Isolated deficits of the long bones form a well-recognized group of anomalies. They may be described as terminal, in which there are no unaffected parts distal to and in line with the deficient portion (fig. 1); or intercalary, in which a middle part is deficient while those portions proximal and distal to it are present (fig. 2) (3).

Ulnar hemimelia is a postaxial longitudinal deficiency of the upper limb, wherein the ulna is completely or partially absent. Clinically, because of the multiplicity of forearm and hand deformities or contours, it may be very difficult to recognize precisely the deficiency without roentgen studies (figs. 3 and 4). The elbow joint may be in extension or in acute flexion. There may be fusion of the radiohumeral joint. The range of motion, if present, may be markedly limited. The proximal part of the radius may articulate with the underdeveloped capitulum, or it may be completelyluxated. If the deficiency is incomplete, the ulnar remnant may vary in length and contour. The digits of the hand may vary greatly in number (figs. 5 and 6). At the shoulder girdle, one may observe considerable muscular atrophy, ligamentous relaxation, and a deep web in the axilla.

In 1932, Kanavel (6) reported 60 cases of ulnar deficiencies. Comparison of Kanavel's findings with those of the cases presented here reveals the digit deficits as shown in table 1.

O'Rahilly (17) presented a resume of 65 cases in the literature up to 1950. This deficit is seen much less frequently than is radial hemimelia, the literature indicating a ratio of 18:1. O'Rahilly's analysis revealed that 67% of the cases were unilateral, and 69% involved the right upper limb. The incidence in males was more common, with a ratio of 2:1. Radiohumeral fusion and/or digital syndactyly were not mentioned.
The absence of a radiohumeral joint (fusion) indicates the failure of cavitation of this structure. It is suggested that the lack of cavitation is an integral part of the total deficit seen in some cases of ulnar hemimelia (38.5% of Frantz's patients).

During the past 15 years, the staff at the Area Child Amputee Center has examined and managed 26 children with ulnar hemimelia. An analysis of these cases reveals a follow-up of from 1 to 15 years. There were 16 males and 10 females.

This deficit appears to be a sporadic lesion, in that there were 59 normal siblings of the 26 patients studied. One patient had a fraternal twin who had no skeletal deficits.

Ten of the patients (38.5%) had unilateral ulnar hemimelia with no other skeletal deficiencies. Three children (11.5%) had bilateral ulnar hemimelia; seven also had lower-limb deficits. Six patients with unilateral ulnar hemimelia had varying deficiencies in the contralateral upper limb. These included terminal transverse hemimelia, phocomelia, absent thumb, and absent fifth finger. Ten patients (38.5%) had radiohumeral fusion accompanying the ulnar hemimelia.

The involvement of carpals and metacarpals is complex. The triquetrum and capitate often are absent. There is an increasing frequency of metacarpal failure as one passes from the radial to the ulnar side of the hand.

Fig. 2. Intercalary longitudinal paraxial hemimelia, ulnar. Note that all five fingers are present.

Fig. 3. Left, the short left upper limb is phocomelic. Note the severe atrophy of the left shoulder girdle. There are three digits in the hand. The right arm (ulnar hemimelia) demonstrates good shoulder musculature and motion. Center, abduction and forward flexion are limited by the axillary web. Right, X-rays reveal fused right radiohumeral joint (failure of cavitation).
ULNAR HEMIMELIA

Fig. 4. Left, bilateral ulnar hemimelia, with monodigital hands. Right, note the deep web at the cubital fossa (pterygium). Center, X-rays reveal the radiohumeral relationship. There is no true elbow joint.

The frequency of digital absence is shown in table 2. It is of interest to note that the three-fingered hand is preponderant, followed closely in occurrence by the monodigital hand.

MANAGEMENT

In our experience, most of these children can be managed without surgical intervention. The goal, of course, is to improve function, with or without the use of a prosthesis. Whether surgery is indicated depends upon whether both arms are involved, and on the range of motion, the number of digits present, and the presence or absence of syndactyly (table 3).

NONSURGICAL

No Fitting

Some of these children had radiohumeral synostosis (figs. 3 and 7).

Opponens Post

Children with one digit (monodigital hand) possessing good flexion power and lateral stability of the metacarpophalangeal joint were fitted to advantage (fig. 8).

