# CASE REPORT: THE ORAL HEALTH STATUS OF SIBLINGS WITH SUSPECTED ICHTHYOSIS IN A PERI-URBAN COMMUNITY OF THE CENTRAL REGION OF GHANA.

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## ABSTRACT

Ichthyosis is a dermatological condition characterized by the presence of dry, scaly skin that sometimes resembles fish scales. The condition may be present at birth, in childhood, or later. Diagnosis is based on clinical features and sometimes on histological features. Management involves a multidisciplinary approach. No cure is currently available. It has an adverse effect on health-related quality of life with a positive correlation with the severity of the disease.
There is a need for awareness and the establishment of treatment protocols to manage individuals with this condition.
This is a case report of two adult siblings with the condition but at different phases.

**Keywords**
Ichthyosis, Quality of life, Oral health, Dermatology, Scaly.

## INTRODUCTION

Ichthyosis is a descriptive term for dry, scaly skin that, in some cases, may resemble the scales on a fish, sometimes referred to as hyperkeratosis.[1,2] Ichthyoses are the phenotypic results of gene mutations that lead to an imbalance of stratum corneum homeostasis and skin barrier failure. It is a rare, heterogeneous group of cornification disorders that typically affect the entire skin.[1,7] Ichthyosis can be present at birth, developed in childhood,[7] or acquired later in life.[7,9,10] Incidence and prevalence rates vary depending on the type of Ichthyosis and the population studied; however, it can be found in both sexes and ill races; at least 20 different varieties have been identified.[12,13]Formally infants with Ichthyosis die within the first three months of life. Improvement in neonatal care and early introduction of retinoids have increased survival rate.[4]

It may be diagnosed based on clinical features,[18] histologic findings, and gene mapping. It can be classified into syndromic and non-syndromic types.[7,18,20] Other clinical symptoms are ectropion, ocular changes, deafness, alopecia, diminished sweating, dermatosis, nail dystrophy, and affected individuals prone to infections.5 The underlying defect, mode of inheritance, character, and extent of scaling differ among the various types, with symptoms varying from mild to life-threatening conditions.[22,23]

No cure is currently available for Ichthyosis, and a multidisciplinary approach is employed in its management.[24] Applying skin-softening emollients after bathing while the skin is still moist with lotions containing alpha-hydroxy acids, urea, or propylene glycol is most effective.

Like many dermatologic conditions, the condition's impact extends beyond the skin. Disease manifestations, symptoms, and daily care affect the patient's self-perception and different patient and family life dimensions.[26]

Recent publications have described the quality of life of people living with Ichthyosis to be correlated with the severity of symptoms and female gender. Time of treatment and advancing age significantly impacted quality of life.[27]

Concerning an individual's oral health-related quality of life, four main dimensions can be considered. These include oral function, oro-facial pain, oro-facial appearance, and psychosocial impact. The oro-facial appearance was found to moderately impact people with aesthetic compromise.

The appropriate needs assessment will help rapidly identify distressing family situations for appropriate and prompt intervention. Unfortunately, to date, there is only a French validated ichthyosis-specific questionnaire.[30]

Genetic testing to confirm the diagnosis aids in management but is challenging to obtain in poor and under-resourced countries.

With advancements in medicine and technology, novel and more individualized treatment options will hopefully soon become available. Thus, diagnosis is mainly based on clinical features, especially in areas where genetic testing is not easily available.[32]

Although literature abounds on this disease, there is a paucity of knowledge about its oral manifestations and dental management.[25,5] This present case report assesses the oral health status of two siblings with Ichthyosis in a peri-urban community in the Central Region of Ghana.

## CASE REPORT

### DEMOGRAPHICS

A.H and K.H are siblings who presented with dermatological and oral conditions at a medical and dental outreach. They have additional seven siblings, three females and four males, who do not have this condition. Both parents are deceased.

They live a secluded life with their siblings in a self-owned compound house built of concrete but with a compound of bare ground in a peri-urban community in the Central Region of Ghana.

They are unemployed but occasionally help to gather farm produce.

Both have no formal education.

**MEDICAL HISTORY**

**A.H.**

34-year-old female born at home by a traditional birth attendant. At birth, she had a membrane covering and suffered blisters, scaly skin, and erosions in infancy.

She suffers from moderate hearing loss and difficulty in walking. She has a good memory but suffers difficulty carrying out daily tasks such as washing clothes.

