Multiple Bony Deformities and Short Stature in a Child with Lamellar Ichthyosis, What more can we do? A Case Report

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Learning Point of the Article:
Surgical management in the form of osteotomy is an acceptable management for patients with ichthyosis and severe bony deformities, it has the potential to drastically improve the quality of life of the patient.

Abstract

Introduction: Ichthyosis is a group of disorders typically characterized by the accumulation of large scales over the skin. Mild bony deformities due to Vitamin D deficiency are commonly associated with this group of disorders which can be successfully treated with conventional Vitamin D supplementation. Severe multiple bony deformities requiring surgical correction are rarely reported and may be associated with various other disorders.

Case Report: We report a case of a 15-year-old male with ichthyosis, short stature, and progressive multiple bony deformities since birth. The child was started on Vitamin D3 supplementation. Once biochemical parameters improved he underwent multiple corrective osteotomies in the bilateral tibia and right femur to improve gait mechanics. Our main concerns while managing the patient were regarding wound healing, secondary infection due to extensive scaling and healing at the osteotomy site. On follow-up we noted good healing at the osteotomy site without any surgical site infection or skin complications as well as improvement in gait mechanics and cosmesis.

Conclusion: Severe bony deformities due to Vitamin D deficient are rare in ichthyosis and other syndromic causes should be ruled out. Surgical management can be beneficial in improving quality of life and gait biomechanics.

Keywords: Lamellar ichthyosis, osteotomy, genetic testing, Vitamin D deficiency.
On physical examination, the child had a height of 102 cm and weight of 24 kg, both these parameters were below the third percentile of the Indian Academy of Pediatrics chart for height and weight for boys. Cognitive and speech functions were intact.

Examination revealed brown scales all over the body, including the scalp, flexural surfaces, and external auditory meatus. The scales were large in size and odorless, adherent to the skin. Thin hair was noted on the scalp with entangled fine scales. Abnormal dentition and missing teeth were also noted. There was fixed kyphoscoliosis with convexity to the right side. The right forearm showed bowing and the right upper limb appeared "S" shaped. Pronation and supination were severely restricted on the right side due to old malunited fracture in the forearm. There was anterior bowing of the right femur and lateral bowing of the left femur as well as anterolateral bowing of bilateral tibia and lateral dislocation of the right patella (Fig. 1). Plain radiographs showed multiple bony deformities in the lower limbs (Fig. 2). Skin biopsy confirmed the diagnosis of lamellar ichthyosis. Oral Vitamin D supplementation was started at 5000 IU daily dose along with oral calcium supplementation [6], oral tretinoin and topical emollients were given for skin disease.

Once biochemical parameters were normalized, he was planned for a staged correction of all lower limb deformities to prevent postoperative infection due to poor skin condition. We first performed a right-sided distal tibia corrective osteotomy. As there were no immediate signs of surgical site infection with good healing of surgical wound, after 1 week a right-sided supracondylar femoral osteotomy was done. Following this procedure, the patella spontaneously relocated. A third osteotomy procedure was performed after 3 weeks for correction of the left distal tibia. We followed standard pre and post-operative protocol for all procedures. All surgical sites healed without any complications and at 6 weeks follow-up assisted walking was started as the osteotomy sites were uniting on radiograph (Fig. 3).

We continued Vitamin D supplementation on a monthly basis. At 18 months follow-up, we observed improved gait mechanics, decreased pain and fatigue while mobilizing. His scaling and cosmetic appearance also improved and biochemical parameters were within normal limits (Fig. 4).

**Discussion**

Vitamin D deficiency is common in ichthyosis and presents with mild or no deformities. Various mechanisms have been proposed
for Vitamin deficiency in these patients (Table 1) [1, 7].

A small percentage of ichthyosis children manifest with severe deformities and short stature. These severe deformities can be either due to prolonged untreated nutritional rickets or due to associated syndromes (Table 2).

Some authors have proposed association of some forms of ichthyosis and Vitamin D resistant rickets following large contiguous deletions of genes on the same chromosome. Deletions of 12q13 coding for keratin 1 can involve 12q13-14 coding for Vitamin D receptor causing a cellular Vitamin D resistance [8]. Similarly, deletion around Xp22.31 coding for steroid sulfatase can involve Xp22.1 coding for Phosphate Endopeptidase Homolog X-linked leading to hypophosphatemic rickets which is also Vitamin D resistant [9].

Based on this hypothesis we should explore the use of genetic testing in such patients, wherever possible. Testing if done before the growth spurt may enable us to determine the sensitivity of rickets to conventional Vitamin D supplementation and may prevent the development of short stature and disabling deformities by the initiation of appropriate treatment. In cases where resources are sparse a close monitoring of biochemical parameters to Vitamin D therapy and timely referral should be considered.

### Conclusion

Vitamin D deficiency in ichthyosis is a common phenomenon but rarely presents with severe deformities. In cases of severe deformities with ichthyosis, one should initiate appropriate treatment and attempt to rule out syndromic causes. Surgical therapy including osteotomy can be considered and has yielded good results for this child leading to an improvement in quality of life.

### Clinical Message

Vitamin D deficiency in ichthyosis rarely causes severe bony deformities. In patients presenting with severe bony deformities, syndromic involvement should be ruled out. Surgical management in the form of osteotomy is an acceptable management and improves the quality of life.

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