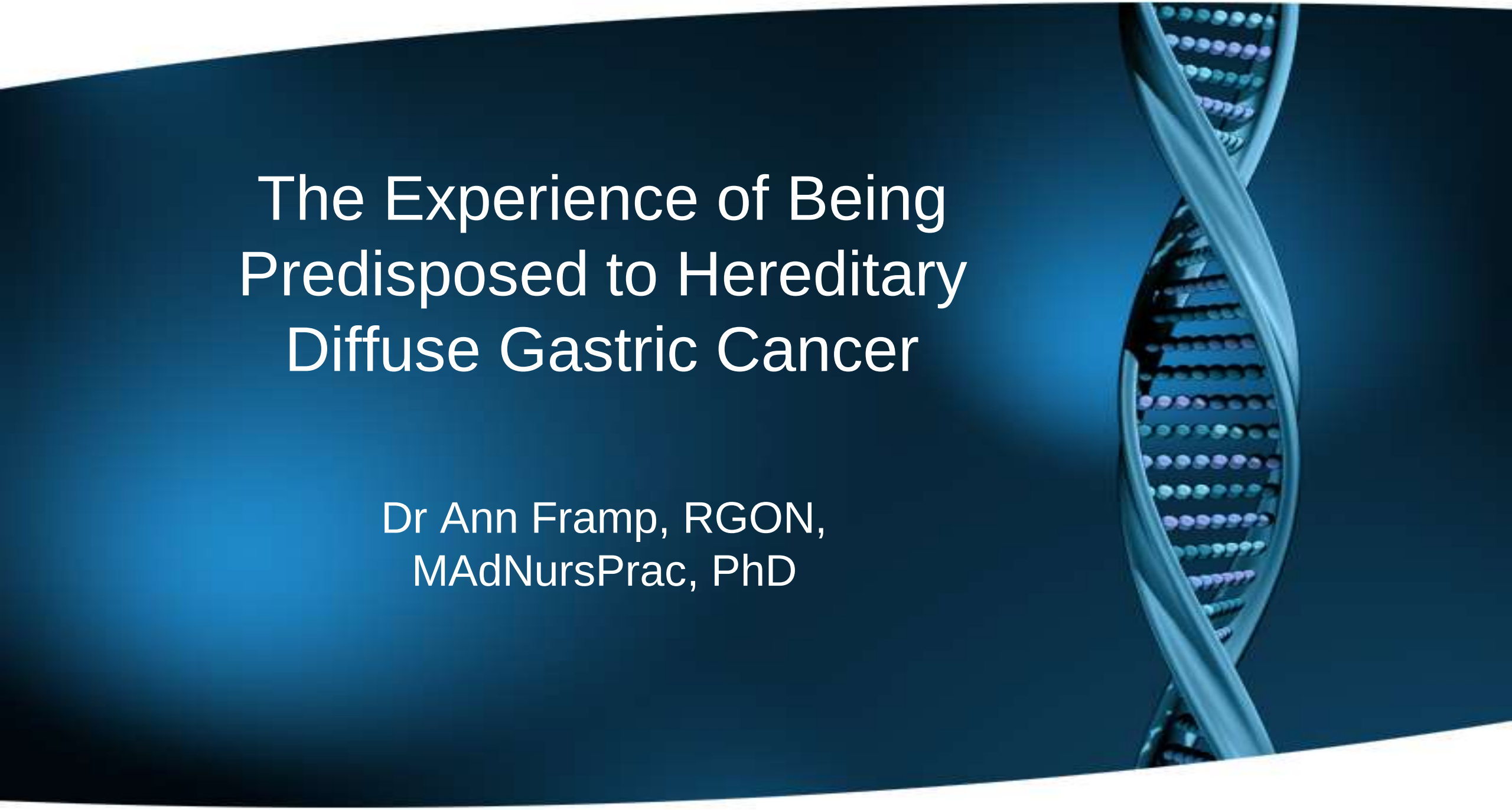
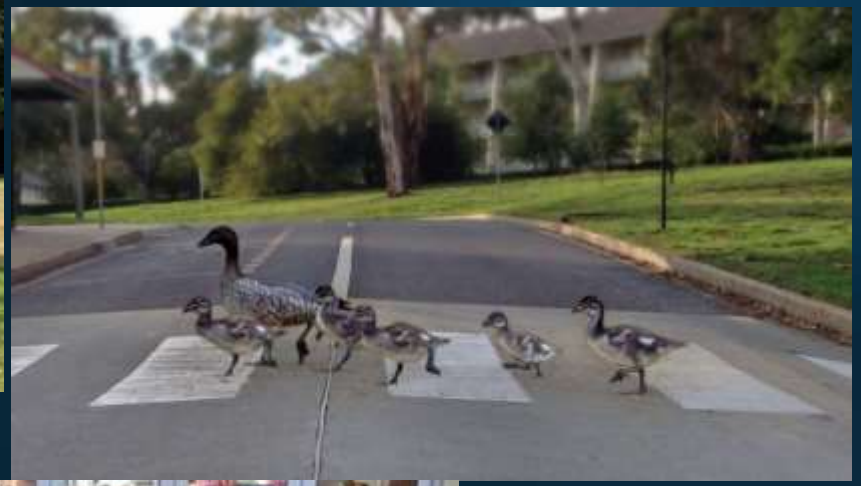
# The Experience of Being Predisposed to Hereditary Diffuse Gastric Cancer