Currently, she suffers from skin fissuring with associated pain, pruritis, ectropion, alopecia, and decreased ability to sweat.

**DENTAL HISTORY**

She has not visited a dentist before.

**FAMILY HISTORY**

She had no other positive family history of their condition except the sibling.

**EXTRA ORAL EXAMINATION**

She presented with generalized dry, flaky scales on the body: this affected the extensor surfaces, trunk, flanks, folds, lower leg,s, and wet areas. She also presented with hyper linear palms, skin fissuring, moderate alopecia, ectropion, keratitis, corneal opacities, skeletal dysplasia, diminutive external ears, and ataxia with moderate difficulty in walking and moderate hearing difficulty.

She had a moderate skeletal class II malocclusion with decreased LAFH. Her face was asymmetrical, and d had a tongue thrust on swallowing. She had incompetent lips with 50% of teeth showing at rest and an acute nasolabial angle.

**INTRA ORAL EXAMINATION**

Oral hygiene was assessed using the Oral Hygiene Index Simplified (Greene and Vermillion, 1964) to be 4.83 (Poor). The periodontal diagnosis was Generalized Periodontitis Stage IV grade B/ Generalized severe chronic periodontitis.

She was in the permanent stage of development with multiple missing teeth and a DMFT of 9. Caries assessment was done with ICDAS Caries Lesion Activity Assessment Criteria modified by Nyvad et al. (1999) and Ekstrand et al. (2008). A.H. had active approximal caries on the mesial aspect of 13 (ICDAS 5) and extensive coronal caries on 17 and 27 (ICDAS 6). She also had a lingually positioned tooth number 3 5. She had a class I molar relationship bilaterally.

**K. IS A 14-YEAR-OLD MALE** born at the polyclinic. At birth, he had a membrane covering and suffered blisters, scaly skin, and erosions in infancy. He suffers from moderate hearing loss and difficulty in walking. He has good memory sufferer difficulty in carrying out daily tasks such as washing clothes.

Currently, he suffers from skin fissuring with associated pain, pruritis, ectropion, alopecia, and decreased ability to sweat.

**DENTAL HISTORY**

He has not visited a dentist before.

**FAMILY HISTORY**

He has an older sister with a similar condition.

**EXTRA ORAL EXAMINATION**

He presented with generalized dry, flaky scales on the body, which appeared less severe than the older sister. It also affected the extensor surfaces, trunk, flanks, folds, lower legs, and wet areas. He also presented with hyper linear palms, skin fissuring, moderate alopecia, ectropion, keratitis, corneal opacities, skeletal dysplasia, diminutive external ears, and ataxia with moderate difficulty in walking, and moderate hearing difficulty.

He had a mild class II skeletal pattern with average vertical proportions. His face was symmetrical. He had competent lips with a 10% incisor show at rest. His swallowing pattern was lip to lip intraoral examination

The oral hygiene Index Simplified score was 3.3 (PoorThe periodontal diagnosis was Generalized Moderate Biofilm-induced Gingivitis. All the teeth were present in the mouth except the third molars. He had a DMFT score of 0. All tooth surfaces of K.H. were sound with no signs of active caries (ICDAS 0), although surfaces showed moderate plaque deposits. He had class II incisor relationship with class I canine and molar relationship bilaterally with overjet of 6mm and overbite of 90%. He had a flat curve of Spee, coincident midlines, and was in a group function.

**INVESTIGATIONS**

OPG and lateral cephalograms were taken for both siblings

**A.H.**

***Microphotographs published with the permission from the patient***

**Fig1 Extraoral photos of A.H. showing typical scaly skin, alopecia ectropian and constricture**

**Fig.2 Intraoral photos of A.H. shows severe active dental disease**

**Table 2- Debris Index Simplified for A.H**

| | Right Molar | | Anterior | | Left Molar | | Total |
|---|---|---|---|---|---|---|---|
| | 17 | 46 | 12 | 31 | 26 | 36 | |
| Labial | 3 | - | 3 | 3 | 3 | - | 12 |
| Lingual | - | 3 | - | - | - | 3 | 6 |

