CONGENITAL DOUBLE ATHETOSIS, DEAF-MUTISM AND MENTAL DEFICIENCY: A REPORT OF FIVE CASES.

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[Received 22 August, 1949.]

CONCURRENCE of the above-named symptoms does not appear to have been reported in the literature. Congenital double athetosis and deaf-mutism usually occur independently of one another. Out of about 1,500 patients at this institution there are 14 with the former condition and 25 with the latter. The relation of the syndrome to its chief constituents may be clarified by a preliminary account of this larger context out of which 3 of the 5 cases here reported are drawn.

Congenital double athetosis has as its central symptom involuntary movements of slow, writhing character, affecting chiefly face, tongue and limbs. There is also the tendency to the adoption of characteristic postures which may lead to permanent deformities. The distal limb muscles are more affected than the proximal. The associated increased tonus of the limbs is at least partly voluntary in origin, brought about to inhibit the movements. There is no pyramidal involvement and the plantar reflexes are flexor.

The degree of disability varies from complete helplessness and speechlessness to a mere maladroitness and clumsiness of the finer and more subtle movements. Of 14 cases here, 2 are unable to walk and practically deprived of speech, 2 have a clumsy and precarious gait and grossly impaired articulation, while the remainder can walk and talk and feed themselves.

Mental deficiency is quite a common concomitant of congenital double athetosis, and is naturally present in most of the cases at this institution. It is seldom severe in degree, nearly all cases being in the dull and backward or feeble-minded class. There appears to be no correlation between the degree of the deficiency and the severity of the athetotic symptoms.

	Uncomplicated.	With epilepsy.	With deaf-mutism.	Total.
Congenital double athetosis	9	3	3	14
Above with disease of pyramidal system	7	8	—	15

A comparison of patients with congenital double athetosis alone and those also showing signs of pyramidal disease reveals some points of interest:

(1) The average intelligence of the former group is appreciably higher than that of the latter.

(2) Epilepsy is rather commoner in the presence of pyramidal disease.

(3) None of the patients with associated deaf-mutism display pyramidal signs.

The table reveals some of these points.

There are 25 cases of deaf-mutism, only one of which has epilepsy, and that particular patient also has athetosis. There is no association with pyramidal disease. The accompanying mental deficiency is mild in most cases, being somewhat similar in degree to that which occurs in uncomplicated athetosis.

It should be noted that neither of the two groups described is homogeneous. For instance, one patient with deaf-mutism also has cretinism, while one case of athetosis is known to be due to Rh incompatibility.

Summarizing the relation of the three patients at this institution with the full syndrome to the two groups described:

(1) The degree of athetosis is relatively mild.

(2) Signs of pyramidal involvement are absent.

(3) The mental deficiency is of mild degree, corresponding to that commonly found in other patients with athetosis or deaf-mutism.

(4) Epilepsy is present in one case. This is quite common in uncomplicated athetosis, but not significantly so in deaf-mutism.

It may be asked whether the concurrence of athetosis, deaf-mutism and mental deficiency is incidental or whether it is based on a single pathogenesis, and it is convenient to consider this point before describing the cases in detail. On the basis of the facts already given it seems probable that the combination is not incidental. The expected admission rate of patients doubly afflicted would be $14/1{,}500 \times 25/1{,}500$. The actual admission rate is $3/1{,}500$. If the double handicap were to double the likelihood of admission, the actual admission rate would still be 6–7 times the expected rate. Moreover, if the association were incidental, the chance of admitting such a patient who also has pyramidal disease would be slightly greater, and yet there is not one such patient in this institution.

CASE 1.—A. R—, male aged 11 years, 7 months. Examined by Dr. S. Nevin and Prof. Penrose. Estimated mental age 3. I.Q. 26. W.R. doubtful. Cephalic index ·82.

History.—Normal birth. Had jaundice three days after birth. Did not walk until 3. Presumably deaf from birth. Mother deaf herself. Patient said to have been mischievous at home and used to play with the fire.

Physical condition.—Diffusely bossed skull; mannerisms and athetosis of hands. Flat-chested. Cardiovascular and respiratory systems normal. Palate and tongue normal. Ears prominent and large. Umbilical weakness. Central nervous system: Reflexes normal. Very poor co-ordination of hands and poor power. Eyes brown. Pupils slightly centrally displaced.

