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MRI of a very rare hereditary ectodermal dysplasia: PIBI(D)S

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Summary. PIBI(D)S is a acronym for a very rare autosomal recessive syndrome consisting of photosensivity, mild non-congenital ichthyosis, brittle cystine-deficient hair, impaired intelligence, occasionally decreased fertility and short stature. We report a 12-year-old female patient affected by PIBI(D)S with previously unreported MRI findings of central nervous system dysmyelination.

Key words: PIBI(D)S – Trichothiodystrophy – Central nervous system dysmyelination – Magnetic resonance imaging – Cockayne syndrome

PIBI(D)S is an acronym for a very rare inherited syndrome characterized by extreme photosensitivity, mild non-congenital ichthyosis, brittle cystine-deficient hair with trichoschisis (trichothiodystrophy), impaired intelligence, occasionally decreased fertility and short stature [1]. Patients with this syndrome may have abnormalities of the nervous system including spasticity with pyramidal signs, hyper-reflexia, ataxia, dysarthria, intention tremor, peripheral neuropathy and absence of deep tendon reflexes, neurosensory hearing impairment, nystagmus, jerky eye movements and unusually sociable behaviour [1–4]. To our knowledge, less than 20 cases have been reported [1].

We report a new patient affected by PIBI(D)S, with neurological impairment and previously unreported MRI evidence of central nervous system dysmyelination.

Case report

A 12-year-old girl was referred to our institution for investigation of developmental delay. She was born prematurely at 32 weeks, after an uneventful pregnancy and delivery; at birth she weighed 1.6 kg, her length was 51 cm and her head circumference 31 cm. Developmental delay and failure to thrive were observed after the 1st year of life: she sat without support at 17 months, stayed upright after 21 months and learned to walk at 2 years. Severe psychomotor retardation was noted thereafter. In her

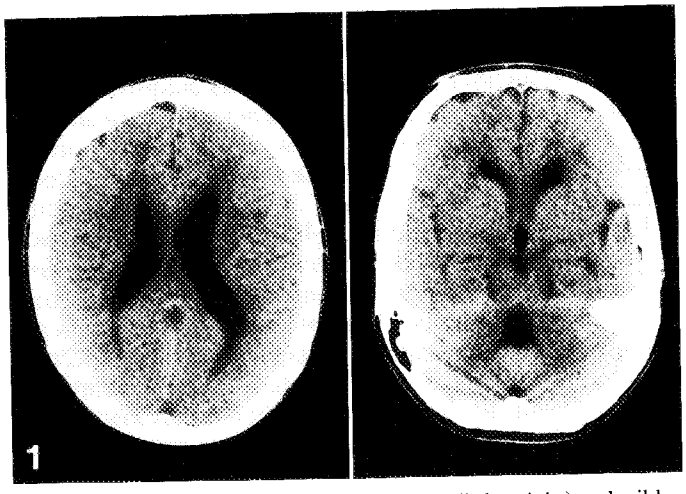

Fig. 1. CT shows slight ventricular asymmetry (left > right) and mild enlargement of the fourth ventricle

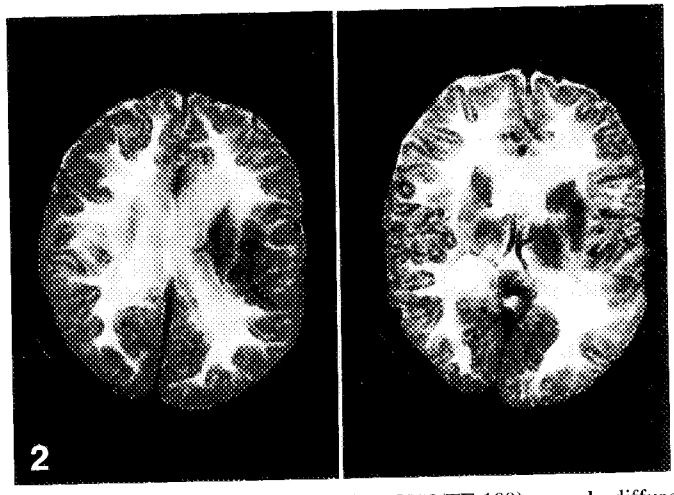

Fig. 2. T2-weighted axial MRI (TR 2000/TE 100) reveals diffuse high signal from white matter and low signal in the basal ganglia

early years she was often in hospital for recurrent infections, most often urinary. She also showed extreme photosensitivity, suffering severe sunburns after a few minutes' exposure.

On admission, she weighed 27 kg (below the 3rd centile), her height was 136 cm (3rd centile) and head circumference 50 cm (3rd centile). She had an unusual face, with receding chin and protruding ears. There was moderate ichthyosis not present at birth; the scalp hair was short, brittle and dry. She had a spastic gait with increased muscle tone; all deep tendon reflexes were brisk and plantar responses extensor. A mild divergent strabismus was observed. Psychometric studies with the Wechsler Intelligence Scale for Children revealed verbal IQ 54, performance IQ 58 and total IQ 56. Ophthalmological examination revealed reduced visual acuity (6/10 bilaterally) and pale optic discs. The EEG showed diffuse slow activity. Visual evoked potentials with flash stimulation were normal, as were audiometry, electromyography, motor and sensory nerve conduction velocities measurement and skeletal age. Cerebral CT demonstrated mild ventricular asymmetry (left > right) (Fig. 1); MRI disclosed diffuse high signal throughout the cerebral white matter on T2-weighted images (Fig. 2).

Routine blood chemistry and urinalysis were negative. Light microscopy of the hair shafts showed clean transverse fractures (trichoschisis); polarizing microscopy showed alternating bright and dark bands giving the shafts a tiger-tail pattern. Amino acid analysis revealed that cystine levels were about 40% lower than in normal hair.

On the basis of the clinical features (photosensitivity, mild non-congenital ichthyosis, brittle hair, impaired intelligence, decreased stature) and the microscopic appearance of the hair, the diagnosis of PIBI(D)S was made.

Discussion

Our patient's neurological deficits (spasticity, with increased deep tendon reflexes, and mental retardation) have been reported in other patients with PIBI(D)S [2, 3]. We have been unable to find any report of MRI studies in patients with PIBI(D)S or with the closely associated neuroectodermal syndromes that, like PIBI(D)S, include trichothiodystrophy [5].

The central nervous system MRI findings in our patient recall those of another genodermatosis with neurological involvement and photosensitivity: Cockayne syndrome, in which changes compatible with diffuse white matter hypomyelination have been reported [6]. Histopathological evidence of demyelinating peripheral neuropathy has also been reported in this syndrome [7]. Our patient does not seem to have involvement of the peripheral nervous system, at least on the basis of electromyography and motor and sensory nerve conduction velocities. However, reduction of peripheral nerve conduction velocities has rarely been found in patients with PIBI(D)S [3, 4].

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