SJOGREN LARSSON SYNDROME
(Two case reports)

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Summary

Two cases showing essential features of Sjogren Larsson Syndrome namely congenital ichthyosiform erythrodermia, spastic disorders, mental retardation and speech defect are presented. There was no history of consanguinity in parents. Macular dystrophy was present in one case. There was no history of convulsions in any of them. Bony age was almost consistent with their chronological ages. Aminoaciduria was absent in both cases. There was no dental or osseous dysplasia in either case. Hyperteliorism, defective sweating or dermatoglyphy were not present in our cases. Additional findings in one case was presence of Diabetes Mellitus in the absence of any family history of diabetes.

Tuberculosis in some form or other was present in both cases. Although this finding is not of much importance in our country where tuberculosis is commonly prevalent, it cannot be ignored that both patients are non-ambulatory and there is no case of tuberculosis among family members or near contacts.

This syndrome first reported by Sjogren & Larsson[1] in 1957 consist essentially of a triad of congenital disorders like ichthyosiform erythrodermia, spastic disorders and mental deficiency of a severe degree. They originally reported 28 cases. Out of these 28 cases, 25 were born in the county of vastorbotten in North Sweden. In this community consanguinity is common (8 out of 13 parents). Although this syndrome was first described by Sjorgen and Larsson[1], Paradocastelle and Fazz[2] had reported such cases under the name of 'Ichthyosis littles disease' as early as 1932.

Later, Witkop and Henry[3] reported 14 cases in another isolated community of inbred people called Haliwas of Halifax and Warren county in North Carolina. This community is a mixture of Caucasian, Negro and Amerindian Stocks. Coincidentally this group of cases was also associated with another ectodermal defect called hereditary benign intraepithelial dyskeratosis. This is transmitted by a dominant gene and produces white plaques around the mouth and eyes.

Upto 1963 there were 53 reports of Sjogren Larsson syndrome[4]. In 1965 six new and four old cases were reported by Hiezer and Read[5]. Salmonovitz and Porter[6] reported another 3 cases in 1967. Later Richard[7] in an insertion in Lancet had stated that upto 1965 only 7 cases of this syndrome were reported from Britain and he knew another 2 unreported cases amongst the Britishers. There was another report of 3 cases of this syndrome by Rosano et al[8] from the University of Rome.

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Many additional clinical features have been added to this syndrome after the earlier description of this disease. Among these are developmental anomaly of globus pallidus and frontal lobes[9], degenerative pigmented lesions in macula[10], defective sweating, hyperteliorism and dermatologlyphic abnormalities like decreased frequency of whorls and interdigital patterns, low ridge count, increased incidence of distal and double axial triradii hypothenar patterns and modified simian lines.

The severity of the abnormalities varies widely among affected individuals. Although generally considered an autosomal recessive trait without sex preponderance[11], Salmonovitz[6] suggests presence of a heterozygous or carrier state. Most of the reported cases are from Scandinavia, Europe, near East and Americas predominantly in Caucasians.

Case Reports

Two cases in the same family a brother and sister presenting the salient clinical features of Sjogren-Larsson syndrome are reported here. One of the cases had Diabetes and both of them had tuberculosis which is not hitherto reported. (Fig. Page No. 52.)

Case 1, was a 26 years old female resident of Delhi, born to non-consanguine parents of Hindu Sikh background. She was 2nd of 6 children all of whom were born normally at full term. Her youngest brother, the 6th child in the family is the 2nd case reported here.

Patient 1 was a F. T. N. D. after an uneventful pregnancy in a Hospital at Rawalpindi. Her birth weight is not known to the parents. At birth the parents noted generalised redness and peeling of the skin. Mile stones were delayed. She started sitting at the age of 9 months. She could stand with support at the age of 1 year. At about 2½ years of age she could walk a little. Only at this stage the parents noticed that the child walked on toes and the heels never touched the ground. Gradually the foot drop and walking inability of the patient worsened. At present she is unable to walk. She is mentally retarded. There is no history of convulsions at any stage of development.

On examination there is generalised ichthyosiform erythrodermia of moderate degree. The patient is mentally retarded with a grinning facies. Mental age as estimated by the performance on proteus maze test was 4 years. Moderate correlation of test result was obtained with performance on seguin board. Test performance was characterised by poor understanding of instruction and task, slow execution with very defective motor control. The speech was of lalling type with some ideoglossia. Patient and her similarly affected brother could converse freely with each other while it was difficult for others to understand them.

