Rud’s syndrome

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ABSTRACT

Rud’s syndrome is a rare autosomal recessive hereditary disorder characterized by congenital ichthyosis, epilepsy, dwarfism, sexual infantilism, polyneuritis, and macrocytic anemia. We report here an interesting case of this disorder in an 18-year-old girl for its rarity and academic interest.

Key words: Congenital ichthyosis, Rud’s syndrome, sexual infantilism

INTRODUCTION

Rud’s syndrome is a rare genodermatosis characterized by congenital ichthyosis, epilepsy, mental retardation, dwarfism, sexual infantilism, and infrequently, retinitis pigmentosa. Subsequent to its initial description in a 22-year-old Danish patient by Rud in 1927, around 55 cases have been reported world-over, with none from India. Herein we report a female patient form India with Rud’s syndrome.

CASE REPORT

An 18-year-old female presented to our outpatient clinic with complaints of dry, scaly, skin all over the body that started when she was six months old. There was no preceding history of convulsions or abnormalities pertaining to vision, hearing, teeth, and bones. She had not attained menarche and was a school dropout with a low IQ. She was born of a full-term normal vaginal delivery to non-consanguineous parents. None of her family members had similar problems.

Her clinical examination revealed ichthyotic skin all over the body, but more prominently on the upper and lower limbs (Figure 1). Palmoplantar keratoderma with hyperlinearity of palms was present. Her stature was short compared to her chronological age, (4’5” height and weight 31 kg) and breasts were small with poor development of areola and nipple (Figure 2), and her external genitalia were infantile (Figure 3). Hair over her axilla and pubic region was strikingly absent, although it was normal on the scalp.

Routine hematologic and biochemical investigations were within normal limits, except that liver function tests showed mild elevation of alkaline phosphatase (212 IU/L) and the serum calcium levels were low (6.8 mg/dl). Hormonal analysis was normal except for elevation of follicular stimulating hormone (FSH - 126.66 IU/L) and luteinizing hormone (LH - 32.47 U/L). Ultrasonography of the abdomen and pelvis showed hypoplastic, infantile uterus (1.6 × 0.4 cm) and ovaries (right ovary - 18 × 7 mm, left ovary - 14 × 7 mm), small right kidney and bulky left kidney with dilated pelvicalyceal system. However, her renal function tests were within normal limits and she had no clinical evidence of the renal impairment. Further evaluation of kidneys such as renal biopsy and immunoflorescence were not undertaken due to lack of facilities. Karyotyping showed female character and her buccal mucosa was positive for X-chromatin.

A 4 mm punch biopsy obtained from the skin over her right leg revealed a histological diagnosis of ichthyosis showing features of hyperkeratosis, thinning of suprapapillary epidermal plates, slight elongation of club shaped rete ridges with non-specific mononuclear inflammatory cell infiltrate in the dermis (Figure 4). Ophthalmological examination revealed no abnormality.

Based on her history, clinical and the laboratory investigations, she was diagnosed as a case of Rud’s syndrome. After explaining about the nature of her skin condition, she was advised emollients and keratolytics for dry and scaly skin and calcium supplements in the form of...
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oral calciferol tablets for low calcium levels. Further, she was referred to the gynecologist for specialized care regarding her delayed menstruation and sexual infantilism.

DISCUSSION

The pattern of inheritance and the nature of primary genetic defect in Rud’s syndrome is not entirely known,[4] although an autosomal recessive and X-linked inheritance pattern have been proposed[5] and the deletion of the steroid sulfatase gene leading to the deficiency of that enzyme has been considered as the basic abnormality.

Rud’s syndrome was considered in the past as a genetically heterogeneous, but distinct clinical entity. Heterogeneity of this disorder is suggested by differences in the clinical features, histological and endocrinological findings, steroid sulfatase activity, and modes of inheritance.[6] However, the existence of such a syndrome has been doubted recently based on the new developments known in the molecular biology of ichthyosis.[2,3]

Our patient has been diagnosed as a case of Rud’s syndrome based on typical history and presence of characteristic features such as low intelligence, congenital ichthyosis, short stature, sexual infantilism, and hypogonadism.

Several abnormalities including dermatologic, neurologic, endocrinologic, ophthalmologic and musculoskeletal have been reported with Rud’s syndrome.[7] The skin changes vary from simple dryness to severe ichthyosis. Eye changes such as cataract, ptosis, nystagmus, strabismus, retinitis pigmentosa have been reported. Neurological abnormalities are mental retardation, epilepsy, psychomotor retardation, personality disorders, polyneuropathy, cranial dysmorphisms, and abnormal electroencephalograms. Nerve deafness, teeth abnormalities, alopecia, pseudoacanthosis
nigricans can occur. Sexual infantilism can be diagnosed only after puberty.

Molecular analysis of the Rud’s syndrome is essential for prenatal diagnostics and proper genetic counseling. Treatment of Rud’s syndrome includes a multidisciplinary approach including dermatologic, endocrinologic, ophthalmologic, auditory, and gynecologic specialties depending upon the manifestations of the disease. Dermatologic management includes local application of emollients for ichthyosis such as soft paraffin, keratolytics, topical retinoids, and vitamin D3 analogues. Endocrinological and gynecological referral is required for the management of infantile sexual organs and delayed menstruation.

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