PELVIC ABNORMALITIES IN MONGOLS

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Caffey and Ross (1956) first observed that the radiological appearances of the pelvis in mongols were abnormal. Kaufmann and Pelargonio (1957) confirmed this original observation, which was later extended to include the detailed study of more subjects (Caffey and Ross, 1958). This paper describes our findings in a further series of 57 mongols whom we have examined radiologically since 1956. During this period of observation four infant mongols died and went to necropsy. The data which were obtained demonstrate that characteristic anatomical configurations of the pelvic girdle appear frequently in these individuals. The superior iliac spine is absent and the wing of the os ilium is widely splayed, causing a decrease in the cephalo-caudal diameter (Fig. 1). The acetabular and iliac angles are decreased and there is a low iliac index (Fig. 2). The iliac index (I.I.) is defined as the sum of

FIG. 2.—Schematic drawing method of measuring the acetabular and iliac angles. Both angles are based on Hilgenreiner's line extending through both Y-cartilages.

both acetabular angles (A.A.) and both iliac angles (I.A.) divided by two. The ramus of the ischium is abnormal in length and tapers at the distal end, while in infants of 9 months or more the femur often shows a valgus deformity.

X-ray Examinations

Our series of 57 mongols was divided into two groups and these were studied separately. (1) Infants: The methods described by Caffey and Ross (1956) were

employed in the examination of the 43 x-ray films obtained from 34 mongols under 1 year of age. Measurements of the A.A. and the I.A. were made and the I.I. was calculated. (2) Older infants, children, and young adults: In this group were 23 mongols, aged 1 to 22 years, who, apart from their basic disorder, were essentially healthy. Most of the older ones were attending the local training school for the mentally handicapped, though some were living at home.

Necropsy Examinations

The post-mortem findings in the four mongols who died were compared with those in seven normal newborn babies or infants of similar ages. The mongols were aged 1 day, 3 weeks, 3½ and 6 months at the time of death. Particular attention was given to the bones of the pelvis, the lumbar region of the spinal column, and the femora. Specimens were preserved in 4% formalin and radiographs of the pelvis were taken both before and after the injection of 20% lipiodol into both hip-joints. The exact positions of the pelvic bones relative to one another were studied carefully, attention being focused on the same fixed points in different views.

Results

Of the 34 infants (Group 1) examined, three had been born prematurely. Their birth weights were 1,270 g., 1,630 g., and 1,900 g., and their respective measurements were: A.A., 15, 9, and 9 degrees; I.A., 45, 40, and 43 degrees; I.I., 60, 49, and 52. The data presented by the entire group of 34 infants were in full agreement with the characteristic patterns that have already been described. The mean values revealed were A.A. 14 degrees, I.A. 42 degrees, and I.I. 50, compared with the average figure, A.A. 25–28 degrees, I.A. 55–58 degrees, and I.I. 79–81 for the normal infants.

In the second group the same characteristic changes were evident. In one child who was radiographed at 2 years of age and again at 11 years the abnormalities had neither changed in character nor progressed in degree.

The radiological appearances of the four necropsy specimens demonstrated the same anomalies that had been observed in living subjects, and differed in appearance from the necropsy specimens of normal infants. The arthrograms did not reveal any abnormality.

The changes that have been noted are well illustrated in the photographs (Fig. 3). The pelvis of the mongol is contrasted with the normal pelvis in the antero-posterior view (Fig. 3 A), in the lateral view (Fig. 3 B), and in the cephalo-caudal view (Fig. 3 C). In the antero-posterior view the ilium has wide laterally flared wings with a reduced cephalo-caudal projection. The

FIG. 1.—Characteristic x-ray appearance of the "mongol pelvis" at different ages. A, 1 day old; B, 1½ years old; and C, 16 years old.

superior iliac spine is absent, and the ischium appears to be elongated. In the other views, also, it is the ilium which appears to be abnormal.

Discussion

Our studies confirm that radiological changes are seen in the pelvis of 4 out of 5 mongols. These anomalies are characteristic of both premature and newborn mongol infants, and are of particular diagnostic significance. The appearances persist into adult life, and further study of x-ray films in adult mongols is obviously indicated. It appears that all who have investigated this problem agree that the x-ray changes are diagnostic of mongolism, and it seems justifiable to postulate the concept of a true "mongol pelvis."

We have evidence that adult mongols can develop dislocation of the hip, and have observed three cases in 18 mongols over 5 years of age. This finding is perplexing, because the appearances in the "mongol pelvis" are the opposite of those seen in congenital dislocation of the hip. We consider that the dislocation of the hip observed in the mongol patient is of a secondary nature, such as that which follows in patients with paralytic poliomyelitis, spina bifida, and spastic diplegia or paraplegia.

We believe that the radiological and anatomical findings could be due to a developmental anomaly of the pelvic girdle which affects the configuration of the ilium, but the possibility that an abnormality of form due to irregular growth as an additional factor cannot be excluded. Further studies are to be undertaken in order to make this point clearer. It is our opinion that the well-known generalized hypotonic musculature in mongolism does not contribute towards the development of these abnormalities. The fact that we have noted two cases of amyotonia congenita in which the x-ray film of the

pelvis was completely normal lends support to this view. In discussing the possible causes of these pelvic changes we must consider other congenital abnormalities which have been reported in the mongol. Lejeune *et al.* (1959) and Jacobs *et al.* (1959) demonstrated an extra autosomal chromosome pattern in mongols. These findings have been confirmed by Ford *et al.* (1959) and by Böök *et al.* (1959). It may be that the presence of an extra chromosome is instrumental in causing the pelvic manifestations which we have emphasized. The means by which this can be brought about is unknown, but it may be of value to investigate the appearance of the pelvis on x-ray examination in other conditions in which an abnormal chromosomal composition is established.

Summary

Characteristic variations from the normal which were observed in the x-ray films of the pelvic bones of over 60 mongols are reviewed. These changes are constant from birth onwards and persist throughout life without signs of any progression in severity.

In three adult mongols dislocation of the hip, which is considered to be a secondary phenomenon, is recorded.

Anatomical studies establish the precise nature of the pelvic anomaly and indicate that the abnormal appearances are due particularly to the configuration of the os ilium.

REFERENCES

- Böök, J. A., Fraccaro, M., and Lindsten, J. (1959). *Acta paediat. (Uppsala)*, **48**, 453.
Caffey, J., and Ross, S. (1956). *Pediatrics*, **17**, 642.
— (1958). *Amer. J. Roentgenol.*, **80**, 458.
Ford, C. E., Jones, K. W., Miller, O. J., Mittwoch, U., Penrose, L. S., Ridler, M., and Shapiro, A. (1959). *Lancet*, **1**, 709.
Jacobs, P. A., Baikie, A. G., Court Brown, W. M., and Strong, J. A. (1959). *Ibid.*, **1**, 710.
Kaufmann, H. J., and Pelargonio, S. (1957). *Schweiz. med. Wschr.*, **87**, 1529.
Lejeune, J., Gautier, M., and Turpin, R. (1959). *C.R. Acad. Sci. (Paris)*, **248**, 602, 1721.

FIG. 3.—Anatomical configuration of the pelvis of a 3-week-old mongol patient removed at necropsy contrasted with a normal pelvis. A, Antero-posterior view. B, Lateral view. C, cephalo-caudal view. Upper row: Normal pelvis. Lower row: "Mongol pelvis." Note: in A and C the opening of the wings of the ilium causing the broader appearance, visible on the x-ray films; and in B the angulation and downward slope of the iliac wing in the "mongol pelvis."