Cranial nerves were normal, Upper limbs showed no wasting but power was less than normal and equal on both sides. There was no hypotonia. Reflexes were exaggerated. Sensations were normal. Lower limbs showed mild degree of flexion deformity of knees, spasticity of lower limbs, foot drop with inversion of feet, positive Babinski, exaggerated ankle and knee jerks and scissors gait. Sensations were normal. Respiratory system showed bilateral pleural effusion. C. V. S. and G.I. system were clinically normal. There was no disturbance in sweating or dental dysplasia. Interpupillary distance was 5.3 cm.

Investigations

Hb. 14 gms., T.L.C. 9000 per cu. mm. D. L. C. : P 70, L 28, E 2., E.S.R. 23 mm., S.T.S.—Negative., serum protein 7.04 gms%, serum albumin 3.2 gms%. serum globulin 3.84 gms%. Sodiumserum

130 mEq/L, serum potassium 3.2mEqp/L. serum cholestrol 210 mg. Blood sugar-fasting 133 mg. PC 210 mg., Glucose tolerence test-Diabetic lag curve, Blood urea 29 mg., L. E. cell phenomena negative, Urine-N. A. D. Urinary aminocide—negative, Stool—N. A. D, Fundus—both maculae dull, E. E. G. failed to show any abnormal electrical activity. Pneumo—encephalogram-evidence of cortical atrophy without any internal hydrocephalus.

CSF: Total proteins 35gm%, Globulins-negative, Chlorides 720gm% Sugar 11 mg%, Cytology-clear fluid, no deposit, no coagulum and no cells. X-ray chest, Bilateral pleural effusion. X-ray skull N.A.D. Bony age about 20 years (chronological age 26 years).

Histopathology: Consistent with congenital non-bullous ichthyosiform erythrodermia.

Case Two

A 14 years old male was younger brother of case No. 1. The history was very similar to that of his sister. About 2 years prior to the hospital visit, patient developed glands in axilla. The biopsy of one of these was reported as tubercular lymphadenitis. Patient was put on antituberculous treatment. There were extensive pyodermic lesions on the scalp with partial cicatrical alopecia because of active folliculitis decalvans. There was moderate degree of generalised ichthyosiform erythrodermia. The patient was mentally retarded. His mental age as estimated by performance on proteus maze test was $4\frac{1}{2}$ years. Moderate correlation of the test result was obtained with performance on seguin board. The performance was characterised by poor understanding of instructions. There was hurried execution with defective motor control. Overall performance and understanding was better in comparison to that of his elder sister. There was lalling speech with some ideoglossia.

Cranial nerves were normal. Upper limbs showed no wasting of muscles. Power was more or less normal. Lower limbs showed flexion deformity of both the knees, foot drop with inversion and adduction of feet. Positive Babinski and exaggerated knee and ankle jerks. Ankle clonus + Scissor gait, + sensation were normal. Respiratory system and C.V.S. were normal. Viscera could not be properly palpated due to spasticity of the muscles.

Investigations

Hb. 11 gms%. T.L.C. 10,000 per cu. mm. D.L.C.: P 75 L 25 E.S.R. 12 mm. Blood S.T.S. negative. Serum protein-7 gms%. Serum alb. 4.23 gms%. Serum glob. 2.68 gms%. Serum sodium—137 mEq/L. Serum cholestrol 150 mg. Fasting blood sugar 91 mg. G. T. T. within normal limit. Blood urea 31mg. L. E. Cell phenomena negative.

Fundus :—Bilateral macular dystrophy.

E.E.G. failed to show any abnormal electrical activity.

Pneumoencephalogram: Evidence of cortical atrophy without any internal hydrocephalus.

C.S.F.: Total Proteins 25mg% Globulins negative. Chloride 700 mg%. Cytology—clear colourless fluid with no deposit or coagulum. R.B.C. + Occasional leucocytes.

Urine: N. A. D. Urinary amino acids: negative. Stools: N.A.D.

Lymphnode biopsy—tubercular lymphadenitis. Skin biopsy–consistent with non bullous ichthyosiform erythrodermia.

X-Ray chest: N. A. D. X-Ray skull: N.A.D. X-Ray wrist: N.A.D. X-Rays of Elbows: epiphyseal centres for trochlea and epicondyle have appeared and fused with capitulum suggesting age

of 12 years. But the centres for medial epicondyle head of radius and olecranon have not fused suggesting that the bony age is less than 14 years. Thus the evaluation of the bony age comes to about 12 – 14 years.

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