Behaviour.—Attention quite good. Understands signs very clearly. Able to use spoon and fork. Walking good. Slight use of gestures. Clean habits. Not subject to fits.

Family history.—Patient is the fourth of eight children. The other children are all normal. Mother normal except for deafness, which followed scarlet fever at the age of 7 years. No miscarriages. Mother's parents and eight siblings are normal. Siblings' children normal. Father normal. Eldest of family of ten, all of whom were healthy. Father's parents were normal.

CASE 2.—A. L. P—, male, aged 23. Estimated mental age, 11. I.Q. 79. W.R. negative. Cephalic index ·77. Height 5 ft. 5 in. Hair fair. Eyes brown. Blood group $O.R_2r$.

History.—Age of mother at birth 26 years. Mother in good health during pregnancy. Labour difficult. Forceps delivery. Patient nearly died at birth and nearly died again on the third day. There was difficulty in getting the infant to suckle. Fed by spoon and later by bottle. No history of jaundice. Learnt to walk after the age of 2 years. Believed to have had three slight fits in infancy. Noticed to be deaf at an early age.

Physical condition.—Of good physique. Cardiovascular and respiratory systems normal. Tongue and palate normal. Good teeth. Nothing of note outside the nervous system. Central nervous system: Tendon reflexes brisk. Plantar reflexes: Right, flexor; left, indefinite. Pupils react to light. Athetotic movements of tongue, face and neck. Hands: poor co-ordination and tendency to adopt athetotic postures. Gait good.

Behaviour.—Deaf and dumb. Understands gestures. Washes and dresses himself. Feeds with spoon and fork. Collects the laundry and works in the ward. Cheerful, sociable and well behaved.

Family history.—Father dead. Suffered from diabetes. Backward in learning to walk and talk. Was, however, a shipowner's cashier and in a good financial position. Father's mother alive and well. Normal. Mother alive and well. Of normal or superior intelligence. Was a hospital nurse. Her siblings are normal. She knows of no physical or mental disorders in her family or that of her late husband. No consanguinity. Patient is the eldest of two siblings. The other is a girl of 20 years who is normal.

CASE 3.—R. W. T—, male, aged 30 years 5 months. Estimated mental age 10. I.Q. 67. W.R. negative. Cephalic index ·79. Height, 5 ft. 2 in. Hair dark. Eyes brown. Blood group O. R_1r.

History.—Age of mother at birth 30 years. No precise information regarding birth available, but had jaundice at birth, which lasted for several weeks. Was able to walk and talk at 6 years, but how long before that is not certain. Has had measles and chicken pox. At age of 10 years was said to have been spiteful and mischievous.

Physical condition.—Of good physique. Cardiovascular systems normal. Palate and tongue normal. Abdomen normal. Stiffness of right leg due to tuberculosis of the right knee-joint. Central nervous system: Tendon reflexes brisk and equal; plantar reflexes flexor. Athetotic movements of tongue, neck face and hands. Gait clumsy but quite good. Some increased tonus of limb muscles.

Behaviour.—Deaf and dumb. Pays attention and understands signs well. On one occasion drew a rough but recognizable outline of his leg with an arrow pointing to the place where the plaster was hurting him. Was able to wash before the joint disease developed. Feeds with spoon and fork. Used to do simple work in the ward. Cheerful and sociable.

Family history.—Whereabouts of parents now unknown. The father was an electrician of normal mentality. His parents were normal. The mother was normal and her parents were normal. The patient is the youngest of three children. The eldest was alive and well at the age of 12 years. The second died of whooping-cough at the age of 5 weeks.

CASE 4.—P. A. C—, female, aged 17. Mental age 12. I.Q. 80 (Drever-Collins). W.R. negative. Cephalic index ·786. Blood group, O. R_1r. Hair dark. Eyes hazel.

History.—Illegitimate. Early history unknown.

Physical condition.—Good physique and attractive appearance. Palate and tongue normal. Cardiovascular and respiratory systems normal. Abdomen normal. Nervous system: Pupils react to light and accommodation. Tendon reflexes normal. Plantar reflexes flexor. Muscular hypotonia. Involuntary movements of limbs and of face, neck and shoulders. These are slighter than in the other cases and rather more choreiform than athetoid.

