AN ANALYSIS OF TWENTY-SIX CASES OF MONGOLISM.*

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LITERATURE.—Until recent years very little has been written on this subject except in Great Britain. Two German and Austrian references, and five American ones (up to February, 1902) are all that could be discovered in the magazine literature of these countries. From French sources I could get nothing. Neumann in 1899 wrote, "This form of idiocy is nowhere mentioned in German literature," but states that it is better known in England. Many of the observations published differ not only on minor but on important points, the difference being due, in some instances, to confusion of this condition with cretinism.

NOMENCLATURE.—In 1866 Dr. Langdon-Down suggested the term "Mongol" or "Kalmuc" to describe a genus of congenital imbecility the members of which presented certain physiognomical resemblances to the members of this ethnological group. Although his classification of the varieties of mental feebleness has now been abandoned, the term "Mongol" has survived, being an expressive and descriptive one.

Several definitions more or less unsatisfactory have been offered of this condition. They have been called "furfuraceous cretins." They have been classified under "simple congenital cases without any other defect of skull and limbs," although

* An abstract of a thesis for the degree of M.D., Edinburgh. The cases were for the most part met with in the out-patient clinics of Drs. Still and Hutchison, Great Ormond Street Hospital for Children, London.
brachycephaly, or, at any rate, a diminution of the normal difference between the lengths of the anteroposterior and transverse diameters of the cranium, is one of the constant features.

Mongolism is different from all the other congenital types of mental feebleness such as cretinism, congenital hydrocephaly, microcephaly and the birth palsies. By some they are ranked as imbeciles, by others as idiots. My own cases varied much, some being of a distinctly low grade, though others again are the opposite. The general term "mental feebleness" so much used in America seems more satisfactory. Mongolism might perhaps be defined as a type of mental feebleness, always congenital in origin, characterized by certain constant cranial, and later by lingual, changes.

The geographical distribution seems to be that of the Caucasian race. My own cases represent London (and its suburbs such as Poplar, Ponders End, Homerton, Woolwich, Leyton, Hampstead, Tottenham, Edmonton) Kingston, Harrow, etc. I have seen cases in Liverpool, Edinburgh, and in Sydney, New South Wales. One case came from Magdeburg, Germany, in which country Neumann,23 the only German writer who has given attention to the subject saw 13 cases in three or four years; and another from Roumania. Carl Looft21 reports cases in Norway, and it is known in Austria. Lombruso long ago gave an account of one in Italy. I have seen a case in South Africa; Dr. Ireland9 mentions one from Australia; and another which I saw came from Bermuda. Thus mongolism probably exists everywhere; it is found in the country and in the towns, and no white race is exempt. Two of my cases were among Jews. It seems to be less common in Scotland (3 per cent. of all mentally feeble children) than in England (5 to 10 per cent.) 1-18. It is a fairly common condition in all large out-patient clinics in London, and is at least four times as common as cretinism. In America West39 saw 9 cases in four years.

Etiology.—A history of drunkenness in the parents was found in 4 cases, of consanguinity in none. All the children were born in wedlock. Some children were born and had always lived in the country, in others the environment was bad, so that no special connection was traceable. In 4 cases there was a family history of insanity or eccentricity; in 3 the father was violent in temper; in 1 the father committed suicide; in 4 the mother was neurotic and excitable (in 1 case with suicidal impulses). In
3 cases there were twins or triplets in the family; in 3 the intrauterine movements were hardly felt; in 3 the mongol was premature; in 1 a previous child was anencephalous; in 8 the labor was prolonged or complicated; and in about half the cases a statement of fright or worry was volunteered.

Langdon-Down¹⁸ says that mongolism arises chiefly from tuberculosis in the parents, but this occurred in only 38 per cent. of my cases.

Sutherland⁴ found evidence of syphilis in 11 out of 25 cases, and suspected it in three others. Still⁷ found it only once in his cases, and it was present in only 3 of mine.