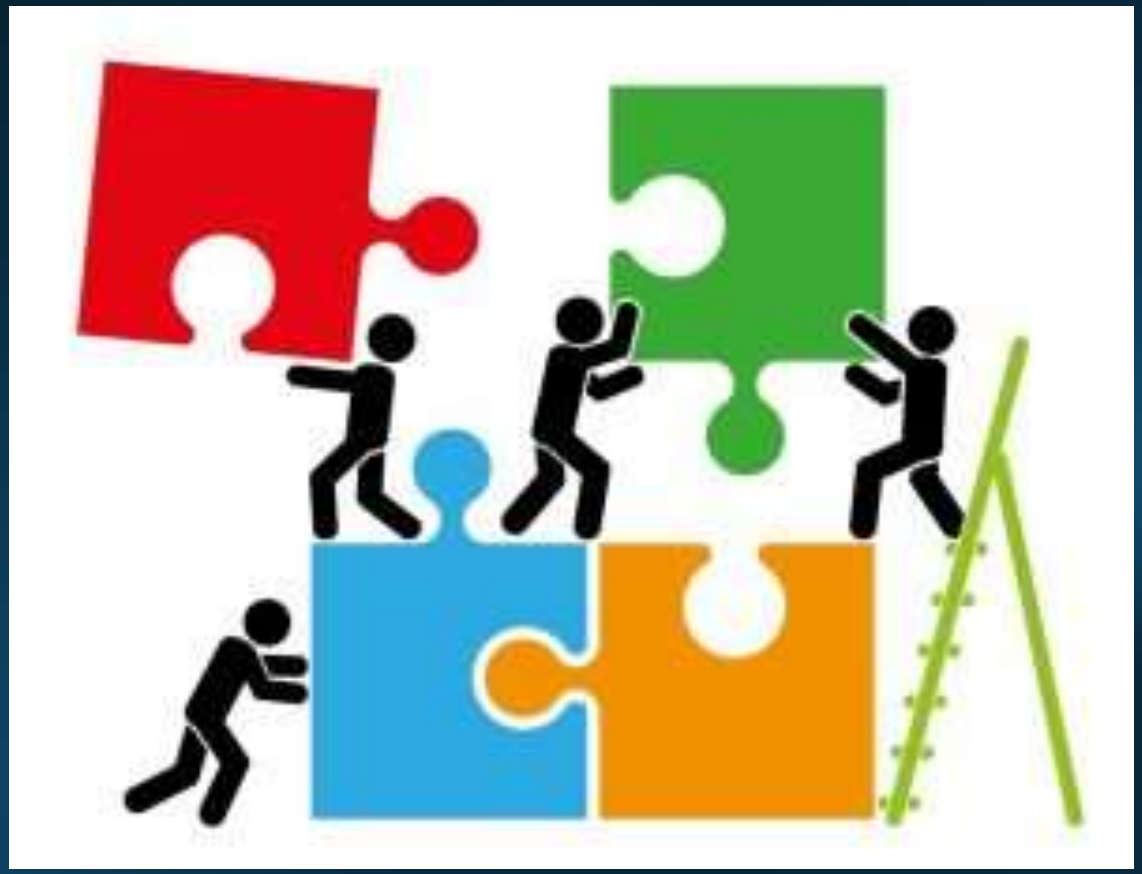
Dr Ann Framp, RGON, MAdNursPrac, PhD



# University of the Sunshine Coast Queensland, Australia




"OUR ENDOSCOPE IS BROKEN, BUT LUCKILY NURSE HAS HER MOBILE PHONE CAMERA — ITS QUITE SMALL..."
CARTOONSTOCK
Search ID: shu0185



https://resourcedevelopment.net/creating-opportunities-for-collaboration-and-teamwork/

# Hereditary Diffuse Gastric Cancer

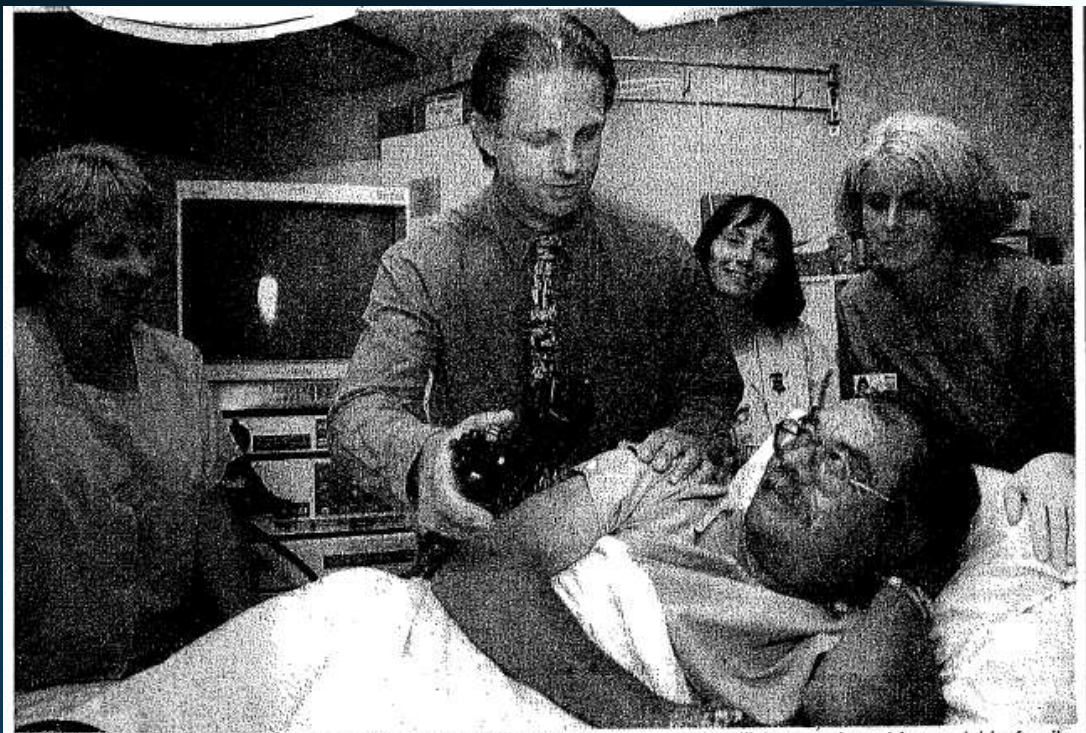
- Accounts for  1-3% of all incidences of gastric cancer worldwide (Mi et al. 2018)

- A genetically inherited aggressive form of stomach cancer that originates in the deeper layers of the gastric mucosa

# Hereditary diffuse gastric cancer

- Mutated gene discovered in 1998 (Guilford et al. 1998)

- Cumulative risk of gastric cancer by 80 years of age is 70% for men and 56% for women
- Average age of onset 38 years



DAVID Shaw shows Rangi McLeod the new ultrasound machine which will be used on him and his family. Looking on are, from left, Ann Framp, Lyn Elliot and Tricia Ogle. Picture: Ross Brown E0077

# Camera on family stomach plight

By Julia Holmes

MEMBERS of a Mount Maunganui family who suffer from a rare stomach cancer came under the international spotlight yesterday.

