a significant number of cases occurring below 50 years of age, [c] Absence of gastric cancer as part of cancer family syndrome, [d] additional risk posed by fried food. Since patient number was small, these results need confirmation by larger field studies.

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Myotonic Muscular Dystrophy

Sir,

The disease is seen worldwide with a particularly high frequency in French Canadians in Quebec where all cases can be traced to a single ancestor.1-3 The molecular basis of disease lies in expansion of a trinucleotide (cytosine – thymine – guanine CTG) repeat sequence in the 3’untranslated region of the myotonic dystrophy protein kinase (DMPK) gene on chromosome 19q. This expanded gene is unstable and its size increases with age. The size of repeat on gene is directly proportional to clinical severity of disease and has inverse relation with age of onset of the disease.1-3

25-years-old male presented with decreased power of gripping, which progressed to difficulty in holding objects in hands over 5 years. There was difficulty in holding neck while getting up from lying down position and dysphagia, and nasal intonation of voice along with nasal regurgitation of fluids for last 3 years. There was no family history of the disease. On examination atrophy of muscles of face (hatchet shaped face), temporalis, masseter along with sternocleidomastoid bilaterally was noted (Fig. 1). Percussion myotonia could be demonstrated both on thanar muscles (Fig. 2) and tongue. Rest of neurological examination and systemic examination was normal. EMG studies showed myotonia along with decreased amplitude of action potentials and polyphasic potentials. Muscle biopsy revealed muscle fibers of variable size with atrophy of fibers. There was presence of central nuclei and ring fibers.

‘Classical form’ of the disease is seen in adolescent or early adult life with variable presenting features. Muscular weakness, myotonia, mental retardation, cataract, neonatal problems are common symptoms and about 18% remain asymptomatic.1, 3 The clinical severity of the disease ranges from death in utero to mild symptoms without physical signs in old age.2 Superficial facial muscles, levator palpebral superioris, temporalis, sternocleidomastoid, distal muscles of forearm and dorsiflexors of foot are most prominently affected. The atrophy of facial muscles gives “typical hatchet shaped appearance” to face. Quadriceps, diaphragm, intercostals, palatal muscles, pharyngeal muscles, and extracocular muscles are also commonly involved. Muscles of pelvic girdle, hamstrings, soleus, and gastrocnemius are spared.1 Smooth muscle involvement of gastrointestinal tract may lead to dysphagia and irritable bowel syndrome like symptoms.
Conduction defect and cardiomyopathy denotes cardiac involvement. Cataract, frontal baldness in male, gonadal atrophy, hypersomnia, mild endocrinial anomalies, bone changes and abnormalities of immunoglobulins can be other associated disorders.1-3

‘Congenital form ’ is evident at birth with history of polyhydramnios and poor fetal movement in pregnancy. There can be respiratory and feeding difficulty in neonatal period with death of few, in those surviving until late teens or early adulthood, hypotonia resolves and motor function improves but during adolescence the features of classic adult form appear. The ‘late onset form’ is associated with a small CTG-repeat expansion and is typically asymptomatic or oligosymptomatic.1-3 Death is usually due to respiratory infections or cardiac cause.

There is currently no specific therapy. The surgical procedures are avoided due to associated anesthetic sensitivity, postoperatively respiratory muscle inadequacy with high incidence of arrhythmias. These patients require regular, general, and neuromuscular assessment for detection and correction of systemic disorders and for better quality of life. Physiotherapy, occupational advice, and speech therapy all have a role in such patients. Genetic counseling by means of chorionic villous sampling can be offered for screening to the members of family affected.1-3

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