SIBLINGS WITH SHORT STATURE AND BRITTLE HAIR- MYSTERY UNVEILED

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PRESENTATION OF CASE

Siblings (6 years and 9 years old/ female) born to 3rd degree consanguineously married couple from a village in Telangana presented to us with complaints of dry scaling of skin present all over the body since birth, short stature, brittle hair and delayed development.

Tay syndrome or trichothiodystrophy (TTD) is a very rare autosomal recessive disorder characterised by congenital ichthyosiform erythroderma and brittle sulphur-deficient hair. Other features include short stature, intellectual impairment, photosensitivity and multiple defects affecting organs mainly derived from neuroectoderm. Till date only less than 110 cases of Tay syndrome have been reported in literature worldwide and there are no cases of siblings with Tay syndrome reported in India till date. We present here a case of siblings from a South Indian family with Tay syndrome.

CLINICAL FEATURES

	Child - A	Child - B
Age/Sex	9 yrs./ Female	6 Years/ Female
Weight	15 kgs Expected Weight - 32 kgs	11 kgs Expected Weight - 21 kgs
Height (Short Stature)	108 cm Expected Height - 132 cm	95 cm Expected Height - 114 cm
Bone Age	3 years	2 years
Skin Findings	Ichthyosiform Erythroderma	Ichthyosiform Erythroderma
Hair	Straight, Coarse, Unruly and Brittle	Scarce and Brittle
IQ (Stanford- Binet Scale)	78	70
Developmental Age	7 years	4 years

Birth History

All 3 siblings were born at home and was delivered by a trained dhai. First sibling had ichthyosis at birth and died at day 5 of life at the hospital in view of sepsis. 2nd and 3rd sibling was taken to the hospital immediately after birth, as they too had ichthyosis at birth and was discharged at Day 10 of life with topical emollients.

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Family Pedigree Chart

DIFFERENTIAL DIAGNOSES

With the above mentioned Clinical Features, 3 differential Diagnoses was Considered-

1. Netherton syndrome- characterised by ichthyosiform erythroderma, trichorrhexis invaginata and atopic diathesis.
2. Chanarin-Dorfman syndrome- characterised by congenital ichthyosis, lipid vacuoles in leucocytes and hepatomegaly.
3. Tay syndrome or trichothiodystrophy- congenital ichthyosiform erythroderma, brittle sulphur deficient hair, short stature and intellectual impairment.

CLINICAL DIAGNOSIS

Since both the siblings had intellectual impairment and short stature, the diagnosis of Tay syndrome was considered.

FINAL DIAGNOSIS

- **Hair Shaft Examination under Polarising Microscopy**
Showed diagnostic alternating light and dark banding pattern called Tiger tail banding suggestive of Tay syndrome.
- **Biochemical Examination of Hair**
Chromatography of hair amino acids done revealed decreased sulphur containing amino acids, cysteine, proline and threonine thereby confirming the diagnosis.

Figure A. Showing "Tiger Tail Banding" seen under Polarising Microscope

Figure B. Picture of Siblings with Tay Syndrome (Short Stature, Ichthyosis, Brittle and Sparse Hair)

DISCUSSION OF MANAGEMENT

Tay syndrome or Trichothiodystrophy is a very rare disease with autosomal recessive inheritance characterised by brittle, sulphur deficient hair.^{1,2} It is a multisystem disorder with a varied spectrum of clinical features. The common clinical features are brittle hair or hair shaft abnormalities (96%), intellectual impairment (86%), short stature (73%) and ichthyosis (65%).³ The name reflects the brittle, sulphur deficient hair seen in all TTD patients (From Greek- tricho- meaning hair; -thio-, sulphur; -dys-, faulty; -trophy, nourishment). Various acronyms have been used to describe the clinical features of this syndrome. PIBIDS,⁴ IBIDS^{5,6} and BIDS⁷ describe the features of TTD: Photosensitivity, Ichthyosis, Brittle hair, Intellectual impairment, Decreased

fertility and Short stature. In 1985, defective DNA excision repair in ultraviolet (UV) exposed lymphocytes by Van Neste⁸ from a TTD patient. Recently, in non-photosensitive patients TTDN1 gene with unknown function was identified.⁸ Till date, four genes causing TTD have been identified: XPD, XPB, TTDA and TTDN1.³

Diagnosis can be made by demonstration of Tiger tail banding in the hair shaft under polarising microscopy, which is diagnostic of and is seen exclusively in Tay syndrome.³ Hair shaft examination under scanning electron microscope reveals flat hair with trichoschisis and chromatography hair amino acids show decreased sulphur containing amino acids.

No specific treatment for Tay syndrome is available. Management is mainly symptomatic. Family psychological and social support is needed. Children should be advised to avoid sunlight exposure and aggressive combing or hair dressing due to weakness of hair shaft.

CONCLUSION

Tay syndrome should be considered as a differential diagnosis in any child with congenital ichthyosis and hair abnormalities. A simple hair shaft examination under polarised microscopy will help to clinch the diagnosis. We report this case for its rarity.

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