

Poster Number 78: The chance for Hope and Quality of Life for Von Hippel Lindau Syndrome patients: A Case Study

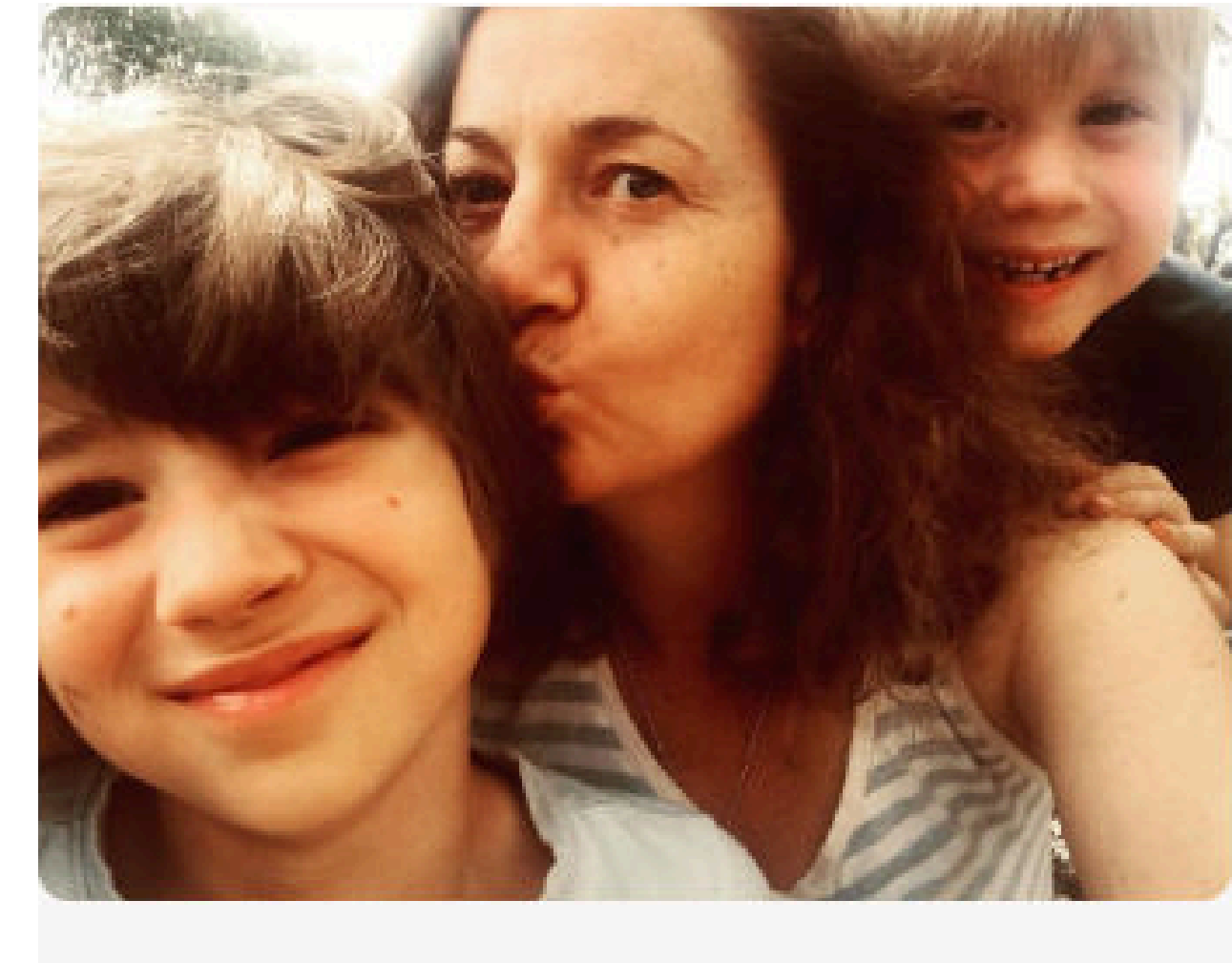
Author: M.Cummins
CEO NeuroEndocrine Cancer Australia

