The impact of permanent muscle weakness on quality of life in periodic paralysis: a survey of 66 patients.

What is Permanent Muscle Weakness?

The PPs cause a gradual loss of muscle power as working muscle is gradually replaced by fibrous tissue and fat, defined as permanent muscle weakness. (PMW)

Early signs of PMW begin to be noticeable in the patient’s 40s-50s. Decreased endurance and increased muscle fatigue are common, time needed for recovery after activity increases.
Our Participants

66 patients participated; 20 males and 46 females. Participants were 41 - 82 years old:
- 11 between 70-82
- 9 between 60-70
- 24 between 50-60
- 22 between 40-50.
Participants Have These Forms of Periodic Paralysis

- 42 (64%) paralytic form of hypokalemic periodic paralysis
- 4 (6%) myopathic form of hypokalemic periodic paralysis
- 6 (9%) hyperkalemic periodic paralysis
- 2 (3%) hyperkalemic PP + paramyotonia congenita
- 6 (9%) Andersen-Tawil Syndrome
- 4 (6%) Not yet determined – i.e. normokalemic PP
Participant’s Identified Mutations

HypoKPP: 15 total
8 with R528H, 4 with R1239H
1 with R897T, 1 unpublished mutation, 1 told they have a “Hypo mutation”

HyperKPP: 3 total
1 with T704M; 1 with P875S; 1 told they have a “Hyper mutation”

ATS: 2 both R67W

PMC: 1 with T1313M
41 (62%) have a family history of PP

16 (24%) are unsure

9 (14%) have no known family history
Age at Onset

Symptoms began in childhood or puberty in 74% of patients
There was an average of 26 years between age of onset and age at diagnosis.
Magic 40\textsuperscript{th} Birthday!
Abortive attacks:
are periods of reduced strength which fluctuates over days to months, never progress to paralysis
serum K+ may not leave normal range.
can last so long that they are mistaken for permanent weakness
are treatable!
Episodic Weakness

No Episodes – 11%
Daily Episodes - 28%
Weekly Episodes – 59%

Decreased frequency – 21%
Unchanged frequency – 64%
Increased frequency – 11%

Less severe - 30%
Unchanged severity - 58%
PMW in Our Participants

We Asked:

❖ Do you have muscle weakness which is always present and varies little from day to day?
❖ Yes – 45 (68%)
❖ Unsure – 15 (23%)
❖ No – 6 (9%)
Muscle Groups Affected?

- face/jaw 3 (6%)
- trunk/chest 9 (18%)
- gluts 11 (22%)
- forearms 12 (24%)
- neck/throat 13 (27%)
- hands 13 (27%)
- lower back 13 (27%)
- shoulder girdle 18 (37%)
- calves 18 (37%)
- upper arms 21 (43%)
- hip girdle 27 (55%)
- quads 40 (82%)
Pain, Fatigue and Muscle Stiffness

Fatigue:
✧ 59 participants (89%) reported daily fatigue
✧ 39 (66%) report fatigue of moderate to severe intensity

Pain:
✧ 54 (82%) reported daily pain
✧ 23 (43%) reported pain of moderate to severe intensity

Muscle Stiffness:
✧ 44 (67%) reported daily muscle stiffness
✧ 26 (59%) reported moderate to severe muscle stiffness
Activity levels between 18-35
Activity levels at time of survey
Incapacity

92% - reduced strength & stamina
89% - difficulty performing ADL
89% - limited in the work they can do
75% - hard to do mild exercise
55% - lack of endurance,
42% - climb only one flight of stairs
25% - unable to walk more than a few steps unaided
91% - dissatisfied with what they are able to accomplish.
Medications

Medication: Vital to have an aggressive enough medication regime to prevent weakness as much as possible.

If commonly used meds don't work, research, seek advice on meds used successfully by others.

Don't accept that nothing more can be done until you have tried **everything**!

Possible Alternatives:

**Hypo:** baclofen, gabapentin, methazolamide, 3,4-DAP

**Hyper:** Tegretol, florinef, Avapro, methazolamide

**PMC:** Paxil

**ATS:** triamterene
Falls:

67% had fallen and injured themselves seriously enough to require medical attention.

The incapacity & injuries caused by falling have not been given enough attention.
Use of mobility aids

- 17% use a walker
- 20% use a scooter
- 23% use a wheelchair
- 29% use a cane
- 26% use other aids
Treatment for other problems

Statins given to lower cholesterol cause muscle destruction in patients with a variant in either of two genes. PP patients should be tested for these variants before starting any statin drug.

Testing is available through 23andMe for $99 plus shipping USF

www.23andMe.com
Nutrition

Eat right diet for type:
In ALL Types small frequent meals
Hypo: High protein, low simple carb, low sodium
Hyper: High protein, high complex carb,
PMC: Varies with type and phase
ATS: Varies with form (Hypo/Hyper)
Supplement as needed, creatine, vit D, fish oil
57% had professional physiotherapy. Of these:
49% had significant or mild benefit
40% experienced deterioration
11% no change.
83% followed self-directed programs of exercise, of these:
62% had significant or mild benefit
16% experienced deterioration
22% no change.
Mental Health

Emotional lability is often a signal that serum K+ is low.

35% of women and 33% of men reported depression.

Some anti-depressants cause increased weakness, but try different ones until you find one that works for you!
It's Your Life!

Stay active as possible, insist on an aggressive enough medication regime to reduce weakness to the absolute minimum, avoid triggers, treat any depression, stay in touch with the PP community, carry your medical information always, and advocate for yourself! We have to do all we can to prevent PMW or postpone its development as long as possible in order to lead satisfying lives.
Thank you!

The paper this talk was based on is available for download in pdf format from a link on the front page of: www.HKPP.org