CASE 4

Patient: A 6-year-old girl from Pathumthani

Chief Complaint: Spastic diplegia for 5 years

Present Illness: The patient was referred for the problem of spastic diplegia since 1-year-old. At the age of 6-month-old, she developed generalized pruritus and fine white scales. The scales became more prominent and darker in color with age. She also had globally delayed development and articulation disorder. She is now studying in Kindergarten 3.

Past History: She is the first child born with normal labor. She had no seizure.

Family History: There was no family history of cutaneous diseases or consanguinity.

Dermatological Examination (Figure 4.1-4):
Generalized thick lamellar scales, palmoplantar keratoderma, scales are prominent on flexure area, nape of neck and relative spare on her face.

Physical Examination:
BW 16 kg (P10), Height 110 cm (P25)
HEENT: no pale conjunctiva, anicteric sclera, no lymphadenopathy
Heart: normal S1, S2, no murmur
Lungs: clear on both sides
Abdomen: soft, no distension, no hepatosplenomegaly
Extremities: no edema
Neuro: Motor power at least grade IV, spastic tone, DTR 3+ all, Barbinski's sign and clonus present both sides

Eye Examination (Figure 4.5)**:** Glistening white dots in the macula of the retina

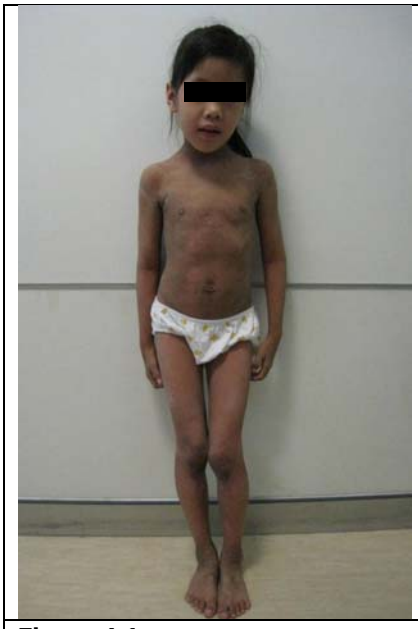

Figure 4.1

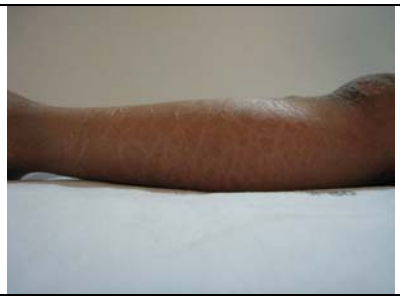

Figure 4.3

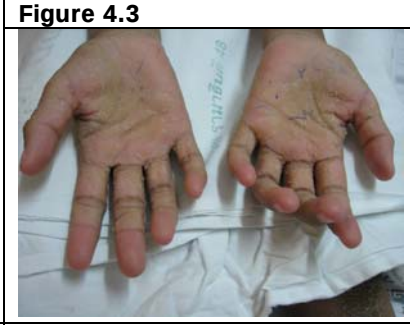

Figure 4.4

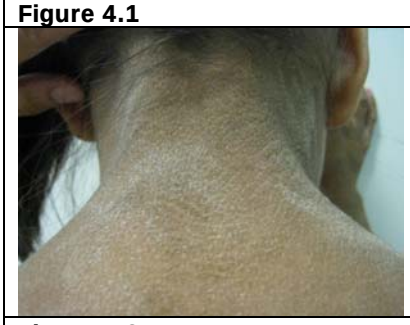

Figure 4.2

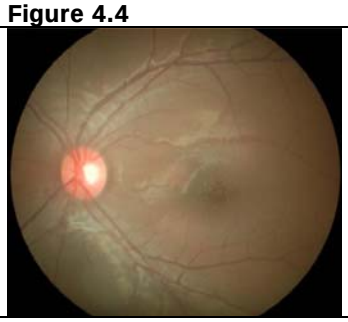

Figure 4.5

MRI and MRS of the brain: Patchy hypersignal T2/FLAIR change along periventricular white matter and bilateral centrum semiovale, showing slight increase of the choline and small lipid peaks.

Diagnosis: Sjögren-Larsson syndrome

Presenter: Yuvaluck Thammagasorn

Consultant: Amornsri Chunharas

Treatment: Physiotherapy
Oral antihistamine
Emollients

Discussion:

Sjögren-Larsson syndrome (SLS) is an autosomal recessive neurocutaneous disorder characterized by the clinical triad of congenital ichthyosis, spastic di- or quadriplegia, and mental retardation[1,3,7].

The cause is recessive mutations in the fatty aldehyde dehydrogenase gene (FALDH) on the short arm of chromosome 17 (17p11.2), which result in deficient enzyme activity[5]. This enzyme catalyzes the oxidation of long-chain aliphatic aldehydes to fatty acids, a pathway that is important for the synthesis of epidermal lipids as well as the catabolism of ether phospholipids and sphingolipids in the brain. The measurement of enzyme activity in cultured fibroblasts or leukocytes is a specific and reliable diagnostic test[2].

The clinical presentation of collodion baby is rarely seen in SLS. It usually presents at birth with varying degrees of erythema and ichthyosis. Diagnosis of SLS is often delayed until the onset of neurological symptoms and/or appearance of perifoveal glistening white dots (pathognomonic sign). The differential diagnosis includes congenital ichthyosiform erythroderma (CIE) and other congenital recessive ichthyoses(Table 1)[6,8].

After infancy, the erythema tends to fade while hyperkeratosis and scaling become more prominent and darker in color. Predilection sites are the lower abdomen, especially around the umbilicus, the side and nape of the neck, as well as

the large flexures and relative sparing of the face[4]. In more than 50% of patients, mild palmoplantar keratoderma is present. Persistent pruritus is common.

During the first 2-3 years of life, central nervous system involvement manifests with abnormal gait, pyramidal signs, and di- or quadriplegia. Neuroimaging studies reveal that most patients have white matter disease with retarded myelination. Other features are corneal dystrophy, enamel hypoplasia, speech abnormalities, seizures, short stature and kyphoscoliosis

The histopathological features are non-specific and include hyperkeratosis, papillomatosis, and acanthosis with well preserved granular layer.

Management of patients requires a multidisciplinary approach, including dermatologic, neurologic, ophthalmologic, orthopedic, and social collaboration. Aggressive physiotherapy, followed by appropriate surgical intervention, has been shown to drastically improve the mobility of children.

References

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TABLE 1. Clinical Findings in Ichthyosis-Related Diseases

	Sjogren-Larsson	Tay	Refsum	Dorfman-Chanarin	Rud	Passwell
Diplegia	+					
Deafness			+			
Retinitis pigmentosa			+			
Ataxia			+	+		
Peripheral neuropathy			+			
Anomalies of hair		+				
Hepatomegaly				+		
Cardiomyopathy			+	+		
Kidney tubular defects					+	+
Hypogonadism		+			+	+
Mental retardation	+	+	+	+	+	+
Dwarfism		+			+	+
Epilepsy	+			+		+
Myopathy						
Cataracts		+				
Hypoplasia of subcutaneous tissue		+				