Behaviour.—Deaf and dumb. Washes and dresses herself. Walks well and feeds without any difficulty. She is now beginning to write and has learnt to say some words spontaneously as a result of lip-reading, although she has not had any special training. She is usually sociable and pleasant but is spiteful at times.

CASE 5.—An abstract of this case was kindly supplied by Dr. Kirman, of the Fountain Hospital.

J. L. F—, male, aged 5 years 4 months. On admission (aged 3 years 6 months): Height 38 in., weight 30 lb. Cephalic index ·77. Left-handed.

History.—Patient is the result of the second pregnancy. The first child was a stlllborn but apparently normal baby. There was a post-partum haemorrhage and the mother was transfused. Patient born in 1944 and suffered from icterus gravis. His blood was noted to be Rhesus positive. During the third pregnancy the mother was noted to be Rhesus negative and to have a rising titre of anti-D. substance in her serum. The father was Rhesus positive (homozygous).

Progress of patient.—(May, 1949). Found to be deaf-mute and also to exhibit a number of choreiform movements. Very restless. Unable to wash or dress himself. Totters a good deal but gait improving. Willingly obeys, but not spoken commands. A mental test in November, 1948, gave him an I.Q. of approximately 49 on the Merrill Palmer.

DISCUSSION.

There are various causes of athetosis, as also of deaf-mutism, acting upon the organism at different stages of development. At what points do the two paths of possible pathogenesis coincide?

Hereditary causes.—According to Denny Brown, "the affection (congenital athetosis) sometimes is hereditary, more often familial, but usually isolated." The hereditary tendency in deaf-mutism is more commonly in evidence, the mode of inheritance being usually recessive, autosomal or sex-linked. There is, however, no known genetic connection between the two diseases.

No familial or hereditary tendency could be established in the cases reported.

Prenatal causes.—Of these the most interesting possibility is Rhesus incompatibility between mother and foetus. The occurrence of athetosis associated with kernicterus of this origin is well known. We know of no published evidence favouring Rhesus incompatibility as a cause of deaf-mutism. The evidence for case 5 being produced in this way is, however, rather strong, since the child is known to have had icterus gravis. The history of Case 3 is, perhaps, similarly suggestive, but not convincing. The affected patient was the third of three children and had jaundice at birth which lasted for three weeks. In no case here reported can the possibility of this origin of the disease be ruled out, for all the patients are heterozygous positive, but on the basis of the history Case 2 is the least likely to be so caused.

Birth injury.—Norman reported three cases of mental deficiency associated with pyramidal involvement. Athetosis was present in one of these cases and the pathological findings showed *état marbré* associated with other changes characteristically due to birth injury, of which there was also a strong family history. Evans found a significant association between congenital athetosis on the one hand and primogeniture, prematurity and a history of birth injury on the other. There therefore appears to be evidence that birth injury may sometimes produce congenital athetosis.

Evidence of any causal connection between birth injury and deaf-mutism is more scarce, but Hallpike reported a case of deaf-mutism in which an unhealed fracture of the labyrinth capsule on the right side was found, probably due to birth injury.

The history of Case 2 is strongly suggestive of birth injury.

No definite conclusions as to aetiology can be drawn. It appears to us, however, that the causes most favoured by the evidence available are Rhesus factor incompatibility and birth injury. The combination of symptoms may, of course, be sometimes due to one factor, sometimes another, and sometimes to an incidental association of unrelated events. For instance, the author has seen a patient with athetosis and deaf-mutism in whom the former condition is due to kernicterus, the latter to bilateral mastoidectomy performed at an early age. (I am grateful to Dr. V. Cowie for drawing my attention to this case.)

The occurrence of deaf-mutism and athetosis as a result of birth injury has some pathological evidence to support it. This paper provides some clinical evidence that Rhesus factor incompatibility may also give rise to the syndrome. We know of no pathological findings that would account for deaf-mutism of this origin.

Summary.

(1) Five cases of a syndrome characterized by mental deficiency, congenital double athetosis and deaf-mutism are described.

(2) A brief account is also given of the athetotic and deaf-mute "population" from which three of the five cases are drawn.

(3) The aetiology is discussed.

I am grateful to Prof. Penrose for the case notes of Case 1 and to Dr. Kirman for the abstract of Case 5; also to Dr. Hilliard for permission to make use of this case and to Dr. Taylar for allowing me to make use of the material at this institution.

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