

GHRELIN

LEPTIN

- Act on the hypothalamus for energy homeostasis through a feedback loop
- Ghrelin – the Hunger Hormone – produced in the stomach muscle tissue when the stomach is empty – secretion stops when stomach muscle is stretched (full)
- Leptin – the Satiety Hormone – made in fat cells

GHRELIN ANALOG

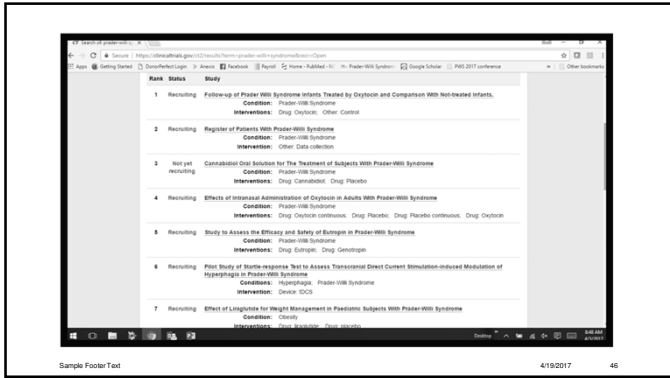
HYPERPHAGIA

- Acylated ghrelin increases appetite
- Unacylated ghrelin decreases appetite
- Available in France – phase two trial – 47 patients

DYSPHAGIA

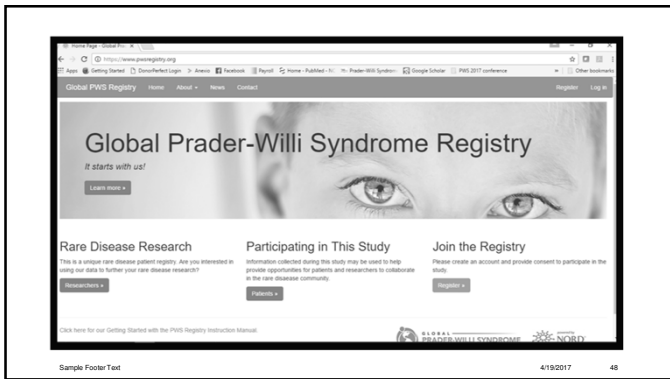
SILENT ASPIRATION

- Two studies with similar outcomes
- Videofluoroscopy - silent aspiration and dysphagia in infants (n=6)
- Incomplete clearance, silent aspiration, impaired coordination of swallowing



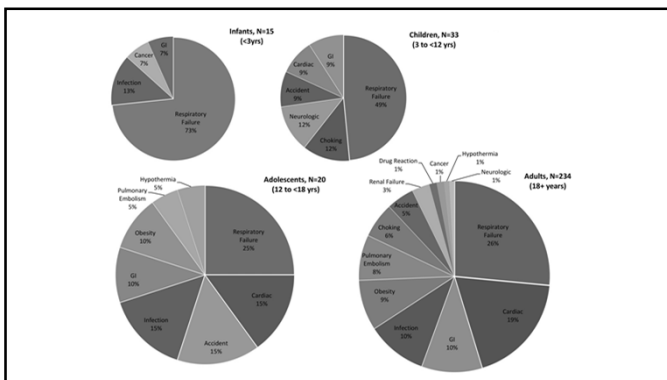
RESEARCH TREATMENTS

- Vagal nerve stimulation
- Transcranial stimulation
- Deep brain stimulation



DEATH RESEARCH

- 40 years (1973-2015; only 8% included an autopsy).
- 486 families reported a death between the ages of 2 months and 67 years
- 70% of these deaths occurred in adulthood
- 312 had a cause of death provided and 36% died "of unknown" cause
- These reports uncovered trends that were previously unknown – gastric rupture and blood clots



TRENDING DIETS



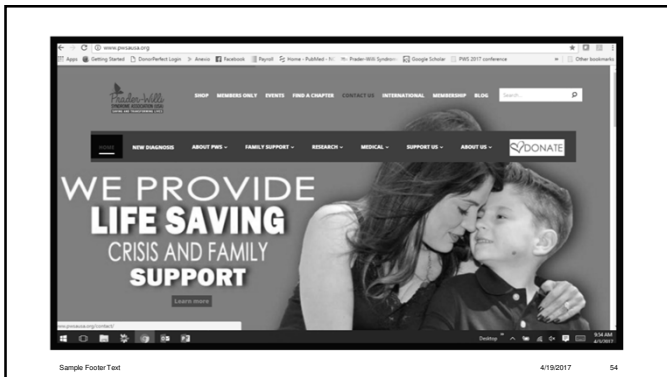
- Stoplight diet
- Modified ketogenic diet
- Soft, lower fiber diet
- Supplements, supplements, supplements

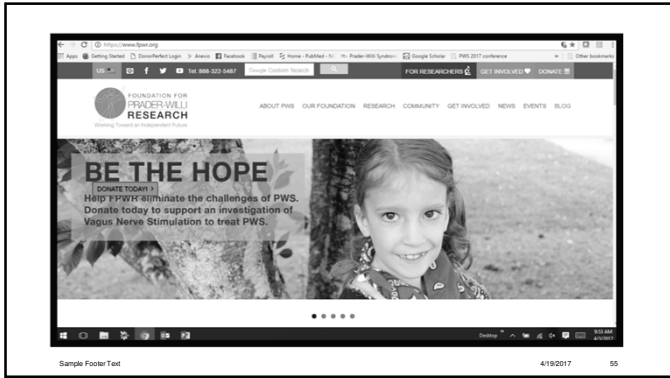
THINGS I DIDN'T KNOW

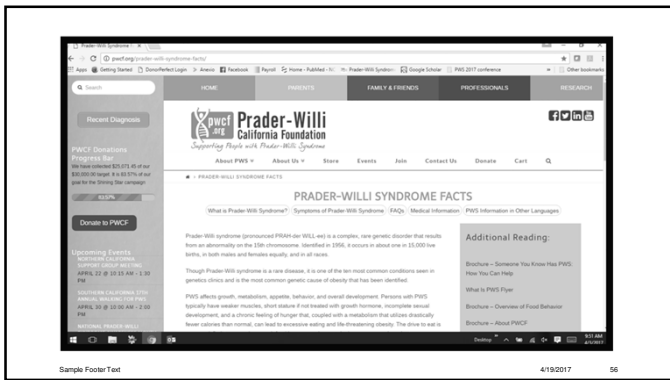
- People with PWS can have profound behavioral challenges starting in the teens
- People do not call PWSA with happy stories
- UPD has significant risk of psychosis (60%)
- UPD is more common in IVF and older parents – the ratios are changing
- There are 2 PWS organizations in the US
- For people with developmental disabilities, living at home after high school is seldom the best plan

THINGS I DID KNOW

- Growth hormone is a game changer in PWS but we don't know how to dose it
- Parents prefer to ask other parents on Facebook for advice rather than make a phone call
- A little compassion goes a long way
- Working from home is pretty sweet







PHARMACOGENETICS

- **Pharmacogenetics** is the study of inherited genetic differences in drug metabolic pathways which can affect individual responses to drugs, both in terms of therapeutic effect as well as adverse effects
- Antidepressants, oncology meds, pain treatment
- Liver enzyme CYP2D6 – converts morphine into codeine – could cause overdose if the individual has more than one copy of this gene
- CYP 450 testing can help determine psychotropic choices in PWS