Much stress has been laid on bad health on the part of the mother during pregnancy. This occurred in only 15 of my series. In the remainder it was perfect. In 1 case the mother had two healthy children and then a mongol, her health being better during the last pregnancy than during the former. In proof of this theory it has been stated that mongols are often the last of a large family, 40 per cent. according to Shuttleworth,¹⁶ 10 out of 18 cases according to Still. But 3 of my cases were first-born, 7 were third, 1 fourth, 6 fifth, 1 sixth, 2 seventh, 2 eighth, 1 ninth, 1 tenth and 1 twelfth. In the cases of all the first-born children, in the sixth-born case and in one third-born case, healthy children have been born since. In many cases, however, the parents were over forty years of age. As regards causation clinical evidence has helped us very little.

Sex.—In my own series there were fifteen males and eleven females. Of 80 cases from all sources 56 per cent. were males and 44 per cent. females.

The Symptoms are present from birth, a point insisted on by Dr. John Thomson,⁶ and confirmed by 2 of my cases in a remarkable way. In one an aunt remarked when the child was born, “Oh! Isn’t she like a Chinese baby.” When the other was eight weeks old the grandfather said “Well, baby, you didn’t have to go to Japan for your eyes.” Thus in well marked cases the peculiar physiognomy attracts the attention even of the laity. The trunk and limbs though small are well formed. Those features which are characteristic are found in the skull, eyes, tongue and hands.

The Skull.—The occipitofrontal circumference is almost always diminished, the average diminution being 1.3 inches, and practically the same in both boys and girls. It varied from nil
to 2.25 inches, which was the maximum. My youngest patients were two, aged two months with circumferences of 14 inches and 15 inches respectively. This compares fairly with the normal birth circumference, viz., 14 inches.

Brachycephaly—a shortening of the normal anteroposterior diameter—is always present in typical cases. I was unable to find in any language statistics showing the normal measurements of this and the transverse diameters of the cranium in children. The average length of the anteroposterior diameter in seven mongols all aged about two and a half years is $\frac{511}{14}$ inches; in fifty healthy children $\frac{64}{7}$ inches; difference $\frac{7}{1}$ inch. The Mongolian transverse diameter averages 5 inches, the normal is $\frac{53}{4}$ inches. In all except one case the occiput was flattened and steep, but no actual depression such as Sutherland mentions was ever present.

The Fontanels remain open late, the latest in my series was four years, but one has been reported open at four years and nine months.$^7$ From my cases the average date of union is about $2\frac{1}{2}$ years of age. The posterior fontanel was often open at the fourth, fifth and even the tenth months. The union of the lambdoidal, coronal, and sagittal sutures was also delayed.

The Face is flat and often depressed as a whole. The lower part of the forehead is then less prominent and on a plane posterior to that of the upper. The complexion is, as a general rule, good, except in the early weeks of life, or when the child is of the thick skinned strumous type. The other common facial peculiarities have already been described in Archives of Pediatrics.

The Eyes.—Strabismus, always convergent and concomitant, occurred in 7 cases. Nystagmus was present five times. Sutherland states that both these symptoms tend to disappear after the first six months, which does not agree with my experience. In 3 cases strabismus appeared in infancy and persisted, and in 2 others it appeared first at eighteen and twenty-six months. In only one case of squint or nystagmus dating from birth did it pass off later. Ametropia is common. I found no noteworthy fundal changes even where there was nystagmus. Ophthalmia tarsi has been described as almost universal, but was seen in only 2 cases. When present it was in older children with marked tuberculous history. Epicanthus occurred seven times, and these children were much more Mongolian looking than those without it. Actual measurements of all cases seemed to show that there is very little increase in the distance between the eyes, although the broad
nasal bridge, and epicanthus, when present, give this appearance. Dr. John Thomson remarks that the eyes are often rather near one another, but I have not met with such a case.

