A case of recessive X linked ichthyosis presenting as a contiguous gene syndrome

K L P D Seneviwickrama[1], N D W Dissanayake[2]

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Abstract

Ichthyoses are common in pediatric dermatology practice. Majority of them are congenital ichthyoses. Ichthyoses may present as an isolated finding or as a part of a syndrome. Here we report a case of recessive X-linked ichthyosis (RXLI) in a three year old boy who also has Kallmann syndrome (KS), mental retardation and autism.

Introduction

Recessive X-linked ichthyosis, a deficiency of enzyme steroid sulphatase (STS) is a relatively common disorder. It is due to a deletion of the STS gene. Frequency of the disease ranged from 1 in 2,000 to 10,000 males[1]. RXLI commonly presents within the first weeks of life as exaggerated desquamation. Over the next few months, large brown, geometrically shaped and tightly adherent scales became prominent. It is symmetrically distributed over the torso and extremities and most pronounced over the extensor surfaces. A number of extracutaneous abnormalities have been reported in patients with RXLI[2]. Many of these abnormalities have been shown to be part of contiguous gene syndrome. Concomitant deletion of neighboring loci results in contiguous gene syndrome[3]. Herein we report a case of RXLI in a three year old boy who also had KS, mental retardation and autism.

Case report

A three year old boy presented with dry skin. He is the second child born to non consanguineous parents. He was born at term by a forcep delivery indicated by prolonged labour and fetal distress. His birth weight was 2.350 kg. At birth he had normal skin. Neonatal examination revealed bilateral undescended testes. Other systems were normal. He was managed at intensive care unit for neonatal meningitis. By day seven he developed dry skin. Later on he developed generalized dark brown polygonal scales (Figure 1, 2). His eyes and ENT examination were normal. His ultra sound scan of brain was normal. But his urological ultra sound scan showed normal left kidney and absent right kidney. Tc-99 DMSA scan failed to visualize ectopic renal tissue. He underwent bilateral orchidopexy at the age of two years (Figure 3). On examination, his skin showed typical features of RXLI. His development was globally delayed. He had bimanual synkinesia of hands (mirror images of hand movements) (Figure 1). Considering the unilateral renal agenesis, bilateral undescended testes and bimanual synkinesia clinical diagnosis of KS was made. He was not mentally sound to establish anosmia which is a part of KS. He was referred to a pediatric endocrinologist for the confirmation of KS. It was planned to do hormonal assay once he is nine years of age. He was put on topical emollients and mild keratolytics for his ichthyosis. He might need hormonal replacement after hormonal assay in future. Autistic behavior was looked into by the pediatric psychiatrist.

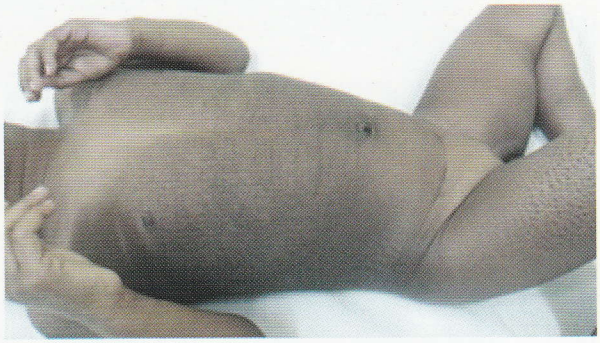

Figure 1.

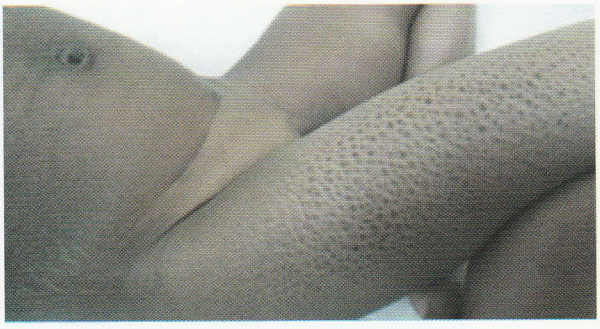

Figure 2.

[1]*Consultant Dermatologist, Base Hospital (Teaching), Gampola, Sirimawo Bandaranayake Specialized Children's Hospital, Peradeniya,* [2]*Medical Officer in Clinical Genetics, Sirimawo Bandaranayake Specialized Children's Hospital, Peradeniya, Sri Lanka.*

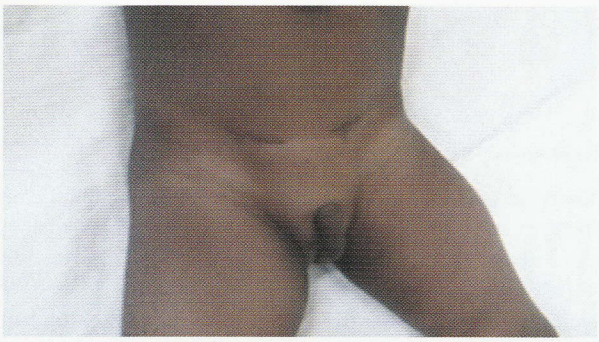

Figure 3.

Discussion

The majority of cases of RXLI are caused by complete deletion of the steroid sulphatase (STS) gene on chromosome Xp 22.3. Other cases result from partial deletion or point mutation of STS gene. Deletion or mutation of STS gene causes complete loss of STS enzyme activity. In 5-8% of cases, the deletion includes genes adjacent to STS gene resulting in a number of contagious gene syndromes[1]. The most commonly associated disorder is Kallmann syndrome. KS is a rare disorder characterized by hypogonadrotropic hypogonadism and anosmia. It is due to deletion of KALI gene. Other features of KS include renal agenesis, bimanual synkinesia, cryptorchidism, pes cavus, high arched palate and cerebellar ataxia[4]. Deletion of genes adjacent to STS gene results in X-linked mental retardation, autism, short stature and X-linked chondrodysplasia punctata.

The index child has features of RXLI, KS, mental retardation and autism. Ichthyosis is a relatively common problem in dermatological practice. The reflection of a rare association in a common presentation is highlighted in this case.

References

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