Neuroradiology (2000) 42: 849–851

Tay's syndrome: MRI

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Received: 7 March 2000
Accepted: 13 April 2000

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Abstract Tay's syndrome is a trichothiodystrophy associated with congenital ichthyosis. We report the findings on MRI and spectroscopy in a young girl with sparse, short, ruffled hair, dry skin and delayed milestones. T2-weighted images showed prominent diffuse confluent increase in signal symmetrically in all the supratentorial white matter. These findings are similar to those in a previously described case, and consistent with dysmyelination. Spectroscopy showed increased myoinositol and decreased choline.

Key words Brittle hair · Tay's syndrome · Dysmyelination · Magnetic resonance imaging · Magnetic resonance spectroscopy

Introduction

Tay's syndrome, first described in 1971, belongs to the trichothiodystrophies. It is an autosomal recessive disorder characterised by sulphur-deficient brittle hair with disturbed synthesis of high-sulphur matrix proteins [1]. The features described congenital ichthyosis, brittle hair, growth retardation and progeria-like facies. The syndrome is associated with mental retardation but the pathogenesis of the neurological manifestations, which include rigidity, spasticity, ataxia, microcephaly and poor neuromuscular development, is not fully elucidated [2]. We report a case of this rare syndrome with MRI features and discuss for the first time the role of spectroscopy.

Case report

A 3 years and 10-months old Turkish girl had had dry skin on her flanks since birth. She was the first child of consanguineous parents; the grandparents of mother and father being half-brothers (with a common father). Three of the father's siblings have sparse hair, which grows slowly. Pregnancy and birth were uneventful. The girl showed increased susceptibility to infections and delayed milestones with retarded speech. After pneumonia at 8 months she developed total alopecia. Hair growth returned to normal, but with each new infection hair loss recurred. Examination showed short, brittle hair and dry, scaly skin (ichthyosis) on the abdomen and thorax. The limbs and the periumbilical area were spared. She also had generally dry skin, presacral mongolian spots, café-au-lait spots on the right arm, nail dystrophic loss of fatty tissue, short stature, mildly oblique palpebral fissures, a short philtrum, high palate, prognathia and microcephaly, with an ataxic gait, intention tremor and mental retardation. Laboratory investigations, including various metabolic tests and examination of the cerebrospinal

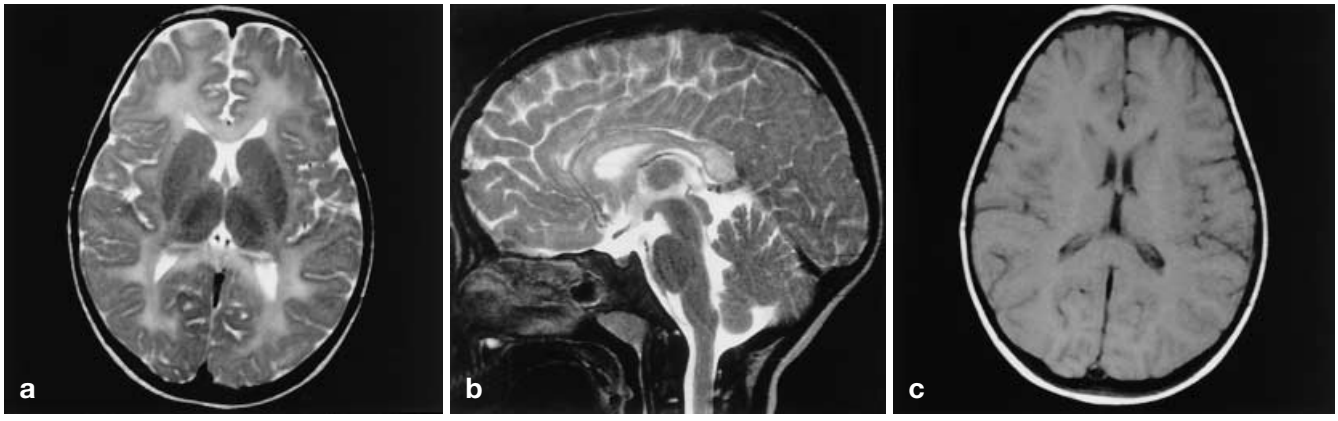

Fig. 1 a, b T2-weighted spin-echo images show diffuse confluent increase in signal intensity in white matter and homogeneous involvement of the corpus callosum. **c** T1-weighted spin-echo image shows decreased grey/white matter contrast

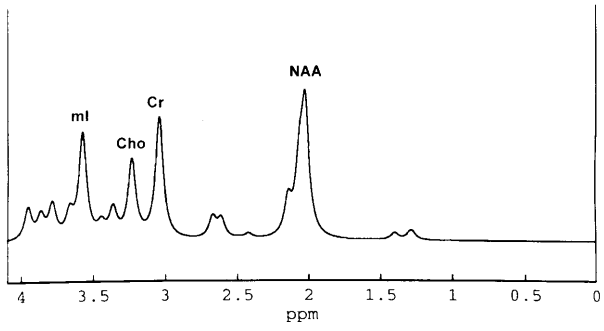

Fig. 2 ^{1}H magnetic resonance spectroscopy (STEAM 1500/20 ms sequence) from left parietal white matter shows lower choline (Cho) and higher myoinositol (mI) peaks than comparable controls

fluid revealed no aetiology. Adrenoleukodystrophy, Canavan's disease, Krabbe's disease, metachromatic leukodystrophy, mucopolysaccharidoses, intrathecal immunoreaction and mitochondriopathies were excluded. Light microscopy of the hair showed trichorrhexis nodosa (swelling on the shaft) and the typical dark and bright banding under polarised light. Photosensitivity was not tested but the history gave no indication of increased photosensitivity. Tay's syndrome was diagnosed on the typical dermatological findings, developmental delay and neurological changes.