Another question that is worth considering is whether the causes of the obliquity of the eyes in mongolism and in the Mongolians are identical in both? Komoto states that the difference between the Mongolian and Caucasian eye rests solely on the epicanthus which exists physiologically among the Chinese and Japanese. In mongolism, however, the obliquity of the palpebral fissure is present even where there is no epicanthus, and is due to skeletal peculiarities. If Komoto is correct, the causes are different. (Quoted Jour. Amer. Med. Assoc., Vol. XVIII., 1892, p. 361).

The Mouth was usually kept open, and in low and medium grade cases there was dribbling of saliva.

The Tongue was never much enlarged, and never more than slightly protruded. The peculiar and characteristic hypertrophy of the fungiform papillae which is followed by fissuring of the surface of the tongue was always present after a certain age and was not seen in any other form of mental feebleness examined. The earliest age at which fissuring was present was twenty-two months. From notes of other cases, I find the following data:—In 8 under 12 months, no hypertrophy nor fissuring. In 2, aged 24 and 26 months, no hypertrophy nor fissuring. In 3, aged 24, 26 and 30 months, hypertrophy commencing, no fissures. In 1, aged 26.5 months, both were present. In 6, ranging from 2.5 to 11 years in age, both were present.

Palatal deformities were present in 65 per cent. of my cases. In seven the palate was high and narrow; in two, high; in one, high with a median ridge; in one, narrow but not high; in two, broad and approaching the circular type; in four, it was "fair," and in the remainder normal.

Rhinitis was common, and adenoids almost invariably present. The ears were often deficient in shape, but in no case were they set too far back.

The Teeth appear late and irregularly. In one case the lower incisors came at 10 months, in another the upper central incisors at 9 months, and these were the earliest I saw. The lower incisors appeared at from 16 to 26 months, the upper incisors at from 17 to 19 months, the right upper molars at from 14 to 18.5 months, and the left upper molar at from 14 to 23 months. One child, of
26 months, had only four teeth; two, of 22 and 30 months, had six. The lower central incisors in one-half of the cases appear first. In many the upper anterior molars come first, or the upper central incisors. The second dentition is also delayed, so that the six-year-old molars may appear two years later. The teeth decay early, and at twenty-six months may all be bad. In other cases they remain perfectly good.

The Hair may be of any color, but is more often light than dark. In infancy it is usually soft and in most cases abundant, but in some cases it is long, fine, dry and standing straight up from the head. In only 5 out of 18 cases could it be fairly described as scanty.

Skin.—Except in one case the skin was soft, and only furfuraceous on the cheeks.

Physical Development.—In 55 per cent. these children were weak from birth, my own experience confirming Sir A. Mitchell's in this respect. The stature was on an average 3½ inches less than normal. The decrease was less in boys than girls, as 23:25. In 4 cases the mongols were .4 to .5 inch above the average height for their ages. The average diminution in weight was six pounds, nine ounces.

A passage in Ireland's book suggested to me to make comparative measurements of the lengths of the upper and forearms. In 1 case they were equal, in another the difference was three eighths of an inch, but in the others no alteration was ascertainable. In the fetal condition up to the fiftieth day and in apes the forearm is longer than the upper arm, in man the reverse is true.

The Thumb and little fingers are relatively very short, and the second, third and fourth fingers often about the same length. The tips are tapering, not square. The peculiar outward curving of the little finger, described by Telford Smith in Archives of Pediatrics in 1896, as being almost distinctive, was present only twelve times, and I found it in microcephaly, cretinism and healthy children. In 9 it was quite well marked, in 3 it was slight and could easily have been overlooked, and in 13 it was entirely absent. Since this was written J. P. West in America has arrived at the same conclusion.

The laxity of the ligaments was in some cases very marked. One child could easily do the stage trick known as the "splits," and another used to hold its leg upright against the chest and face like a doll. Congenital morbus ceruleus occurred five times,
and in one case showed considerable improvement later on. Constipation was complained of in 28 per cent. of cases; mucous disease (of Eustace Smith) in two, and diarrhea in others.