A crew from the BBC science programme Tomorrow's World was filming at Tauranga Hospital where doctors began screening members of the McLeod family who have been identified as being at risk.

It is the first screening to take place since genetic testing was used to identify which family members carried the gene responsible for the cancer.

The screening co-incided with the arrival of an endoscopic ultrasound machine which would help doctors detect cancer early enough for treatment.

Three of the McLeod family who are gene positive were yesterday tested using standard endoscopic procedures and vital dyes. The dyes are absorbed differently according to the tissue type and show up any abnormalities in the stomach lining.

Gastroenterologist David Shaw said the procedures were not new but it was the first time they had run a continuous list of people.

If any abnormalities showed up further investigatory procedures would be employed. This would include the use of the ultrasound machine.

"It is very sensitive technology which is able to pick up very small stomach cancers and the depth of invasion. It's the best method of doing that."

The $260,000 machine is the only one of its kind in New Zealand and was bought mainly to reduce the family's death rate from the hard-to-find cancer.

Dr Shaw and radiologist Gerrard Eager will go to Japan and Australia to learn how to use the ultrasound machine.

The cancer is a particularly aggressive type that develops in the lining of the stomach. This means it is undetectable by regular endoscopic techniques. As a result it spreads to other organs before any clinical signs appear and by that stage life expectancy is no longer than six weeks.

Seven out of 10 people identified as having the gene are expected to develop cancer.

There are about 10,000 descendants of the McLeod family throughout New Zealand.

The McLeod project is the subject of a joint research project with Otago University scientists and the specially formed Kimihauora health clinic.

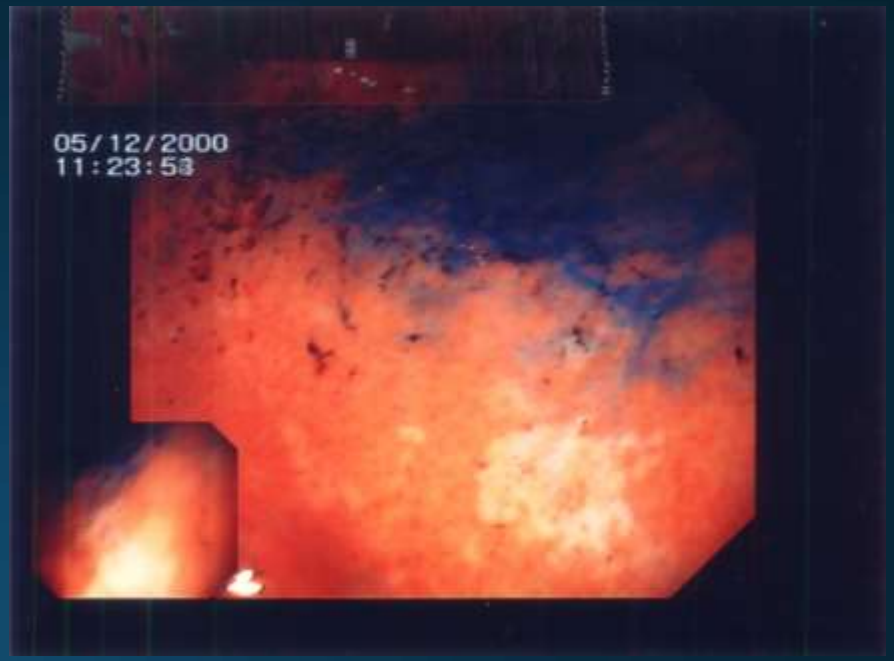
It has produced the world's largest genealogical record of a disease.

