CASE REPORTS

CONGENITAL HEMI-ATROPHY

BY

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Not much detailed information is available as to the causes and origin of structural asymmetries. Congenital hemi-atrophy is a rare developmental abnormality in which parts or all of one side of body are of diminished size. As a rule all tissues are affected to a similar extent. The condition should not be confounded with the unilateral atrophy associated with motor disorders such as unilateral paralysis and unilateral athetosis.

In 1927, in a survey of world literature, starting from 1859, Kraus and Perkins found only eight cases of congenital hemi-atrophy in five of which the whole side of the body was affected. The other three were confined to the leg in two cases, and to the face and leg in one case. More cases have been reported since, and in 1939, in another survey of world literature, Landauer found twenty-two cases of congenital hemi-atrophy, eight of which only showed an affection of the whole side of the body.

Congenital hemi-hypertrophy is, in many ways, the counterpart to congenital hemi-atrophy. Although also a rare condition, more cases of hemi-hypertrophy have been reported than of hemi-atrophy. Both conditions are sometimes associated with retardation in mental development and with vasculo-cutaneous disturbances, but these are met with much less frequently in unilateral atrophy than in unilateral hypertrophy. Perhaps these two latter affections are not caused by two different agents, but by either hypo- or hyperfunction of the same agent respectively.

The mechanism which brings about preferential asymmetry seems to act during a comparatively early stage of embryonic life. Probably the agency responsible for developmental asymmetry must be looked for within the developing embryo, and not in conditions of the uterine environment, as so many different structures are affected. Various authors assume that the central nervous system has a trophic function in development and that thus unequal differentiation of cerebral centres may modify growth asymmetrically. Kraus and Perkins hold the view that cerebral portions of the visceral nervous system exert a trophic control not only on glands and smooth muscle but also on striated muscle, tendons, bone, skin, hair, in short, on all tissues of the body. They assume that these centres are located in the hypothalamic region in or about the floor of the third ventricle, as well as in the cell

groups lying about the tuber cinereum. Furthermore, it is now also generally believed that from cell groups in the posterior part of the hypothalamus a directing influence is exerted over the function and trophic activities of the sympathetic system. On the other hand, evidence has been obtained that dysfunction of the sympathetic can be the cause of unilateral atrophy. Cases of facial hemi-atrophy have been reported associated with changes in the sympathetic on the affected side.

From this point of view it may be of interest to discuss some conditions which are frequently associated with unilateral atrophy. There are, to begin with, two clinical conditions in which hemi-atrophy is frequently met with, chondrodysplasia (Ollier's disease) and scleroderma. With regard to both these affections theories have been brought forward which assume that dysfunction of the sympathetic plays an important part in their pathogenesis. Furthermore, various instances of progressive (non-congenital) facial hemi-atrophy have been reported which show a definite relation to changes in the sympathetic system on the affected side. There are, lastly, various instances of total or partial hemi-hypertrophy associated with vasculo-cutaneous abnormalities, and which have been attributed to unbalanced action of the sympathetic.

Of these conditions Ollier's disease will be more fully described here as the case reported below showed some bone changes similar to those seen in chondrodysplasia.

Chondrodysplasia (Ollier's disease)

Chondrodysplasia is an osseous dystrophy affecting the long bones and the metacarpophalangeal skeleton. In some cases the vertebral bodies have also been found to be affected. (The osseous changes can be demonstrated by skiagram.) It is an affection of growth with arrest of growing parts of the skeleton. Ollier (1899) emphasized as an outstanding clinical character of the disease an asymmetrical involvement of the body, a 'one-sidedness.' One of the essential features of the disease is the retardation of growth of one or both limbs on the affected side, sometimes associated with compensatory scoliosis. The term 'Ollier's disease' has now been confined by a number of authors to those cases of osseous dystrophy which show an asymmetrical involvement of the body as an outstanding clinical feature.

Chondrodysplasia usually starts in early life. Cleveland (1928) describes a case in which the first signs of asymmetry were noticed at the age of six months, so perhaps the determining agent begins to act during embryonic life. The affection is progressive for several years but later on tends to heal spontaneously, and in the later years of childhood the limbs on the affected side grow at the same rate as the limbs on the sound side.

