

# Vaccine and Immunization-Research and Development

## **Case Report**

# Cure Of Cowden'S Diseases

## **Kunal Joon\***

University of Noida International Institute of Medical Sciences.

**Corresponding Author:** Kunal Joon, University of Noida International Institute of Medical Sciences.

**Received: ≅** 2024 Feb 15

Published: 

2024 Mar 20

## **Abstract**

It is the rare autosomal dominant diseases, multiple hamartoma syndrome with characteristics of mucouocutaneous lesion. It is related with abnormalities in breast, GIT, and lead to multiple hamartoma in GIT like Trichiliemmomas, oral papillomatosis, facial papules and acral keratosis. This disease derived from the mutation of PTEN gene.

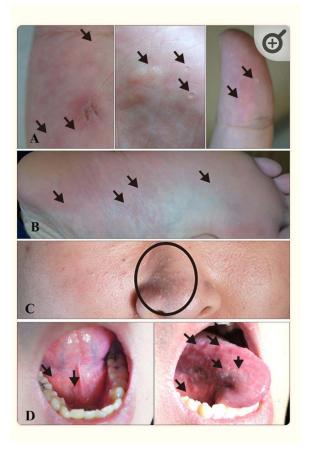
Keywords: Hamartoma Syndrome, Multiple, Pten Phosphohydrolase and Skin Neoplasm.

#### 1. Introduction

Cowden 's diseases is the autosomal dominant diseases and can lead to cause thyroid , breast and other types of cancers and also the high growth of benign hamartoma or over growth on skin or any organ like thyroid . There are also

report of sporadic case [1]. The common mucouocutaneous finding is facial Trichiliemmomas and oral papillomas. Cowden 's diseases is the most common of the phosphatase and tensin homolog (PTEN) hamartomatous which include other syndrome like Proteus syndrome [2].

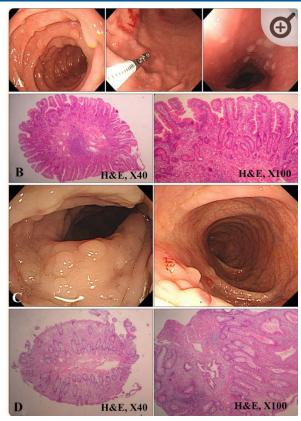
## Case Report Images of 21 Years Old



**Figure 1:** Showing multiple punctate hyperkeratotic male are localized on both 1) palms and both 2) soles (black arrow) [3]. Multiple skin coloured papules are observed on his 3) nose (black arrows) [4]. On his tongue and oral mucosa, several erythematous papules are found resembling cobblestone or fibroma like – polyps.

Also observed abnormal esophagogastroduodenoscopy and colonfibroscopy as shown in figure (2).

Volume - 1 Issue - 1



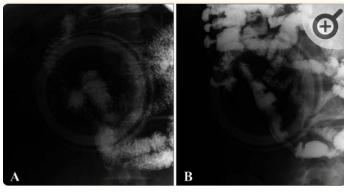
**Figure 2:** (A) Endoscopic photograph showing multiple hamartomas papillomatosis in the duodenum and stomach, and oesophageal keratosis like nodular lesions in the oesophagus [5]. (B) By biopsy of the gastric lesion and polypectomy, histopathology of a gastric polyp reveals a hyperplastic and inflammatory polyp with prominent reactive lymphoid follicle (H&E; ×40, ×100). (C) Colonoscopy shows several small polyps [6]. (D) Histopathology of a colorectal polyp shows it to be inflammatory (H&E; ×40, ×100).

Also skin biopsy was performed on palm and forehead. Biopsy of a punctate lesions show palmoplantar keratosis with a large hyperkeratotic mount and thickened granular layer [7].



**Figure 3:** Haematoxylin and eosin (H&E) stained specimen from palm shows a large hyperkeratotic mount with thickened granular layer [8]. Typical histologic backgrounds of palmoplantar keratosis, including marked hyperkeratosis and epidermal proliferation, are noted (H&E, ×40). (B) Epidermal hyperplasia with hypergranulosis, a thin layer of parakeratosis, and overlying orthokeratosis can be observed [9]. Slightly dilated blood vessels in the papillary dermis and a sparse infiltrate of lymphocytes are noted (H&E, ×100). (C, D) A specimen from forehead shows plugging of the hair follicle by keratin with typical histologic backgrounds of acne including perifollicular lymphocyte infiltration (H&E; C: ×40, D: ×100).

The small bowel series shows a filling defect lesion which suggest a localized polyp on junction of duodenum as shown in figure 4.



**Figure 4:** The small bowel series shows (A) Filling defect lesion on the junction of duodenum 2nd and 3rd portion (2 cm long). (B) No other specific filling defect can be observed in the small bowel loops, especially on the terminal ileum [10].

### **Genetics Analysis**

Defect in the gene of PTEN sequencing on chromosome 10q23 was observed and is a result of DNA backward rolling and lead to mutation.

#### **Treatment**

Genetherapy and the monoclonal antibody with the corrected gene can help and most suitable is the treatment by correct PTEN gene in the stem cells or stem cell therapy with the correct PTEN gene from amniocentesis cells.

#### 2. Discussion

- Cowden diseases
- Effect of Cowden diseases in the body
- Treatment of Cowden disease

### References

- 1. Seol, J. E., Park, I. H., Lee, W., Kim, H., Seo, J. K., et al (2015). Cowden syndrome with a novel germline PTEN mutation and an unusual clinical course. Annals of dermatology, 27(3), 306.
- 2. HONG, E. J., KIM, H. K., CHO, Y. S., JI, J. S., KIM, C. W., et al (2006). A case of Cowden syndrome associated with breast cancer and thyroid cancer. Korean Journal of Gastrointestinal Endoscopy, 293-297.
- 3. Tural, E. (2020). 2D ve 3D ishikawa endometrial karsinom hücre kültüründe lityum, metformin ve everolimus maddelerinin hücre büyümesi üzerine etkileri.
- 4. Lee, H. R., Moon, Y. S., Yeom, C. H., Kim, K. W., Chun, J. Y., et al (1997). Cowden's disease-a report on the first case in Korea and literature review.
- 5. Bagan, J. V., Penarrocha, M., & Vera-Sempere, F. (1989). Cowden syndrome: clinical and pathological considerations in two new cases. Journal of oral and maxillofacial surgery, 47(3), 291-294.
- Ward, S. K., Roenigk, H. H., & Gordon, K. B. (1998). Dermatologic manifestations of gastrointestinal disorders. Gastroenterology Clinics of North America, 27(3), 615-636.

Volume - 1 Issue - 1

- 7. Gustafson, S., Zbuk, K. M., Scacheri, C., & Eng, C. (2007, October). Cowden syndrome. In Seminars in oncology (Vol. 34, No. 5, pp. 428-434). WB Saunders.
- 8. Salem, O. S., & Steck, W. D. (1983). Cowden's disease (multiple hamartoma and neoplasia syndrome): A case report and review of the English literature. Journal of the American Academy of Dermatology, 8(5), 686-696.
- 9. Eng, C. (2000). Will the real Cowden syndrome please stand up: revised diagnostic criteria? Journal of medical genetics, 37(11), 828-830.
- 10. Son, J. H., Chung, B. Y., Jung, M. J., Choi, Y. W., Kim, H. O., et al (2019). Cowden disease: case report and review of the literature. Annals of dermatology, 31(3), 325.