**Mental Condition.**—In 14 cases the cerebral and mental condition was one of idiocy of a low grade; in four of medium, and in seven of a high grade. Fifty-five per cent. gave some sort of warning, usually a grunt or other sign before defecation, less frequently before micturition. This was present as early as the tenth month, but in another no warning was given at two and one quarter years.

*Speech* came late. Five children between the ages of twenty-six months and four years could not speak at all. One at twenty-six months could only say “Dada,” and two at eighteen and twenty-four months could call the cat by name. The development of speech is in the same order as in healthy children; first “mama,” “tata,” “papa,” then the names of persons, then the names of things, and next a few verbs. My most accomplished mongol got no further than this. No abnormal conditions of moment were found in the sensory or motor functions. The extensor reflex of the foot which disappears usually about the end of the first year persisted somewhat longer. The pyramids therefore probably develop late.

**The Temperature** of the body is often subnormal ranging from 96.0° to 97.3° F.

**Symptoms of Degeneration.**—Almost every conceivable malformation and stigma of degeneration are found among mongols. In addition to those mentioned I saw harelip, contraction of the palmar fascia, webbing of the toes (2 cases), supernumerary toes, umbilical hernia, naevi (thrice), cryptorchidism, phimosis, etc. The literature contains mention of others such as spina bifida occulta, congenital club-foot, imperforate anus, etc.

**Diagnosis.**—These children are usually brought by the parents for intestinal disturbance, nystagmus, inability to hold the head up, or “weakness of the back.” Clinically the diagnosis is usually easy and always so in a well marked case. Some normal children have a facies something like that of mongolism. Syphilis may occasionally cause difficulty. Cretinism, however, is usually the source of error. In mongolism we find tapering fingers, the little finger sometimes curved, brachycephaly, obliquely set eyes, pink cheeks, sometimes epicanthus, sometimes congenital heart disease, thyroid gland normal, no local deposits of
fat, peculiar appearances on the tongue, stature below normal but the body well formed, and the symptoms present from birth. In cretinism we find on the other hand, the finger tips square, the little finger rarely curved, nothing special about the shape of the skull, the eyes not oblique, coarse features, epicanthus absent, congenital heart disease very rare, thyroid gland not palpable, edema of whole body, fat deposits frequent, large protruding tongue, bodily deformities frequent, and the symptoms do not appear until the sixth month (Thomson). Cretins are more lethargic, and are benefited by the thyroid treatment. Mongols are not so.

The Prognosis varies but is usually bad with respect to the mental condition. Some of my higher grade cases could feed themselves. A mongol may not hold up his head until two and a half years, may not sit up until twelve to twenty-eight months, may not walk until three and one half years or later, and may not feed himself until four or five years; in bad cases not even then.

My best educated mongol knew his name, and could spell "Monday," and also knew the year and month. He could spell or read no other words, and what he did say was spoken in a parrot-like way. He knew some of the letters and all the figures. Even this one was refused admission to the special classes of the London School Board for Backward Children.

The prognosis as to life is still worse. The parents often told me these children bore the winter badly. One of my cases died of congenital heart disease; one of bronchopneumonia, and one of tubercular meningitis. Most die of pulmonary tuberculosis. Neumann mentions that of his 13 cases, only 6 are now living.

I have not yet had the good fortune to make a postmortem examination of a mongol, but have been able to collect the results of eleven cases. All are agreed that there is an anteroposterior shortening at the base of the skull, but nothing else (up to February, 1902) has been discovered of importance. A summary of these examinations shows that there is no evidence in favor of the premature primary ossification of the os trigonale, but that there is an arrested development of structures at the base of the brain followed by premature osseous union. The autopsy work done by Wilmarth in America on 5 cases points to the latter theory as well as that of other observers in London.

I never found in life any backward extension of the vomer
into the nasopharynx such as was found postmortem in 1 case by Still.  

TREATMENT has availed nothing in my hands. The general condition sometimes improves under malt and oil, but thyroid treatment was useless. Carl Looft found thymus gland treatment was of no benefit.

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