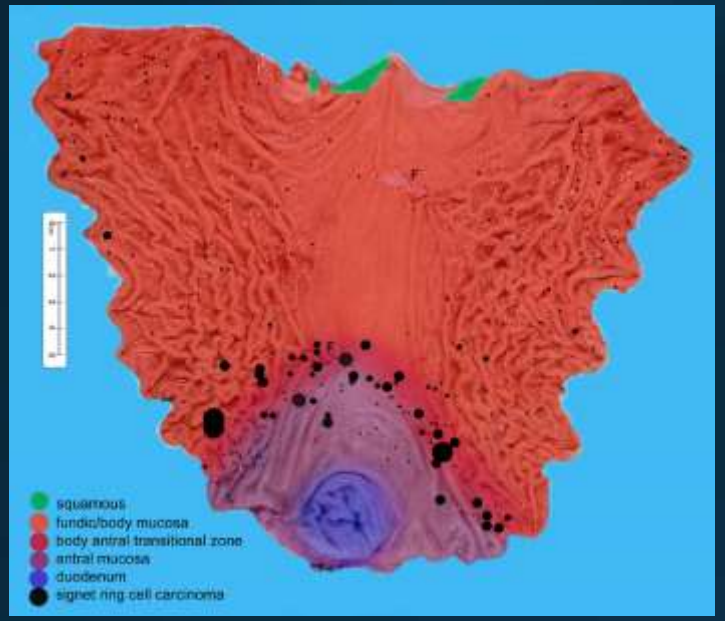
# Chromoendoscopy


https://www.healthproductsexpress.com/congo-red-stain-by-american-m

https://www.birchmeier.com/en/content/produkte/rose-star-1.25/index.php

# Chromoendoscopy



# Recommendations:

## Prophylactic gastrectomy preferred

For those unable or unwilling to have surgery:

- Detailed 30 minuter upper endoscopy every 6 – 12 months with multiple random biopsies
- Screening beginning 5 – 10 year prior to the earliest cancer diagnosis in the family

Recommend care by a multidisciplinary team with expertise in clinical genetics, gastric surgery, gastroenterology, pathology, and nutrition.

(Kaurah & Huntsman 2018; van der Post et al. 2015)

# PhD Study – A Narrative Inquiry into the Experiences of One Family's Predisposition to Hereditary Diffuse Gastric Cancer

- To understand the experience of living in the world predisposed to hereditary diffuse gastric cancer (HDGC)
- To build awareness of the unique psychosocial challenges related to this disease in order to deliver care sensitive to the specific needs of those predisposed to HDGC
- To contribute to existing knowledge about HDGC and other genetically inherited disease

# Methodology: Narrative Inquiry

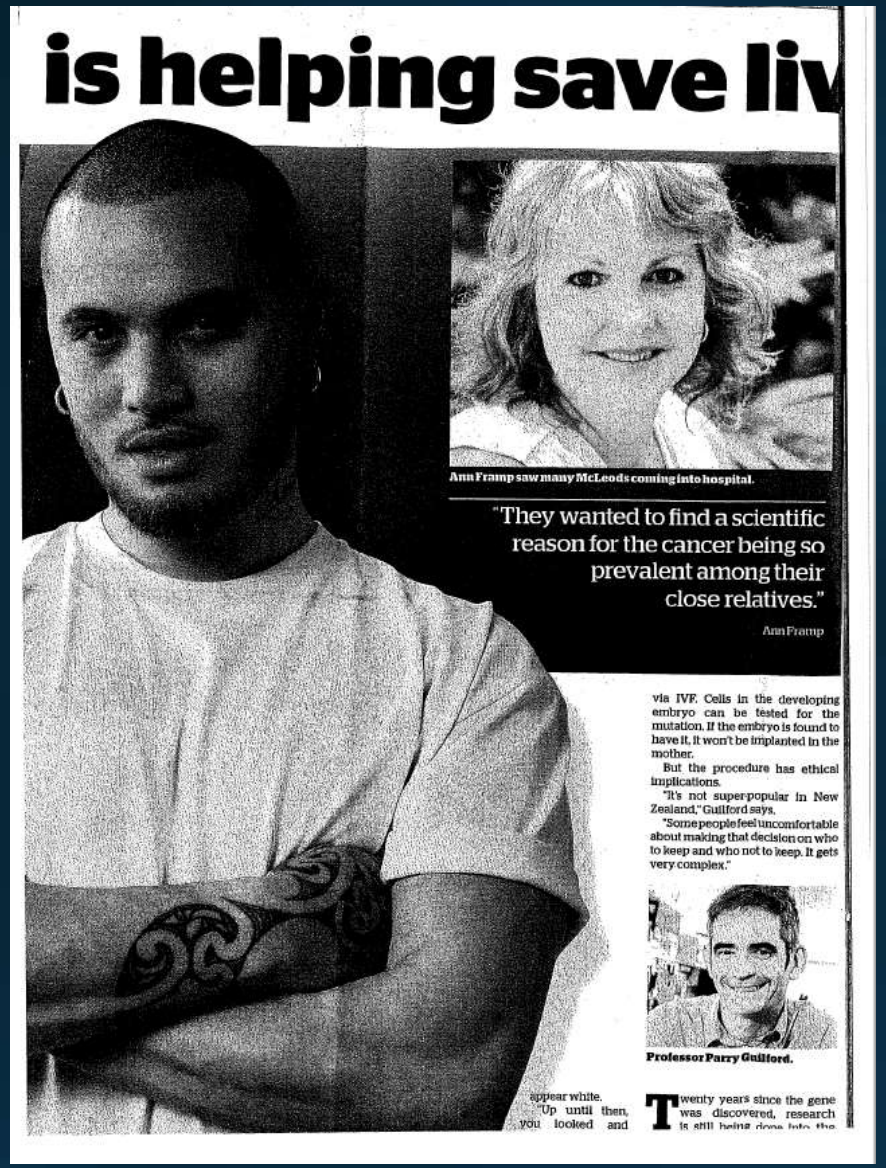
- Stories are a powerful portrayal of interpretation and meaning of life (Moules & Streitberger 1997)
- Provides a humanistic and social understanding of the person with the illness

