Summary

1. An attempt has been made to give brief practical details concerning the estimation of the blood pressure both in adults and in children.
2. The interpretation of the results have been given.
3. The difficulties in the estimation have been tabulated and discussed.
4. Reference to some of the important literature has been freely used.

REFERENCES

1. Joint Report, *British Heart Journal*, 1939, 1, 263.
2. KOROTKOW, N. (1905), *Mitt. Kaiserl. milit-mediz. Akad. St. Petersburg*, 11.
3. BRAMWELL, C., and KING, J. T. (1942), *The Principles and Practice of Cardiology*.
4. TREADGOLD, H. A. (1933), *Lancet*, 1, 733.

DEVELOPMENTAL ABNORMALITIES OF CERVICAL VERTEBRAE IN A CASE OF GENERALISED NEUROFIBROMATOSIS

By ALBERT PEACOCK, M.B., B.S.(LOND.), M.R.C.S., L.R.C.P.
(*Demonstrator in Anatomy, London Hospital Medical College.*)

The skeletal abnormalities described below were present in a patient suffering from generalised neurofibromatosis, who was admitted to the London Hospital in October 1943. The salient clinical features were as follows:—

History

The patient was a male of 49 years, a labourer by occupation. He had long standing cutaneous lesions on face, neck, trunk and limbs and complained of loss of weight over the previous 6 months. Seven days prior to admission to hospital, he complained of difficulty in speaking, swallowing and breathing and of the simultaneous and rather sudden appearance of a swelling in the left side of the neck. The character of the voice changed, swallowing became progressively difficult and the patient was admitted on account of difficulty in breathing.

The previous history was of minor illness only and nothing relevant to his present condition. In the family history, there was no mention of generalised neurofibromatosis.

Physical Examination

The patient was rather emaciated. Characteristic cutaneous lesions of generalised neurofibromatosis, both sessile and pedunculated, were present practically over the whole body. On the left side of the neck, a large firm mass was present projecting from beneath the angle of the jaw and deep to the sternomastoid. On examination from the mouth, the mass was seen to bulge into the left side of the pharynx almost halfway across that cavity, measuring about 2 in. in the longitudinal and $1\frac{1}{2}$ in. in the transverse axis respectively. The trachea was displaced to the right. No enlarged lymphatic glands were discovered and the thyroid appeared to be normal. On examination of the chest, slight emphysema was found to be present. No cardiac signs were found. The blood pressure was 120/70. Nothing abnormal was found in the abdomen. The patient exhibited a left lateral rectus ocular palsy. The arms showed wasting, but muscular tone was normal and the tendon jerks were diminished but equal on the two sides. No abnormality was seen in the legs and the tendon jerks were present and normal.

The patient complained of some coldness of the hands but there were no other symptoms present. No sensory disturbance was detected.

Accessory Examinations

Cerebrospinal fluid	..	20 c.c. of clear colourless fluid was withdrawn. There was less than one white cell per c.mm.
		Protein = 20 mgm. per 100 c.c.
Blood Urea	.. ..	188 mgm. per 100 c.c.
Wassermann Reaction		Negative
Kahn Test	.. ..	Negative.

Operation

During induction of anaesthesia, respiration ceased on account of obstruction and laryngotomy was performed. Following this, anaesthesia proceeded normally and a collar incision was made below the mandible on the left side. The wound was deepened and the internal jugular vein divided between ligatures. A large firm tumour was removed, which had the appearance of a sarcoma. The operation wound was closed with drainage. On the following day, the respiration fell to 4 per minute, and the patient was cyanosed and pulseless. Coramine was administered and a Bragg-Paul apparatus was applied. The patient improved somewhat but some 3 days later he suddenly collapsed and died.

Autopsy

This revealed collapse of lungs. Neurofibromatous lesions were discovered on left cervical sympathetic and in the upper left thoracic sympathetic and extradural lesions on 2nd sacral nerve root and on several spinal nerves. No abnormalities were found on the cranial nerves or brain. Microscopical examination of the tumour of the neck revealed a spindle and polymorphic celled neurofibrosarcoma. An abnormality was noticed in the cervical vertebral column and part of the base of the skull, surrounding the foramen magnum, together with the upper three cervical vertebrae, was removed for further investigation.

Detailed Examination of Abnormal Cervical Spine

(1) **The Undissected Specimen.** Examined from above, the dura mater was seen to be raised and was projected for 2 cm. into the cranial cavity above the level of the foramen magnum, by the underlying odontoid process. Behind this, the cervical portion of the spinal cord was seen, and the spinal root of the accessory nerve and the vertebral artery on both sides.