DI-S = (12+6)/6 = 3 implying poor oral hygiene

**Table 3- Calculus Index Simplified for A.H**

| | Right Molar | | Anterior | | Left Molar | | Total |
|---|---|---|---|---|---|---|---|
| | 17 | 46 | 12 | 31 | 26 | 36 | |
| Labial | 1 | - | 1 | 3 | 3 | - | 8 |
| Lingual | - | 1 | - | - | - | 2 | 3 |

CI-S= (8+3)/6= 1.83
OHI-S= DI-S + CI-S= 3+1.83= 4.83

**K.H**

***Microphotographs published with the permission from the patient***

Fig3 Extra oral photos of K.H showing typical scaly skin, alopecia ectropian and skin constricture

**Fig 4. Intra oral photos of K.H shows milder but active dental disease**

**Table 5- Debris Index Simplified**

| | Right Molar | | Anterior | | Left Molar | | Total |
|---|---|---|---|---|---|---|---|
| | 16 | 46 | 11 | 31 | 26 | 36 | |
| Labial | 2 | - | 1 | 3 | 2 | - | 8 |
| Lingual | - | 2 | - | - | - | 2 | 4 |

DI-S= (8+4)/6= 2

**Table 6- Calculus Index Simplified for K.H**

| | Right Molar | | Anterior | | Left Molar | | Total |
|---|---|---|---|---|---|---|---|
| | 16 | 46 | 11 | 31 | 26 | 36 | |
| Labial | 1 | - | 1 | 1 | 2 | - | 5 |
| Lingual | - | 2 | - | - | - | 1 | 3 |

CI-S= (5+3)/6= 1.33

OHI-S= DI-S+CI-S= 2+1.33= 4.33. An oral hygiene score of 4.33 indicate poor oral hygience which will need scaling and root planning and modification of oral habits

## DISCUSSION

Increasing severity of hyperkeratosis and presence of erythema negatively impact the quality of life in people with Ichthyosis, while better therapies and increased patient education may improve it.[28] A.H. and K.H. suffer from hearing impairment, ocular problems, mobility limitations, and the dermatological manifestation had severely affected their quality of life, as evidenced by their secluded lifestyle and inability to obtain formal education, and they are also unemployed. In addition, A.H. is 34 years, unmarried, and has never been in a relationship. Ichthyosis also hurts their health-related quality of life.[8]

There is very little information about the dental symptoms of Ichthyosis and how to treat them, with few authors drawing correlations between the two.[5,33] Patil[5] have suggested that over-retained teeth can cause the malocclusion observed in persons with Ichthyosis and delayed eruption of permanent teeth, obstructing normal jaw growth and development.

A.H. showed severe periodontal disease with multiple tooth loss, which may be attributed to years of oral neglect resulting in severe compromise of oral tissues.[39] K.H. also showed periodontal disease, though much milder. Dental findings in multiple case reports of Ichthyosis have included gingivitis, periodontitis, xerostomia, and hyperkeratotic plaques on the tongue.[24,33,] However, whether periodontal disease is a direct result of Ichthyosis, as occurs with certain syndromes,[36] or is a complication that arises out of neglect is yet to be established.

K.H. had no caries experience compared with A.H., who had some teeth showing extensive caries. Case reports in the literature about ichthyosis mention patients being prone to high-risk caries,[5] which is observed with A.H. A.H. had about three teeth showing carious lesions, which places him in a high-risk group. However, all parameters of a caries risk assessment need to be assessed to conclude. Both AH and K.H. may benefit from a diet chart.

Severe compromise of oral tissues precluded A.H from benefiting significantly from orthodontic care.

K.H had an IOTN score of 4a. This implied that there is a moderate need for orthodontic care. However, orthodontic care had to be delayed to allow for comprehensive management of oral and perioral tissues.

Extreme care needs to be taken when rendering dental treatment in patients with Ichthyosis since manipulation of their perioral skin tends to worsen the fissuring usually found in those areas. Treatment for these siblings, therefore, mainly focused on a preventive approach.

They were given skin-softening emollients, particularly after bathing, and were referred for periodontology and dermatology consult.

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## CONCLUSION

The impact of congenital Ichthyosis extends beyond the skin and affects oral health and quality of life. Early diagnosis at birth and early intervention can help prevent the severe oral manifestation of this disease.

## RECOMMENDATION

Need for awareness creation about the disease to make family and social support available to those afflicted.
Establish guidelines and protocols to improve the care of those afflicted through education of health workers on the diagnosis and management of the condition.

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