Nothing definite is yet known as to the pathogenesis of chondrodysplasia. Bentzon (1924) holds the view that Ollier's disease represents the typical reaction of the bone to certain disorders in the innervation of their blood-vessels. He also stresses the fact that the affection is often confined to one half of the body. From an experimental standpoint, working with rabbits, he was able, by interrupting the sympathetic nerves to produce in several instances structural changes similar to those seen in Ollier's disease. Murk Jansen (1928) believes that chondrodysplasia is the result of a retardation of the differentiation of cartilage cells consequent upon a deficient blood-supply which may be due to vasoconstriction from faulty action of the sympathetic nerves. It may thus be concluded that, perhaps, sympathetic dysfunction plays an important part on the pathogenesis of Ollier's disease.

Scleroderma

Scleroderma is now considered by many authors to be an angiotrophoneurosis in which unilateral atrophy frequently develops as a secondary symptom. If the disease starts during youth, the growth of limbs on the affected side may suffer.

Cockayne (1916) found several instances of complete hemi-atrophy (non-congenital) with scleroderma of the atrophic parts of the body. Meyer (1936) assumes that the common cause for hemi-atrophy and scleroderma may be looked for in an abnormal function of the sympathetic system, which reacts in some instances by causing scleroderma, in others by causing a hemi-atrophy or a combination of both abnormalities according to the part of the sympathetic which is particularly affected.

Facial hemiatrophy

In 1846 Romberg described a case with wasting of one side of the face. He called this affection 'facial hemi-atrophy' as a proof for the existence of trophic nerves.

Jendrassik (1884) assumes that the cause of facial hemi-atrophy is a lesion of the sympathetic cervical ganglia or of the fibres of Remak connected therewith, whereas Archambault and Fromm (1932) are of the opinion that sympathetic implication is its only underlying cause. This view has been confirmed by clinical observations. Brüning and Kroll (Meyer, 1936) found alterations in the ganglion cervicale superior in some instances of facial hemi-atrophy. Manthey (1928) reported a case of facial hemi-atrophy developing after cut-injury of the cervical sympathetic and in a case reported by Bost (1927) the cervical sympathetic had been injured by fracture of the clavicle.

Although in all these instances the hemi-atrophy is affecting the face only, and is mostly of a non-

congenital type, they may furnish a proof for the essential part which the sympathetic system plays in the pathogenesis of unilateral atrophy. The condition is strictly unilateral. It extends gradually, involving skin, fat and subcutaneous tissues until the entire side of face is affected. The bones show retarded growth or atrophy.

Hemi-hypertrophy and vasculo-cutaneous disturbances

Congenital hemi-hypertrophy is frequently associated with vasculo-cutaneous disturbances, e.g. pigmentation or haemangioma. Furthermore, diffuse angioma are often accompanied by local gigantism or partial hemi-hypertrophy. It has been assumed that this unilateral overgrowth might be the result of an unbalanced action of the sympathetic system, giving rise to an inequality in blood flow or distribution.

Report of case

J.E. a boy, aged 11 years, was admitted to the Queen Elizabeth Hospital for Children, Bayford, Herts., in January 1944.

Family history: The patient's mother is healthy, but 'highly strung.' The patient's father, who deserted the mother soon after the child's birth, is said to have been healthy and nothing abnormal has been reported in his family. No physical or mental abnormality can be determined in the mother's family.

Patient's history: The patient, an only child, was a full-term infant. His was an instrumental delivery. Birth-weight was 5 lb. 6 oz. He was bottle-fed. No definite date is available as to the starting of dentition, but he is said to have acquired his teeth rather early. He sat up when ten months' old, began to talk at fourteen months and started walking with support of a specially constructed orthopaedic boot at the age of two years. He has had measles, rubella, mumps, whooping-cough and chicken-pox.

At birth the child presented a marked underdevelopment of the right side of the body. The right side of the face was much smaller and flatter than the left one, the right arm and leg showed a remarkable shortening, he had a dorsal scoliosis to the right and a kyphosis. Testicles were absent from scrotum. During the first year of life the atrophy of the right side of the face became gradually less marked. During the following years, according to his mother's statement, this did not increase to any remarkable degree, the rate of growth of both apparently almost keeping pace with each other. The kyphoscoliosis, however, became much more conspicuous. He was treated in various hospitals where he received all sorts of endocrine therapy (elityran, ant. lobe pituitary hormone), as well as osteopathic treatment, but with no success. He could move all limbs well and never showed any paralysis. Mentally he was bright and intelligent and went to a normal school until September 1943, where he was backward in physical attributes but appeared mentally alert. Since then he has been taught at home.