(2) **On Dissection** the posterior arch of the atlas on the right side was found to be imperfectly developed. It tapered to a point and failed to reach the posterior aspect of the atlantal lateral mass, the gap being bridged by a fibrous membrane which appeared to be a continuation of the posterior occipito-atlantal ligament.

Disarticulation of the skull and atlas was next performed; the joint capsules of the occipito-atlantal joints were present, as were the transverse and check ligaments and anterior occipito-atlantal ligament. Between the anterior surface of the odontoid, and the posterior aspect of the right half of the anterior arch of the atlas, passing up to the skull base, was a mass of firm greyish tissue which had to be divided to permit the separation of the skull and atlas.

Examination of the skull base revealed the occipital condyles which were seen to be asymmetrical. The right one though almost bisected by a saw cut, was elongated and had an indented medial margin. It was on a plane posterior to the left condyle. The left condyle was shortened antero-posteriorly, the anterior portion being turned upwards and facing anteriorly to articulate with the corresponding articular surface on the atlas. To the medial side of the right condyle was seen the firm mass described above, which impinged on the basi-occiput, the base of which was very thin. This mass was quite invisible from the exterior and could not be detected during the ordinary routine autopsy. It was only visible on separation of the skull from the atlas.

(3) **Maceration** of the vertebrae was next carried out.

Examination of the fused vertebrae revealed that the atlas, axis and the 3rd cervical vertebra were fused together, the fusion having occurred mainly in the region of the articular processes. The composite mass was asymmetrical, the right side being advanced anteriorly.

The Atlas showed a well-developed anterior arch, bearing an anterior tubercle. The upper articular processes were widely separated, the right approached a normal appearance, but the left was placed on the posterior aspect of the anterior arch, was almost circular in shape and faced directly backwards. No facet was present for the odontoid process, a median elevation taking its place. The posterior atlantal arch (as far as could be seen from the specimen), was well developed on the left side, a median posterior tubercle was present, while the right half was imperfectly developed as described above.

The Axis showed marked asymmetry. No facet was present on the anterior surface of the odontoid, which was 1.5 cm. posterior to the anterior atlantal arch. A groove for the transverse ligament was seen at the root of the odontoid.

The 3rd Cervical Vertebra also showed marked asymmetry of the body; on the right side, fusion with the axis was seen, but the articular pillar was absent on this side. On the left side, the articular pillar was poorly developed and projected downwards and forwards, the superior articular facet was fused with the inferior corresponding facet of the axis, while the inferior articular facet was small.

In reviewing the abnormality of the cervical spine, certain features require comment. The adaptation of the medulla oblongata to the abnormal position of the odontoid is of interest, and of the intraspinal manifestation of the neoplastic tissue, it may be noted that pressure on the medulla was prevented by the odontoid and right side of the body of the axis bounding the mass posteriorly. In life, limitation of movement would be marked. The effects of modification of the cervical vertebral column and occipital condyles would be, complete absence of rotation at the atlanto-axial joint and limitation of flexion and extension, and more particularly flexion, at the occipito-atlantal joint, and absence of movement between the axis and the 3rd cervical vertebra.

An extensive anatomical literature exists in reference to osteological abnormalities in this region and reference to monographs must be made for details.

It may be remarked that abnormalities at the atlanto-axial joint are particularly concerned with the question of the vertebralisation of the occiput, and occipitalisation of the atlas, dependent upon singular embryological segmentation of the occipito-atlantal region.

Fusion of the atlas and axis is very rare, and of this Cave states that "non-pathological fusion of atlas and axis constitutes a very rare anomaly, perhaps the rarest of all the many and varied manifestations of irregular segmentation affecting the vertebral axis." Macalister considered "atlanto-axial ossification and ankylosis as always the result of disease" and as Cave points out this probably resulted in examples of developmental fusion being overlooked in the past. Elliot Smith commented on this rarity and in his cases he discovered no pathological changes and he considered the fusion to be due to "developmental eccentricity."

Fusion of the 2nd and 3rd and sometimes the 4th cervical vertebrae is very common and not infrequently there is congenital maldevelopment of the 3rd cervical vertebrae present in these instances. Dwight commented on the fusion of the 2nd and 3rd and considered this area as one of the regions of the column exhibiting a predilection for fusion. Leboucq was in agreement with this and described two cases and cited Murie to the effect that reduction of cervical vertebrae in the Manatus arose from an almost complete regression of the 3rd cervical vertebra. Dwight considered this as a "critical point" and stressed the fact that it was "fusion not mixing" of vertebrae in the cases described.

