our patient sustained two respiratory arrests related to the use of narcotics. Therefore, avoidance of hypnotics and narcotics in this group of patients is strongly recommended.

Recognition of the fact that adult-onset nemaline myopathy may present with pulmonary symptoms due to diaphragmatic paralysis rather than muscle weakness is of utmost importance to the clinician, since these patients, otherwise, might be subjected to unnecessary and often invasive investigations or inappropriate therapy.

Coincidence of myotonic dystrophy and Down's syndrome (Trisomy 21)

Sirs: In 1981, Bird1 described the first case of myotonic dystrophy associated with Down's syndrome (trisomy 21) and suggested that myotonic dystrophy could have a small but significantly increased frequency of chromosomal aberrations, bearing in mind previous descriptions of myotonic dystrophy associated with other chromosomal anomalies.2–5 The following is a report of a new case of myotonic dystrophy associated with Down's syndrome.

A 15 year old girl was diagnosed as having Down's syndrome (trisomy 21 with deletion of long arms) since birth. In her family, the father, a sister of the father and a sister of the patient suffered from myotonic dystrophy. At her birth, the father was 36 years of age and her mother 34, their respective karyotypes being normal. She had weak suction with cough and nasal reflux, very little spontaneous mobility and muscular hypotonia. She began walking on her own at 18 months of age. At the age of four, she was prescribed corrective lenses because of visual difficulty. Menarche was at the age of 9 years.

At the age of 15 years, she was examined in our department. There was obesity, intellectual retardation, inexpressive and myopathic facies, thick lips, mongoloid palpebral clefts, cataracts in both eyes, high arched palate, caries and badly positioned teeth. She had weakness of the masticatory muscles and, to a lesser degree, distally in the extremities. There was spontaneous myotonia of the hands, which improved with exercise but was not influenced by the cold, and percussion myotonia of the tongue, forearms and hands; generalised muscular hypotonia without a clear muscular atrophy was observed; global incapacity was slight. The electromyogram showed typical myotonic discharges. The levels in blood of CK, aldolase, GGT, GPT, LDH, T3, T4, TSH, PRL, FSH, LH, GH, ACTH and cortisol were normal. An oral glucose tolerance test following ingestion of 75 g of glucose showed: fasting 68 mg/100 ml; 30 minutes 106 mg/100 ml; 1 hour 115 mg/100 ml; 2 hours 71 mg/100 ml and 3 hours 60 mg/100 ml. The IgG in serum was 700 mg/100 ml (normal 800–1200), with normal IgA and IgM. The WAIS intellectual coefficient was 40. Cerebral CT scan was normal.

Considering that the frequency of Down's syndrome (trisomy 21) is 1 in every 600 births6 and that myotonic dystrophy has a prevalence of 2 to 5 cases per 100 000 population,7 the probability of both diseases being present in the same individual is extremely low. Besides our patient, there exists only one report of this association.8 However, the coexistence of myotonic dystrophy with another chromosomal anomaly, such as Klinefelter's syndrome (47 XXX),2–5 has been described. In addition, Nadler et al9 reported two brothers with myotonic dystrophy and phenotypic features of Klinefelter's syndrome before cytogenetic evaluations were available, and Sparkes and colleagues10 described a patient with Klinefelter's syndrome and myotonic dystrophy, although there was not a family history of myotonia, frontal balding, cataracts or endocrine dysfunction.

Further descriptions of new cases are necessary to determine whether these associations are merely coincidence, or whether myotonic dystrophy is associated with an increase in frequency of chromosomal anomalies.

References


2 Grumbach MM, Blanc WM, Engle ET. Sex chromatin pattern in seminiferous tubule

Letters


2 Grumbach MM, Blanc WM, Engle ET. Sex chromatin pattern in seminiferous tubule

References


2 Grumbach MM, Blanc WM, Engle ET. Sex chromatin pattern in seminiferous tubule

Letters


2 Grumbach MM, Blanc WM, Engle ET. Sex chromatin pattern in seminiferous tubule

References
Letters

dysgenesis and other testicular disorders: Relationship to Klinefelter's syndrome. J Clin Endocrinol Metab 1957;17:703-36.

