

Angelman's "Happy Puppet" Syndrome

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Angelman in 1965 described a syndrome of mental subnormality, jerky movements, brachycephaly, seizures, episodes of unprovoked laughter and protruding tongue in children(1). He used the term "puppet" children for them. This was later on changed to 'ehappy puppets' because they always looked happy(2). The recognition of the entity is increasing. However, less than 100 cases have so far been reported. A typical case of this syndrome is reported here.

Case Report

A five-year-old male child was brought to the Rehabilitation Centre for delayed development especially speech delay, and seizures. He was born at term by a low forceps-assisted delivery to a primigravida mother. The antenatal, perinatal and neonatal periods were uneventful. He weighed 2.3 kg at birth and had an immediate cry. His milestones were delayed from early infancy in all sectors; speech was particularly affected. Generalized tonic-clonic seizures were first noted at two years of age, for which he received Phenobarbitone, and was seizure-free for the past one year.

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On examination, the facial features were typical, a wide mouth, thin upper lip, a pointed jaw and a constant smile (*Fig. 1*). He had episodes of inappropriate laughter. His head circumference was 48 cm (<3rd centile) and occiput flat. There was generalized 'etremulousness' and a jerky ataxia of all limbs. The gait was unsteadily and wide based. The muscle tone and all the reflexes were normal. He could follow simple commands, point towards objects, differentiate 3 colors, and could barely speak Mama-Papa.

The CT scan of the head was normal. The sleep EEG showed a background rhythm of 8-9/sec. Sharp waves were seen in frontal regions with a tendency to become generalized. Posterior slow waves 1-2/sec of high amplitude (220-250 uV) were also seen (*Fig. 2*).

Discussion

This patient fulfilled all the diagnostic

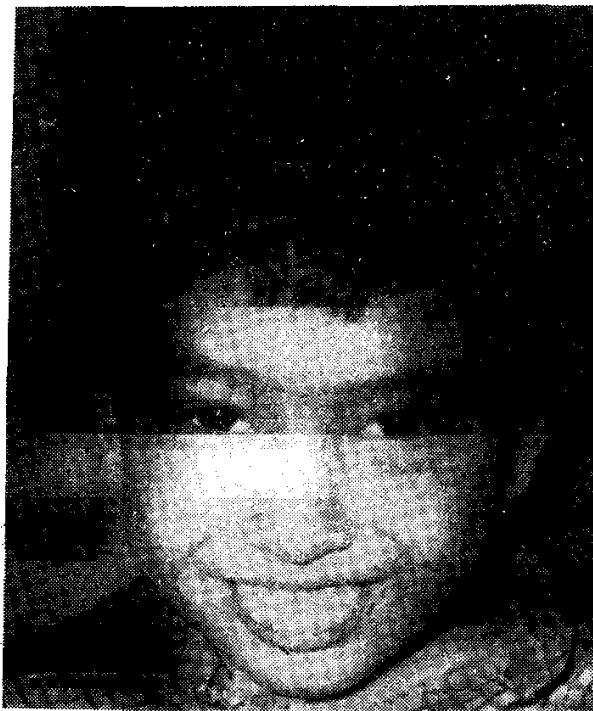


Fig. 1. Photograph showing happy looking child with wide mouth, thin upper lip, pointed chin and tongue thrusting.