Examination: The patient is a small boy, his height corresponding to that of a child aged five

years. The most marked deformity, at first sight, is the conspicuous kyphoscoliosis. The face is somewhat asymmetrical, and the right arm and leg are shorter and thinner than the left ones. He walks with a limp, owing to the shortening of the right leg.

HEAD: The head is of normal shape. The face shows some degree of asymmetry, the right side being somewhat flatter and smaller than the left.

CRANIAL NERVES:

1st to 10th: Normal.

11th nerve: Function of both sternomastoid and trapezii muscles somewhat restricted, but this is probably mostly due to the deformity and fixation of the spine which does not allow free movements.

12th nerve: Tongue protrudes in midline. No fibrillation.

TEETH: The teeth are sound, but rather crowded together and in bad position.

THROAT: Normal.

NECK: The neck is short, owing to the deviation to the left of the cervical spine. Movements restricted.

THORAX: The thorax is grossly deformed. The shoulders are kept high, the right one slightly higher than the left one. The upper thoracic spine shows considerable lateral deviation with the convexity to the right, the cervical and the lower thoracic spine show a compensatory scoliosis with the convexity to the left. There is a conspicuous kyphosis of the dorsal spine. The sternum protrudes considerably, thus causing a vaulted appearance of the anterior wall of the chest. The inferior angle of the right scapula is somewhat higher and farther from the middle line than that of the left scapula.

HEART: The heart is displaced upwards and outwards. The dullness extends over an almost quadrangular area, the lower border running in the third intercostal space from the left sternal border to just outside the midclavicular line, the upward border running almost parallel in the first intercostal space. The apex beat is palpable in the third intercostal space, $\frac{1}{2}$ in. outside the midclavicular line. All sounds are normal. The pulse is normal in volume and rate. B.P. 95/60 mm. Hg.

RESPIRATORY SYSTEM: The percussion note over the right side of the chest is hyperresonant, more so anteriorly than posteriorly. The percussion note is normal over the left lung. Breath sounds are feeble, especially posteriorly, but everywhere there is normal vesicular breathing. Respiration rate is normal.

ABDOMEN: The abdomen is flat. Muscular development fairly good. Liver and spleen not palpable. Abdominal reflexes present and equal. Both testes are absent from the scrotum and cannot be felt in the inguinal canal. No hernia.

EXTREMITIES:

Arms: Left arm normal.

Right arm: The right arm is shorter ($2\frac{1}{2}$ in.) and thinner than the left. The palm and thenar eminence are flattened. The middle phalanx of the ring finger shows a slight lateral abduction, whereas the distal phalanx is kept in a slightly flexed position. The little finger is small and shows a marked contraction, with palmar flexion in both phalangeal joints, and slight dorsal flexion and ulnar abduction in the metacarpo-phalangeal joint. All movements

with the exception of the ring- and little finger, are normal and not restricted, but there is some degree of decreased muscular power as compared with the left arm.

There exists some laxity of the elbow joint and, especially, of the metacarpo-phalangeal joint of the thumb, thus allowing hyperextension. All reflexes are normal and equal on both sides. The patient is left-handed.

LEGS:

Left leg: Normal.

Right leg: The right leg is shorter ($2\frac{3}{4}$ in.) and thinner than the left one. All movements are normal and not restricted, but the muscular power is somewhat decreased in comparison to the left leg. All normal reflexes present and equal. No abnormal reflexes.

There is no evidence of ataxia or dysergia. No tremors, twitchings, choreiform movements or muscular spasms are present. All tests for touch, pain, temperature, vibratory, muscle, bone and tendon sense elicit normal and accurate responses.

THE SPEECH is normal.

THE SKIN is smooth, of normal consistency and there is no hyperhidrosis. The scalp hair is dense and equally distributed.

SLEEP: Normal.

APPETITE: Normal. Patient does not suffer from abnormal thirst.

MICTURITION AND BOWEL ACTION: Normal.

MENTAL CONDITION: The patient is of average intelligence. He is bright and alert, but rather unstable emotionally and will break into tears on the slightest provocation. He is noisy, over-talkative and boisterous and tries to attract attention by every conceivable means. When together with other children he adopts a domineering and provoking, at times even aggressive attitude. He is witty, although in a somewhat affected manner. He draws extremely well. He gets on well with younger children, who admire him because of his witticisms and among whom he quickly makes himself the centre of attention, but he has difficulties in adjusting himself to the company of children of his own age group who are inclined to resent his boastful, provoking behaviour and, in their turn, deride him because of his physical deformity.

