

INTRODUCTION

- Pachyonychia Congenita is a rare congenital dermatological disorder affecting the keratin gene.
- Mutations in the keratin genes KRT6A and KRT16 are associated with type 1 phenotype and KRT6B and KRT17 genes with type 2.
- All patients affected by this disorder have specific changes of the nails and skin. Most common abnormalities:
 - hypertrophic nail dystrophy
 - palmar keratoderma
 - plantar keratoderma
 - plantar pain
- The plantar pain is due to formation of blisters in the affected areas of hyperkeratosis

PATIENT HISTORY

- 11yo female with Pachyonychia Congenita type 1
- Referred to our pain clinic by her dermatologist at 8yo
- Presented with extensive blisters and keratoderma on her feet. Her feet looked similar to the figure 1 photo.
- She refused to walk 2/2 pain. She ambulated with a wheelchair
- Our patient was unable to participate in activities of daily living
- Multiple hospital admissions for infections

USUAL TREATMENT FOR PLANTAR HYPERKERATOSIS

- Emollients
- Keratolytics
- Comfortable shoes
- Debridement of nails and areas of hyperkeratosis

BOTOX

- Works by cleaving a protein involved in the release of acetylcholine from the axonal terminal.
- Intradermal administration produces a chemical denervation of the sweat gland.
- Botox is used for treating hyperhidrosis.
- Botox use has been reported in the treatment of hyperhidrosis in adult patients with Pachyonychia Congenita.

Figure 1. Plantar Keratoderma



Images copied from pachyonychia.org

Fig 2: Improvement after Botox, Fall 2017



Fig 3: Improvement after Botox, Jan 2018



PATIENT'S TREATMENT

- Intradermal Botox injections q3-6 months over 3 years.
- Average dose of Botox administered was 50 units per foot.
- Immediate improvement in plantar pain had been reported.
- Improvement in hyperkeratosis and blister formation was observed (Figure 1 & 2).
- Now she is able to ambulate and participate in activities of daily living. She even started playing soccer this fall.
- No hospitalizations since starting Botox.

DISCUSSION

- Significant improvement in plantar pain and foot hygiene.
- No adverse side effects to three years of botox treatment.
- Pediatric patients with Pachyonychia Congenita may benefit from intradermal Botox injections for treatment of plantar hyperhidrosis.

References

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