clinical condition improved after spontaneous drainage in combination with trimethoprim-sulfamethoxazole therapy.

Currently, the infecting strains of CA-MRSA in Taiwan are thought to have unique microbiologic characteristics such as resistance to multiple antibiotics (including clindamycin, erythromycin, tetracycline and chloramphenicol), different exotoxin gene profiles (*e.g*., PVL and SEB), common pulsed-field gel electrophoresis patterns (which are different from those of the major pandemic clones of hospital-acquired MRSA), ST 59 genotype by MLST, and smaller SCC*mec* variants: SCC*mec* type V_T, or less frequently, type IV [5]. The microbiological characteristics of the CA-MRSA strain infecting our patient are consistent with the results of previous studies. The present case suggests that CA-MRSA should also be considered a potential cause of both suppurative OM and SSF in children.

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Congenital Myotonic Dystrophy with Asymptomatic Mother

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Myotonic dystrophy is an autosomal dominant neuromuscular disorder characterised by extreme pleiotropism and variability in disease expression. A congenital form is rare and is observed in infants born to symptomatic mothers with multisystem involvement. We report a case of a neonate with congenital myotonic dystrophy born to an asymptomatic mother.

Key Words: *Asymptomatic mother, Congenital Myotonic dystrophy, Diaphragmatic weakness, Floppy infant.*

Myotonic dystrophy (DM) is the most common inherited neuromuscular disorder with a prevalence of 1:8000 [1]. It is caused by a triplet repeat expansion (CTG) in the non-coding region of the myotonin gene at 19q13.3. The typical form has an onset in early adult life and is characterised by myotonia, skeletal muscle weakness and wasting, cardiac conduction defects, and cataracts. We report an infant with genetically proven congenital myotonic dystrophy, the diagnosis of which was clinched by the combination of generalised floppiness and diaphragmatic weakness.

CASE REPORT

The proband was born via caesarean section to non-consanguineous parents at 32 weeks of gestation, which was complicated, by polyhydramnios and breech presentation. Baby cried immediately after birth. The couple had a previous first trimester miscarriage. Baby was admitted to neonatal intensive care unit following mild respiratory distress. Physical examination revealed severe tachypnoea, generalised floppiness, cold peripheries, microcephaly, narrow palpebral fissure, antimongoloid slant, low set ears and bilateral simian crease.

Blood parameters including complete blood count, electrolytes, blood urea, serum creatinine and C reactive protein were all within normal limits. Chest radiograph showed bilateral high domes of diaphragm. Based on sudden unexplained respiratory failure and radiographic findings, possibility of a neuromuscular disorder was considered.

Electrophysiological studies were not done as the baby was on ventilator. Care was withdrawn on day 2 of life after discussing with the parents, as the ventilatory requirements were very high and the prognosis was grim. Echocardiogram and electrocardiogram were normal. DNA from the baby was subjected to Southern blot analysis. The baby was found to be heterozygous as the number of CTG repeats in one of the alleles was >700 (normal 4-37 repeats) whereas in the other allele, the repeat size was only 4. This confirmed the diagnosis of congenital myotonic dystrophy in the baby. Karyotype of the baby was 46XX.

Both parents were apparently healthy and did not show any classical symptoms of neuromuscular disorders. However, on examination, mother had minimal percussion myotonia of thenar muscles and tongue. Electromyography of the mother revealed myotonia from all tested muscles of upper and lower limbs. Her CPK levels were 92 U/L, fasting blood sugar was 92 mg/dl and TSH level was 3.23 uIU/L. Ophthalmological evaluation revealed bilateral early cataracts. Southern blot analysis of the mother showed >300 repeats from one allele and 13 repeats from the other allele. This confirmed that the mother is also affected with myotonic dystrophy.

DISCUSSION

DM is characterised by extreme variability, anticipation and differential expansion in the maternal and paternal germline, so much so that congenital myotonic dystrophy (CDM) is always transmitted from the maternal allele. The usual number of repeats is 4-37. The repeat number goes up to 200-500 in classical myotonia and in CDM the CTG repeat are usually >1000 [1].

CDM was first described in 1960 and is the most severe phenotypic expression of DM1 [2]. Congenital form is frequently fatal and is usually observed in infants born to classically affected mothers. One interesting feature in this case is the fact that mother was totally asymptomatic. The explanation for selective maternal transmission as the major cause for CDM is that mature spermatozoa can carry only small expansions whereas ova can accommodate much larger expansions. Koch, *et al*. found that only women with multisystem signs of DM1 at the time of pregnancy and delivery were likely to have congenitally affected offspring and that the chance of having a more severely affected child increased with maternal disease severity [3,4]. These observations have been supported by more recent molecular studies which showed that infants with CDM and their mothers had greater amplification of the CTG repeats than those with non-CDM and their mothers and the maternal expansion was three times greater in the CDM group than in the non-CDM group [5,6].

The differentials include congenital muscular dystrophy, neonatal myasthenic syndrome, conge-

nital myopathy and spinal muscular atrophy with respiratory distress type 1. Congenital muscular dystrophies are characterized by hypotonia and need at birth or shortly thereafter with multiple joint contractures. Neonatal myasthenic syndromes includes familial myasthenia with prominent respiratory and feeding difficulty at birth, and congenital myasthenia with predominant ocular findings. Congenital myopathies presenting as neonatal hypotonia needs muscle biopsy for confirmation. Spinal muscular atrophy with respiratory distress type 1(SMARD1) is a distinct genetic disorder and these babies are usually born with intrauterine growth retardation with a weak cry, and foot deformities [7]. Polyhydraminos and reduced foetal movements are two important ultrasound markers that should alert the clinician to consider the diagnosis of CDM. In a mother with 200-500 repeats and with a previous child with CDM, the risk or recurrence is 40-50% [1]. The mother was counselled regarding the prospects of prenatal diagnosis in her future pregnancy by determination of CTG expansion in the fetus by chorionic villus sampling or amniocentesis.

The diagnosis of congenital myotonic dystrophy is often difficult in babies whose parents are not diagnosed. The lack of family history in a baby with severe manifestation has only been rarely reported in literature [8]. The number of trinucleotide repeats is known to predict the degree of muscular disability in DM [9], but as shown here, may not correlate with individual clinical features.

Contributors: KMA was involved in patient management, conception and design of study, analysis of data, SN has done the genetic work up and confirmed the diagnosis and has critically reviewed and modified the manuscript. JP has evaluated the mother and has helped in drafting the manuscript. VMB is involved in acquisition and interpretation of data, drafting of manuscript and review of literature.
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