

families, both chromosome 9q34 and 17p11.2 JS loci were excluded in 26 JS families. They have also searched and excluded the candidate genes EN1, EN2, FGF8, and BARHL1 from a direct pathogenetic role in JS(5). Genetic studies and many variable phenotypes of JS suggest that there is heterogeneity in genetic basis of JS.

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## X-Linked Adrenoleuko-dystrophy Presenting as Addison Disease

X-linked adrenoleukodystrophy (X-ALD) is a group of peroxisomal disorders characterized by an impaired beta-oxidation of very long chain fatty acids. This defect results in an extremely variable phenotype including presymptomatic form, isolated adrenal insufficiency and cerebral disease. An incidence of about 1: 10000 male subjects has been reported(1).

Two brothers (8 years and 5 years old), born of a non-consanguineous marriage were brought with complaints of progressively increasing pigmentation and darkening of

skin, starting from face since the age of 4 years. There was history of recurrent episodes of loose motions, vomiting and fever after the age of 4 years in the elder sibling for which he was hospitalized in a state of shock. Family history, perinatal history and developmental history were non-contributory. General and systemic examinations were unremarkable except for the generalized hyperpigmentation of the skin. Black colored spots were also noted on the tongue and oral mucosa.

Investigations revealed a normal hemogram. Basal serum cortisol levels were low [1.82 µg/dL and 2.8 µg/dL (normal= 9-25 µg/dL)] and response to ACTH was subnormal [5.86 µg/dL and 6.7µg/dL after ACTH stimulation test.]. Serum electrolytes, albumin,

alkaline phosphatase, calcium and phosphorus levels were normal. Thyroid function tests and antimicrobial antibodies were negative. All these investigations suggested a primary hypofunction of adrenal glands. An abdominal ultrasound and chest X-ray were unrevealing.

The siblings were further investigated for a possibility of adrenoleukodystrophy (ALD) by estimating plasma levels of very long chain fatty acids (VLCFAs). The pattern of increase in VLCFAs levels was consistent with X-ALD (*Table I*). An MRI scan of brain was normal. The sibs are currently receiving replacement doses of corticosteroids with an adequate stress cover and are under continuous follow up. They have not developed any neurological complaints till now so they may be rare entities of "Addison only" variant of ALD.

Majority of cases of primary adrenal insufficiency have autoimmune or tubercular etiologies but X-ALD is now being recognized as not an uncommon cause of adrenal insufficiency in males, especially in children and young adults. Many of these patients who have clinical features of only Addison disease at the time of presentation, go on developing neurological manifestations of X-ALD in future, but this interim period is

highly variable and may extend up to 32 years(2-5).

Although establishing X-ALD as a cause of adrenal insufficiency doesn't affect the endocrinal management of the patient, it opens the possibility of therapy before the onset of overt neurological disease, since both bone marrow transplantation and Lorenzo oil are beneficial only at an early stage of disease(2,5). It also allows screening of other male members and relatives who may be at risk of developing Addison disease or neurological complications. Early diagnosis also brings the possibility of genetic counseling; carrier detection and antenatal diagnosis and thus has the potential for radically reducing the incidence of this devastating disease.

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**TABLE I**—*Levels of VLCFAs in Two Siblings with X-ALD.*

Very Long Chain Fatty Acid	Patient (µg/mL)	Normal controls (µg/mL)	X-ALD Hemizygote (µg/mL)	X-ALD Heterozygote (µg/mL)
C 26: 0	Sib A: 1.47 Sib B: 2.04	0.24 ± 0.14	1.30 ± 0.45	0.68 ± 0.29
C 24/22	Sib A: 1.98 Sib B: 2.14	0.78 ± 0.1	1.71 ± 0.23	1.30 ± 0.19
C26/22	Sib A: 0.13 Sib B: 0.16	0.01 ± 0.003	0.07 ± 0.03	0.04 ± 0.02

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## **Influence of Child Care Practices on Prevalence of Diarrheal Diseases**

Diarrhea is a common illness and a leading cause of malnutrition and death in under five children of developing countries(1). Child-care practices including personal and domestic hygiene play an important role in diarrhea prevention.

A cross-sectional, community based, observational study was undertaken in a suburban area of Kolkata, to study the role of child care personnel in occurrence of diarrhea. Municipal records showed 40% diarrhea prevalence among under fives in the study area during two weeks time. Based on this a sample size of 300 was estimated, and the subjects were selected by systematic random sampling technique.

The study population showed a diarrhea incidence of 31.67% in a two week recall period, which was much higher than the figure reported in children under three years of age, by NFHS 2 in West Bengal(2). The incidence of diarrhea was inversely proportional to the increase in educational level of the mother,

this difference being strongly significant ( $P < 0.005$ ). Incidence of diarrhea increased with the increase of income levels of their families ( $P < 0.05$ ). Diarrhea cases were significantly more ( $P < 0.001$ ) among under five children of working mothers. Children who were looked after by their mothers showed least incidence of diarrhea, followed closely by those looked after by trained outside nurse. Children who were cared for by untrained outsider suffered the most. This trend was strongly significant ( $P < 0.005$ ). Exclusively, breast-fed infants showed least incidence of diarrhea, followed by spoon fed and bottle fed ones. Those who were fed by hand showed a very high diarrhea incidence, which was significantly more than that among exclusively breast fed babies ( $P < 0.01$ ).

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