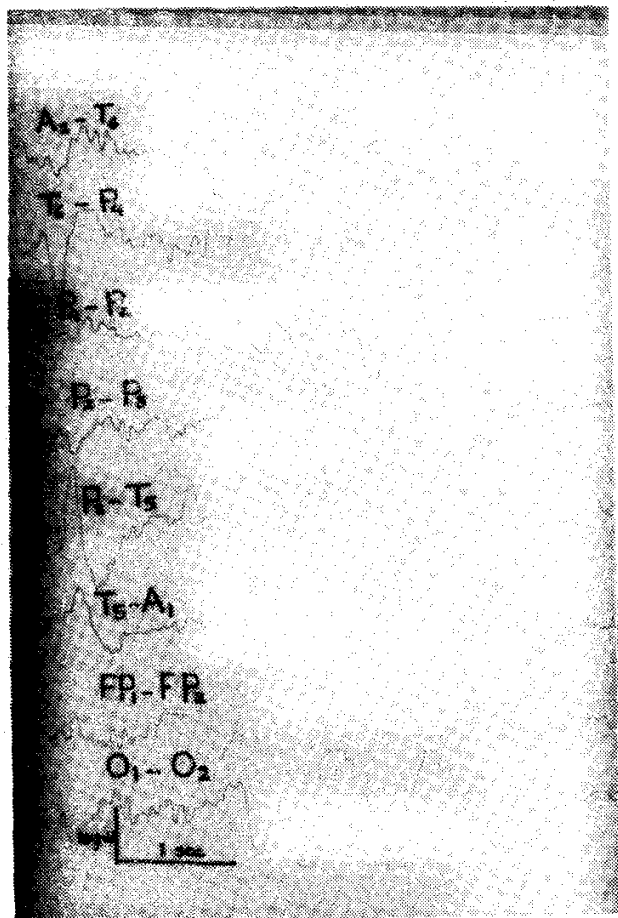


Fig. 2. Sleep EEG showing high amplitude posterior slow ($1.2/\text{sec}$) waves.

criteria of Angelman's syndrome namely: delayed development, jerky ataxia, head circumference below the 50th centile for age, dysmorphic craniofacial features (prominent lower jaw and wide mouth), happy disposition, and episodes of paroxysmal electroencephalogram (EEG) (3).

The syndrome is seen equally among boys and girls. Most of the children with the syndrome are born normally and have normal growth. The neurodevelopmental delay occur from early infancy, and is a particularly profound in the area of language development. The facial features, though not pathognomonic, are highly suggestive. These are mid-facial hypoplasia, prominent mandibles, pointed chins, and thin upper lips. Frequent tongue thrusting

may lead to bowed upper dentition. There may be an evolution of facial features so that below the age of one year clinical diagnosis may be difficult. Episodes of paroxysmal laughter are common but may not always occur in infancy. The laughter is not an expression of happiness; probably it is due a defect at the brain stem level (4). Many children have microcephaly or microbrachycephaly with horizontal occipital depression as seen in our case. Jerky ataxia involving all the limbs, and a wide based gait are generally normal though some children may show variations in muscle tone with brisk reflexes. The plantar reflexes are flexors.

Seizures are present in most cases, mean age of onset being two years. A few may not have clinical seizures, but have abnormal EEG in form of frequent spikes, short wave discharges and high amplitude slow waves with excess slow activity (5). Generalized, rather rhythmic intermediate slow waves 4-6 cycles/second (C/S) have been frequently reported in children less than 4 years of age. Runs of 2-3 C/S activity anteriorly often with discharges have also been reported. On eye closure, posterior high amplitude slow waves mixed with spikes 2-4 C/S have been considered diagnostic. Our patient had this finding (Fig. 2). Computerized tomography of the brain is often normal; mild cerebral atrophy is seen occasionally.

Biochemical screening and hematological parameters are mostly normal. Chromosomal analysis has shown deletion of 15q 11-13 in some cases (6). Familial cases suggest that the syndrome may have genetic implications (7,8). Accurate recognition of the syndrome is important for discussion of prognosis and management, and for genetic counselling.

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Maternal Knowledge of Childhood Immunization

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Childhood immunizations are a cost effective large scale method of protecting children's lives. Mothers are the key figure

of support around their children and are thus the most effective primary health care workers for the children. Any successful breakthrough in the child health must necessarily centre on the mother(1). A substantial proportion of mothers have been found to be unaware of the protective values of immunization(2-4). This study was undertaken to evaluate the mothers' knowledge of immunization in an Urban Medical College Hospital.

This study was conducted in the Department of Pediatrics, Medical College, Baroda between January and May, 1990. Three hundred and four mothers of children aged 6 months to 5 years attending Pediatric OPD and indoors were the subject of the study. The mothers were explained the purpose of the study by a Pediatrician and interviewed about their knowledge and awareness of immunization in childhood. Their responses were recorded on a pretested semistructured proforma. Statistical analysis was performed by the Chi square test.

Results

Of the 304 mothers, 218 (71.6%) were aware of the need and availability of immunization, while the remaining 86 (28.4%) mothers had no knowledge about immunization. The mean age of mothers was 23.5 years. Of the 218 mothers, 44 (20.1%) were uneducated while 174 were educated (107 had education up to middle, 40 up to higher secondary and 18 were graduates

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