Proton-density and T2-weighted MRI showed symmetrically prominent, diffuse, confluent increase in signal of supratentorial white matter (Fig. 1), with diffuse involvement of the corpus callosum but sparing of the immediately subcortical white matter. There was no abnormal contrast enhancement. Brain metabolism was investigated measured by localised ^{1}H magnetic resonance spectroscopy (MRS), using the STEAM (TR/TE 1500/20 ms) and PRESS (TR/TE 1500/135 ms) sequences in volumes of interest (voxels) of 8 cm^{3} in the parieto-occipital white matter. In comparison with appropriate age-matched ^{1}H spectra, this revealed decreased choline (Cho) (Cho/creatine (Cr) ratio 0.63, healthy controls 0.88 ± 0.098), N-acetylaspartate (NAA) within the normal range (NAA/Cr ratio 1.57, healthy controls 1.51 ± 0.059) and increased myoinositol (mI) (mI/Cr ratio 1.01; healthy controls 0.65 ± 0.063).

Discussion

The pathophysiology of the central nervous system involvement in Tay's syndrome is unknown. The deranged synthesis of high-sulphur matrix proteins (the basis of which is not known) may affect synthesis not only of hair and nail, but also of similar matrix proteins in other tissues [3]. Mayer-Puttlitz et al. [4] showed that the high-sulphur matrix proteins, neurocan and phosphacan, have an overlapping or complementary role in axon guidance, cell interactions and neurite outgrowth during nervous tissue histogenesis. Abnormal myelin is unstable and may break down [5], so that it is reasonable to hypothesise that children with Tay's syndrome produce abnormal myelin. Necropsy in a case of (trichothiodystrophy with photosensitivity) showed hypomyelination [6].

Myelin represents a large portion of mature adult brain. Myelin maturation in children is part of normal development. Some diseases that primarily alter synthesis of myelin: demyelination refers to destruction and removal of normally formed myelin from the central and peripheral nervous system, while dysmyelination indicates deficient or defective synthesis of myelin. Recognising either or distinguishing between them is not possible with current imaging. Adrenoleukodystrophy, for example, may have components of both, but this is known from histology and histochemistry and not from imaging [7]. The term hypomyelination may be especially useful when referring to the abnormal appearance of white matter on MRI, in children in the first 2 years of life, which deviates from age-matched standards, irrespective of aetiology or pathophysiology. Symmetrical well defined increase in the signal of white matter with diffuse homogeneous involvement of the corpus callosum and sparing of the subcortical layer

suggests a primary white matter disorder. Similar findings have been reported in one other case of Tay's syndrome [2].

We described the use of MRS in this syndrome for the first time, showing increased myoinositol, a glial marker [8] (also sensitive to osmolarity), which may change in white matter diseases, as here. Choline-containing compounds are membrane components, their level is sensitive to myelin disorders and often decreased. The absence of increased Cho excludes acute demyelination, in which cell-membrane turnover is increased. The MRI and spectroscopy in this case are consistent with abnormal production of myelin. We believe the integrated analysis of MRI and ^{1}H MRS could be of benefit in investigating and counselling families with Tay's syndrome.

References

1. Chen E, Cleaver JE, Weber CA, et al (1994) Trichothiodystrophy: clinical spectrum, central nervous system imaging, and biochemical characterization of two siblings. J Invest Dermatol 103(Suppl.5):154S-158S
2. Ostergaard JR, Christensen T (1996) The central nervous system in Tay syndrome. Neuropediatrics 27: 326–330
3. Price VH (1992) Trichothiodystrophy: update. Pediatr Dermatol 9: 359–370
4. Meyer-Puttlitz B, Junker E, Margolis, et al (1996) Chondroitin sulfate proteoglycans in the developing central nervous system. II. Immunocytochemical localization of neurocan and phosphacan. J Comp Neurol 26: 44–54
5. Barkovich AJ, Lyon G, Evrard P (1992) Formation, maturation, and disorders of white matter. AJNR 13: 447–461
6. Tolmie JL, de Berker D, Dawber R, et al (1994) Syndromes associated with trichothiodystrophy. Clin Dysmorphol 3: 1–14
7. William SB (1997) Pediatric neuroradiology. Lippincott-Raven, Philadelphia New York, pp 175–231
8. Zimmerman RA, Wang ZJ (1997) The value of proton MR spectroscopy in pediatric metabolic brain disease. AJNR 18: 1872–1879