Synostosis of the 2nd and 3rd cervical vertebrae are referred to by Brash and Barclay Smith describes an example of this in a case of 8 true cervical vertebrae. Fusion however is described in cervical regions of normal number by Rokitsky, Goodhart, Arbuthnot Lane, Dukes and Owen.

As regards incomplete development of the posterior atlantal arch, this is not frequent but not excessively rare. Macalister described a median and a lateral deficiency, stressing the fact that lateral deficiency is less common. Ledouble described a number of cases of partial absence of the posterior arch, which he considered as due to arrested development of the centre of ossification forming the posterior arch, lateral mass and posterior half of the transverse roots of the first cervical body.

A feature of a number of the abnormalities in the region of the occiput and atlas, and atlas and axis described was the extraordinary degree of compensatory modification of the articular surfaces, to overcome the limitation of movements imposed by these conditions.

In most of the described cases there was no known history to the spines examined and no information of any or other associated developmental diseases.

The specimen here described is remarkable for the fact that fusion affects the three upper cervical vertebrae, that the configuration of the vertebrae is not obscured, thus showing no assimilation of one vertebra by another, or assimilation of the atlas by the occiput, further that there is a marked lack of compensatory modification of the atlantal or the occipital condyles to overcome the restriction imposed by maldevelopment. Moreover, the abnormality is known to be associated with a disease which is considered to have a developmental abnormality as its basis.

The osseous conditions associated with generalised neurofibromatosis fall into two main groups. In one, congenital skeletal defects occur. Kyphoscoliosis is frequently present, spina bifida and defects in the bony wall of the orbit are described. In the other, there are osseous manifestations directly due to the disease, exemplified by the results of periosteal lesions as described by Parkes Weber resulting in overgrowth of bony tissue and the enlargement of bones seen in various situations. It has been suggested that the curious lesions that were present in the case of Merrick, the "Elephant Man" were of a neurofibromatous nature.

In conclusion it may be stated that no such changes are seen in the specimen described, and in the absence of pathological inflammatory or of neoplastic changes, it may be considered as a rare example of the congenital skeletal abnormalities associated with generalised neurofibromatosis.

I am indebted to Professor H. M. Turnbull and Dr. W. W. Woods for the specimen and pathological findings, to Mr. G. Neligan for clinical information and to Professor J. D. Boyd and Professor A. J. E. Cave for their help and advice.

REFERENCES

- ANZINGER, F. P., *Journal of Heredity*, 1933, Vol. XXIV, pp. 309-312.
 BATESON, W., *Materials for the Study of Variation*. 1894.
 BROOKS, B., LEHMAN, E. P., *Surg. Gynec. Obst.*, 1924, Vol. XXXVIII, pp. 587-595.
 CAVE, A. J., *Journ. Anat.*, 1930, Vol. LXIV, pp. 337, 343.
 COCKAYNE, E., "Inherited Abnormalities of the Skin and its Appendages," 1933.
 DWIGHT, T., *Anat. Anzeiger*, 1901, Bd. XIX, p. 331.
 ELLIOT SMITH, G., *Anat. Anzeiger*, 1907, Bd. XXXI, s. 166.
 ELLIOT SMITH, G., *Anat. Anzeiger*, 1909, Bd. XXXIV, s. 357.
 GLADSTONE, A. E. & ERICKSEN-POWELL, J., *Anat. & Phys.*, Vol. XLIX (3rd Series, Vol. X), 1915, pp. 190-209.
 GOULD, E. P., *Quart. J. Med.*, 1918, Vol. XI, pp. 221.
 GRAY, S. H., *Arch. of Neur. & Psych.*, Chicago, 1929, Vol. XXII, p. 91.
 HARROWER, G., *J. Anat.*, 1922, Vol. LVII, p. 173.
 HUBBLE, D., ROGERSON, C. H., *B.M.J.*, 1944, April 8th, No. 4344.
 HUNTER, R. H., *J. Anat.*, Vol. LVIII, pp. 140-141.
 LEBOUCC, *Arch. de Biologie*, 1898 (Quoted by Dwight).
 LE DOUBLE, A. F., *Trasie des variations de la colonne vertebrale de l'homme*.
 LEVIN, O. L., BEHRMAN, A. T., *Arch. Derm. & Syph.*, Chicago, 1940, Vol. 41, No. 3, pp. 450-502.
 MACALISTER, A. J., *Anat. & Phys.*, Vol. III, pp. 54-64.
 MACALISTER, A. J., *Anat. & Phys.*, Vol. XXVIII, pp. 257-268.
 MACALISTER, A. J., *Anat. & Phys.*, Vol. XXVII, pp. 519-542.
 WEBER, F. P., *Quart. J. Med.*, 1930, Vol. XXIII, p. 151.