My first memories about the cancer are when I was 16 and my cousin was dying from stomach cancer. When I was in my thirties, I was at my brother's unveiling ceremony my younger sister was sobbing for my dead brother. She died from the cancer herself that same month. My older sister had already died from the cancer and around that time I also remember some of my cousins being admitted to hospital, but there was nothing that could be done to help them, so they were also sent home to die. My older sister's son died at 31. When I visited him before he died, he was crying and pleading for my help – I remember feeling so helpless.

I remember sitting with my three children in the shed waiting for them to open their test results to find out if they had the gene that caused the stomach cancer. Two of the children were my sisters' children as I was now looking after them since she passed away. This day just happened to be on the anniversary of their mother's death. I remember the relief as my son and niece opened their results and they saw they did not have the mutated gene, but then I watched my nephew open his result and he just hung his head (*crying)*.

**Prophylactic Gastrectomy:**

- Mortality rate 2 – 4%
- Early post-operative complications 10 – 20%
- Late post-operative complications 100%



is helping save liv

Ann Framp saw many McLeods coming into hospital.

"They wanted to find a scientific reason for the cancer being so prevalent among their close relatives."

Ann Framp

via IVF. Cells in the developing embryo can be tested for the mutation. If the embryo is found to have it, it won't be implanted in the mother.

But the procedure has ethical implications.

"It's not super-popular in New Zealand," Guilford says.

"Some people feel uncomfortable about making that decision on who to keep and who not to keep. It gets very complex."

Professor Parry Guilford.

appear white.

"Up until then, you looked and

Twenty years since the gene was discovered, research

My body has changed since I had my stomach out. I can be asleep and then I wake up to these bright flickering lights, I start sweating and then I know I have to get up and eat something like chocolate.

Food tastes differently to what it did before, and the way I eat is different. It's not the same as before. It's just not the same. It is so frustrating when I am eating and food becomes stuck and I have to wait for it to pass through and then I just lose my appetite. I get so tired from chewing my food so much. Yeah, that's a problem, the eating and the tiredness.

When I got home, I could only eat seafood and toast. I even tried baby food but I just vomited it up. I felt like I had to force feed myself and it took just under two years to eat a normal meal. I still can’t eat bread and some meats.  After the surgery, I got sudden bouts of diarrhoea and had to carry spare undies everywhere I went.