X-ray examination:

SKULL: There is a slight flattening of the right side of the skull. Otherwise the skull is normal. Pituitary fossa normal (fig. 1).

THORAX: There is a gross deformity of the upper dorsal spine and upper right ribs, with marked kyphoscoliosis convex to the right with a wedge-shaped deformity of the vertebral bodies. There is some degree of compensatory scoliosis convex to the left of the cervical and lower dorsal spine. The upper thoracic vertebral bodies show a deficiency in bone structure, with abnormal ossification, and contain irregular strands of calcareous material. Their cortex is thin and irregular, and there appears to exist some fusion between II and III, and IV, V, VI and VII. The right ribs are thin and atrophic, especially the upper six, and take a downward course, forming an acute angle with the vertebral bodies. The fourth to eighth left ribs are crowded

FIG. 1

FIG. 2

together and their intercostal spaces are narrow (fig. 2-4).

The lumbar spine appears to be normal.

The right os ilium is smaller than the left one.

EXTREMITIES: All bones are slender. The bones on the right side, including the carpal and tarsal bones, are smaller and thinner than the left ones. There is some lateral abduction of the middle and distal phalanges of the right ring finger, and marked plantar flexion of the middle and distal phalanges of the right little finger, with abduction of the 1st phalanx. The middle and distal phalanges of this finger show a high degree of atrophy. Both feet are small, the right one smaller than the left one, but otherwise well developed. The bone structure is normal, the development is normal for the age and there is no delay in ossification (fig. 5-9).

FIG. 3

MEASUREMENTS (IN INCHES)

Length of body (standing on both legs, without correction of the shortening of the right leg)	..	..	..	..	42½
Normal length of a child 11 years old	..	..	..	..	54
Vertex to symphysis	..	..	..	..	21
				Right	Left
Symphysis to sole	..	..	..	21½	23½
Proc. zygomaticus to chin	..	..	..	3	3½

ARMS:		Right	Left
Length of whole arm	..	18½	21
Humeral-clavicular juncture to lateral condylus humeri	..	8	9
Lateral condylus humeri to styloid process of radius	..	6	7
Radio-carpal joint to tip of middle-finger	..	4½	5
Circumference upper arm	..	6½	7
Circumference forearm	..	5	5½

LEGS:			
Total length of leg	..	21½	24
Spina iliaca ant. sup. to medial malleolus	..	23½	25
Trochanter maj. femoris to lat. condyle of tibia	..	11	12
Lateral condyle of tibia to lat. malleolus	..	9½	10½
Medial condyle of tibia to medial malleolus	..	9	10
Medial malleolus to top of big toe	..	4½	5
Circumference thigh	..	10½	12½
Circumference lower leg	..	7½	8½

FIG. 4

FIG. 5

Laboratory examinations

URINE:

Specific gravity: 1023.
Deposit: Nothing abnormal detected.
Total output within 24 hours: 1280 c.c.

BLOOD:

Erythrocytes	..	..	..	4,560,000 per c.mm.
Haemoglobin	..	..	..	82 per cent.
Colour index	..	..	..	0.9
Leucocytes	..	..	..	8,600 per c.mm.
Polymorphonuclears	..	..	..	61 per cent.
Lymphocytes	..	..	..	32 per cent.
Mononuclears	..	..	..	6 per cent.
Eosinophils	..	..	..	1 per cent.
Basophils	..	..	..	0

SUGAR TOLERANCE:

		Blood sugar (mgm. per cent.)
0 hours (23 gm. dextrose by mouth)	..	97
½ hour	..	170
1 hour	..	131
1½ hours	..	103
2 hours	..	95