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Taste loss as an initial symptom of primary amyloidosis

Sir: One of the major clinical features of amyloidosis is polyneuropathy, especially of the sensory dominant type, observed in 90% of those with familial and 10-20% of those with non-familial amyloidosis. Taste is a chemical sense conducted via the sensory nerves, but taste disorders have not been described in the reported sensory disturbances of amyloidosis.1 2 Taste was occasionally accompanied by syncope. She subsequently developed constipation in August, liver dysfunction and slight hepaticomegaly in October, urinary disturbance in March 1985, and anosmia and sensory disturbance of the limbs in June.

She was admitted to Kagawa Central Hospital in June 1985. Taste was completely lost in all areas of the chorda tympani, glossohyphgeal, and vague nerves. Electrogustometry was negative at the maximal stimulation of 32 dB (normally positive at more than 8 dB). Qualitative and quantitative clinical gustometry (using filter-paper discs4) were all negative for 80% sucrose (sweet), 20% saline (salty), 80% tartaric acid (sour), and 4% quinine HCl (bitter). These concentrations are several to more than ten times the normal threshold levels. Touch sensation of the tongue and the oral cavity was normal, and no local tongue lesions such as macroglossia were noted. Other neurological findings included left anosmia, left mydriasis, loss of left light reflex, decreased salivation and lacrimation, areflexia of distal extremities, and peripheral neuropathy of the socks-gloves type. The motor nerve conducting velocity showed a slight decrease, being 38-9 m/s for the right median nerve and 38-6 m/s for the left posterior tibial nerve. Sensory nerve conduction could not be evoked. Other clinical findings were goitre, hepatomegaly, nephrotic syndrome, constipation, neurogenic bladder, and orthostatic hypotension. Ultrasonic echocardiography showed thickness and granular sparkling texture of the interventricular septum and posterior wall, suggesting cardiac amyloidosis. Laboratory findings in addition to the abnormal values corresponding to the clinical findings above were positive serum electrophoresis for M-protein in the gamma-globulin fraction, and immunoelectrophoretic analysis demonstrating M-proteinaemia IgG-L type. Urine concentrated 25-fold also yielded M-protein and Bence Jones protein of lambda type. Plasmocytes of the bone marrow showed a slight increase to 4-4% without abnormal nuclei. Biopsy of the minor salivary gland and thyroid gland revealed deposition of amorphous substances which were stained with Congo-red and showed green birefringence under polarising microscopy. These substances were identified by electron microscopy as scattered amyloid fibrils and a diagnosis of primary amyloidosis IgG-L type was made.

Upon diagnosis, treatment was initiated with merpharan (2 mg/day), prednisolone (10 mg/alternative day), and dimethylsulfoxide (10 ml/day). Although this treatment produced moderate improvements in the sensory disturbance of the upper extremities, the loss of taste persisted and other symptoms either remained unchanged or deteriorated. Moreover, oedema, anaemia, petechiae, and nausea developed thereafter, and slight enlargement of the tongue was noted in November 1985.

The patient diagnosed as having primary amyloidosis IgG-L type was noted particularly for showing loss of taste as the initial presenting symptom. Taste disturbances may be caused by head trauma, tumour, lesions of the conduction route for taste, systemic disorders such as diabetes mellitus, cold, psychological disorders, and side effects of drugs.5 The loss of taste in our patient could be readily differentiated from gustatory disorders arising from these causes. Zinc deficiency, another frequent cause of taste disturbance,6 was also excluded since the administration of zinc sulfate was ineffective. Acute pandysautonomia also causes taste loss, but the other symptoms are confined to the autonomic nervous system and recovery may occur.7 Taste disturbances have not been described in reports of amyloidosis,3 4 8 and thus their severity or incidence has still to be clarified. However, the tongue is one of the frequent sites of amyloid deposition and may show various pathological conditions such as induration, yellowish nodules, haemorrhagic bullae, and aphthous ulcer.8 Amyloidosis, therefore, may well be expected to complicate taste disorders. Our patient presented with a tendency to macroglossia in November 1985, suggesting amyloid deposition also involving the tongue. The loss of taste may be ascribed to this condition, and the appearance of taste loss preceding that of macroglossia by 3 years in our patient indicates that a small amount of amyloid is sufficient to induce loss of taste. Therefore, taste loss may be an initial symptom of amyloidosis. Our experience emphasises that quantitative gustometry is necessary for investigating amyloidosis, and it may allow an earlier diagnosis of amyloidosis.

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References

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