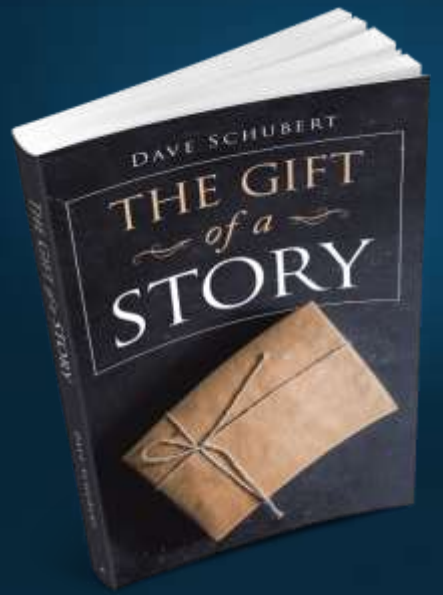
After my operation, I began eating the chicken stew that was brought to me but suddenly the nurse came into the room and snatched it away from me and said “you can’t have that!” I looked at the nurse in amazement and said “do you think I am a Maori in a grass skirt or something?”

After the operation Lucy and I started to worry about Hemi.  We were trying to tell the nurses that something wasn't right but they were not listening and trying to get him up.  We knew - we could see it – we had been through it already – myself and my two older children. We were trying to tell them and they wouldn't listen to us. Hemi had to go back for another operation because of complications. I was just so angry I had not been heard and I lost total confidence.

One of the things that helped me recover was the nurses – it was the nurses who helped me get it together. I had been in there so long because of all the complications and they knew they needed to do something to get me out of there because they could see I was slipping into a depression. I thought that nurses just see you as another person they have to look after, but now I understand how important patients are to nurses. I got along really well with some of them; it was like my illness became personal to them as well.

It has been eleven years since we began the genetic testing and we've had some setbacks, but the recovery time is getting shorter and the operation is getting faster. I've always said we're the lucky ones you know. At least we had a choice ay? We had a choice. Everyone else who died never had that choice.

*To give the gift of listening is to appreciate receiving the gift of a story. Not just understanding this reciprocity but embracing it seems to me to be the beginning of clinical work (Frank 1998)*



# Reference List


Frank, A. (1998). Just listening: narrative and deep illness. *Families, Systems & Health, 16*(3), 197-212.

Guilford, P., Hopkins, J., Harawira, J., McLeod, M., McLeod, N., Harawira, P., Taite, J., Scoular, R., Miller, A,. & Reeve, A. (1998). E-cadherin germline mutations in familial gastric cancer. *Nature*, *392*, 402 – 405.

Kaurah P., Huntsman D.G., (2002). Hereditary diffuse gastric cancer. In M.P. Adam, H.H. Ardinger, R.A. Pagon et al. (Eds.), *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019.

Mi, E.Z., Mi, E.Z., di Pietro, M., O'Donovan, M., Hardwick, R.H., Richardson, S., Ziauddeen, H., Fletcher, P.C., Caldas, C., Tischkowitz, M., Ragunath, K., Fitzgerald, R.C. (2018). Comparative study of endoscopic surveillance in hereditary gastric cancer according to CDH1 mutation status. *Gastrointestinal Endoscopy, 87*(2), 408-418.

Shaw, D., Blair, V., Framp, A., Harawira, P., McLeod, M., Guilford, P., Parry, S., Charlton, A., Martin, I. (2005). Chromoendoscopic surveillance in hereditary diffuse gastric cancer: an alternative to prophylactic gastrectomy? *Gut 54*, 461–8.

van der Post, R., Vogelaar, I., Carneiro, F., Guilford, P., Huntsman, D., Hoogerbrugge, N., Caldas, C., Chelcun Schreiber,K., Hardwick, R., Ausems, M., Bardram, L., Benusiglio, P., Bisseling, T., Blair, V., Bleiker, E., Boussioutas, A., Cats, A., Coit, D., DeGregorio, L., Figueiredo, J., Ford, J., Heijkoop, E., Hermens, R., Humar, B., Kaurah, P., Keller, G., Lai, J., Ligtenberg, M., O'Donovan, M., Oliveira, C., Pinheiro, H., Ragunath, K., Rasenberg, R., Richardson, S., Roviello, F., Schackert, H., Seruca, R., Taylor, A,. ter Huurne, A., Tischkowitz, M., Tjon A., Joe, S., van Dijck, B., van Grieken, N., van Hillegersberg, R., van Sandick, J., Vehof, R., van Krieken, J., & Fitzgerald, R. (2015). Hereditary diffuse gastric cancer: updated clinical guidelines with an emphasis on germline CDH1 mutation carriers. *Journal of Medical Genetics 52*, 361 – 374.