History/Background

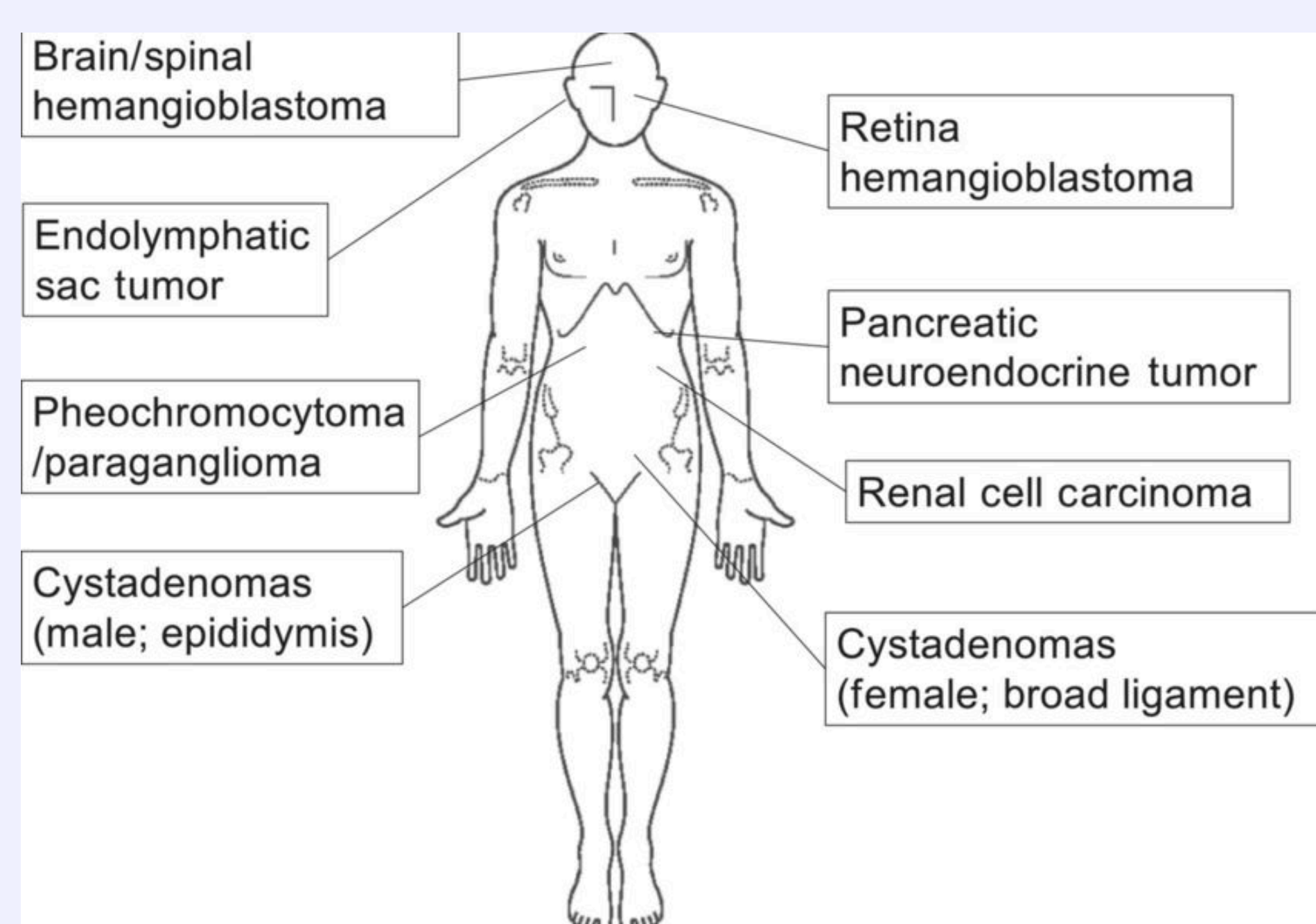
- ✓ Patients with Von Hippel Lindau (VHL) Syndrome have very limited treatment options. VHL is predominantly a hereditary condition with a genetic mutation.
- ✓ VHL causes Pancreatic Neuroendocrine Cancer, Renal Cell Carcinoma, tumours in the brain, retina, spinal cord and adrenal glands.
- ✓ A treatment is available in the US, however not in Australia, which can control the tumour growth reducing the need for ongoing surgery and improving quality of life (QOL).

“Mummy promise me you won’t kill yourself if you go blind” Lucy’s 10-year-old said.

“I won’t love” I promised him and myself no matter how hard my life gets.



Lucy had said in a conversation we had – **“without being able to see or hear my children – I have no Quality of Life.**



Assesment

Without access to this medication patients can only have surgery and / or radiation therapy.

A Case Study

Lucy is young mum of 10 & 8-years old boys.

She has been relentlessly attached by VHL

- Age 11 - eye tumours lasered, and since then ‘innumerable’ retina tumours in both eyes.
- Since the age of 19 she has had four surgeries on her brain to remove multiple tumours. Last year she learnt to walk again for the fifth time, caused by C5 tumour.
- Luckily neither of her children have the gene as she went through IVF
- VHL has left her deaf in one ear lost due to a brain tumour with hearing loss in the other.
- She is no longer able to drive as visually impaired.
- She still has tumours in her brain, spinal cord, and her pancreas.
- She also lives with neurological deficits such as weakness and sensation changes.
- She walks with a stick most days to steady herself.
- Recently scar tissue ripped the retina off my seeing eye – Lucy went blind in a day – thankfully it was able to be repaired

Goals of Care

Neuroendocrine Cancer Australia (NECA) could see the devastating impact of this disease and has advocated for the VHL community to expedite access to this vital medication – not a cure - however controls growth of tumours and improves QOL.

What is a petition?

A petition to the House of Representatives is a request for action on an issue which must be a matter for which the federal government has responsibility.

All petitions must include a reason and request.

PARLIAMENT OF AUSTRALIA

PETITIONS

Intervention (s)

NECA worked with the VHL community to highlight the need for this medication to reduce ongoing surgery, rehabilitation and disruption to their lives by media stories, House of Representative Petition (17,800 signatures), public consultations submitted to PBAC (519 submissions) and held a VHL Parliamentary Event in Canberra attended by Senators, MPs, patients and their families.

Evaluation/Outcome:

Through advocacy and community coming together we had a successful recommendation for listing on the PBS by PBAC in September 2024. In the interim VHL patients who meet the criteria have access to the medication free of charge. Advocacy and patient stories continued, and the medication was successfully listed on the PBS 1 December 2025- \$7.70 for concession card holders and \$31.60 for PBS script.

Conclusion/Implications for Practice:

- ✓ Prior to the inclusion of the medication to the PBS Lucy stated “Living with VHL is the never-ending story of hospital theatres, ICU, stints in rehab to regain functions, physio, monitoring scans then repeat Even after a tumour is resected, more pop up like weeds. I live in fear of when it will strike again and this takes a massive toll”
- ✓ To date patients are seeing tumour reduction meaning surgery is not looming and there is HOPE. Since commencing the medication Lucy has not had any further surgeries.



For the first time in my life living with this awful troll of a disease I have hope.