Summary and conclusion

The case under discussion is a total congenital hemi-atrophy in a boy aged eleven years, affecting

the right side of the body, with a high degree of kyphoscoliosis. The body length corresponds to that of a child five years old, but this is probably due to the deformity of the spine. The extremities on the sound side are of about the normal length for a child aged eleven years. The long bones and the metacarpo-tarsal skeleton on the affected side are shorter and thinner than the ones on the sound side, but the bone structure, as seen by x-ray, is normal and there is no delay in ossification. The under-development is not confined to the bones alone, but also affects the soft tissues, as shown by the lesser circumference and the decreased muscular power of the limbs on the right side. The upper six right ribs show a high degree of atrophy. The vertebral bodies of the upper thoracic spine, especially II to VII, show gross deformity and abnormal structure of the bone tissue. Their appearance with the thin, irregular cortex, the deficiency of bone structure, with irregular strands of calcareous material, is similar to the changes seen in Ollier's disease. The extreme degree of wedge-shaped deformity is, of course, mostly due to the kyphoscoliosis which, in

FIG. 6

its turn, is partly due to the postural disturbances exerted by the underdevelopment of the right side of the body. It is possible, however, that the soft consistency of these bones, caused by the osseous dystrophy, played a part in aggravating their deformity.

The patient showed two more abnormalities, the contracture of the right hand little finger and cryptorchidism. The contracture of the little finger, with fixed palmar flexion of the middle and distal phalanges and slight dorsiflexion of the proximal phalanx, and the slight palmar flexion of the middle and distal phalanges of the ring finger, lead to the suspicion that this is a congenital contracture of the affected fingers, which is a comparatively common inherited deformity. It has been suggested that this abnormality is probably due to imperfect development of the anterior ligament of the first interphalangeal joint. Regarding the cryptorchidism it cannot be determined here whether this condition is due to hormonal deficiency or to anatomical deficiency. The former is the more common cause of undescended testicles, but lately the fact has been

stressed that anatomical defects appear to play an important part in many cases of cryptorchidism.

One further note may be made here about the patient's mental make-up. His very noisy, boastful, provoking and, at times, aggressive behaviour can be explained as a defence mechanism. By over-stressing his mental capacities he tried, unconsciously, to counteract the feeling of inferiority caused by his physical deformity. He thus 'over-compensated his inferiority-complex.' As a consequence, his social adjustment with children of his own age-group was rendered rather difficult.

Nothing definite can be said regarding the pathogenesis of this case. The factor determining the underdevelopment of the right side must have acted during embryonic life as the hemi-atrophy was found to be present at birth. Furthermore, the action of this agent was probably mostly confined to the period of intrauterine development, as the discrepancy in length between the two sides of the body is said not to have much increased after birth.

An influence by the pituitary gland cannot be excluded. But one of the main features of hypo-

FIG. 7

FIG. 8

physcal dwarfism is unimpaired symmetry of the body. The cryptorchidism in this case may be a symptom of hypopituitarism, but, on the other hand, the incidence of this abnormality is high and it cannot therefore be concluded, from this fact alone, that the hemi-atrophy is due to hypophyseal deficiency, i.e. lack in growth hormone. Besides, that would not explain the unilaterality of the affection.

It has been suggested that cerebral portions of the visceral nervous system exert a trophic control on all body tissues, perhaps partly via the sympathetic system, and may thus influence growth asymmetrically. It has also been shown that facial hemi-atrophy may be brought about by changes or injury of the sympathetic on the affected side. It has further been mentioned that scleroderma which is frequently associated with hemi-atrophy as a secondary symptom, may have its origin in an abnormal function of the sympathetic system.

Lastly it has been pointed out that sympathetic imbalance may play a major part in the pathogenesis of chondrodysplasia, in which a unilateral arrest of growth is frequently met with. From that point of view one further note may be made regarding the relations existing between congenital hemi-atrophy and Ollier's disease. In both affections the responsible agent acts during early life. Hemi-atrophy is congenital (as in the reported case). Ollier's disease starts in infancy or early youth (the case reported by Cleveland showed the first symptoms at six months), i.e. it may already be present at birth. Both conditions are not progressive during the whole time of adolescence. In the reported case of hemi-atrophy the growth of both sides apparently almost kept pace with each other after birth. Ollier's disease usually comes to a standstill in the later years of growth. Ollier's disease is frequently (according to several authors always), confined to one side of the body and the

FIG. 9

affected limbs show an arrest in development. Both unilaterality and underdevelopment are the main features of hemi-atrophy. Furthermore the reported case showed changes in several vertebral bodies similar to those seen in Ollier's disease. It thus appears that some relationship might exist between these two abnormalities.

A final conclusion regarding the etiology of congenital hemi-atrophy can, however, not be given, as some still unknown morphogenetic factor is probably concerned.

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