Below-elbow Prosthesis

Modified below-elbow sockets were sometimes prescribed (fig. 9). However, range of elbow motion is significantly lacking.
Fig. 5. Left, bilateral ulnar hemimelia. The left is intercalary, since there are five digits; the right is terminal because there are only four digits. Patient has complete anonychia with distinctive pulp prints on the dorsum of the fingers. Right, X-rays reveal complete dislocation of the radiohumeral joints.

Fig. 6. Left, monodigital ulnar hemimelia, incomplete. Right, X-rays reveal proximal remnant of the ulna, with a bowed radius.
Above-elbow Prosthesis

This is a highly satisfactory method offering patients with unilateral, monodigital, ulnar hemimelia. The forearm segment is acutely flexed against and parallel to the humeral shaft and then encased within the humeral socket. The elbow-locking mechanism has a lever with which the single digit controls the elbow lock and unlock mechanism (fig. 10).

SURGICAL

Elbow Z-plasty

Z-plasty in the cubital fossa was performed in two instances in an endeavor to decrease the cubital web and in the hope of allowing a greater range of elbow flexion and extension. This procedure is somewhat advantageous in that it allows a better fit of the forearm socket, but it fails to offer any significant increased range of motion and therefore is not recommended (see fig. 9).

Elbow Disarticulation

This surgical procedure is followed by fitting the limb with an elbow-disarticulation type of prosthesis. The surgeon should be meticulous in his technique so as not to disturb the distal humeral epiphysis during the disarticulation procedure.

The application of the elbow-disarticulation type of prosthesis with an outside locking elbow offers 11 different positions of the elbow joint.

Humeral Derotation Osteotomy

Two children received a humeral derotation osteotomy of at least 90 degrees (fig. 11). One was lost to follow-up after early union.
DISCUSSION

From this brief outline of management, it is obvious that the treatment of these children is highly individualized. The timing and procedure may be dictated by the age of the patient, the question of bilaterality, and the scope of the handicap. The decision as to whether or not to prescribe a prosthesis may be a difficult one. The approach to handling these children with ulnar hemimelia has been developed over the years by trial and error and by functional analysis.

In figure 3, severe as the deformities may appear to be, the right shoulder functions normally, and the boy is able to abduct and forward-flex the shoulder, which allows him to prehend with his right hand. The left upper limb is phocomelic; however, he has a functional "pinch force" with the digits for close-in functioning. In the occupational therapy department, he demonstrated a very acceptable level of accomplishment in the activities of daily living and therefore was not fitted with prostheses.

This logic is in accord with the problem faced by the boy shown in figure 7. The efficiency of this four-year-old's performance in dressing, undressing, and toilet care is such that he needs no prosthetic aids. Utilizing the ulnar hemimelic limb, this boy is able to feed himself and care for most of his daily living demands.

Bilateral ulnar hemimelia with monodigital hands is a severe handicap (see fig. 4). One male in this group had the Cornelia de Lange syndrome. If a child is seen at an early age (i.e., before two years), one may be tempted to procrastinate. How long? The major question is whether one should fit one or both sides with a passive type of prosthesis (terminal devices with no cables, but with small rubber bands on the hooks) or whether to interfere surgically.

Fig. 8. This boy was born with bilateral ulnar hemimelia with monodigital hands (see fig. 4). At 4 years of age the right upper limb was fitted with an opponens post. The left limb was managed by elbow disarticulation and prosthesis replacement. The elbow unit has 11 positions, allowing from 45° flexion to 180° extension.

Fig. 9. Left, monodigital ulnar hemimelia, with extension limited to 70°. Web release in the cubital fossa offered little additional motion. Initially the child was fitted with a below-elbow type of prosthesis (center). After a 2-year trial, the family expressed dissatisfaction with the limited motion and function of the arm. At 4 years of age an elbow disarticulation was performed and prosthetically fitted (right).
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It has been stated that a Z-plasty at the cubital fossa offers little improvement of the radiohumeral arc of motion.

One approach may be to fit one side with an opponens post and the opposite side with a modified below-elbow prosthesis. Should the prosthetic side prove to be inadequate with a below-elbow type of prosthesis, one may then elect to perform an elbow disarticulation one year before kindergarten, allowing a year of prosthetic wearing before formal schooling. This was done in the patient shown in figure 8. At this writing, the boy is 14 years old. He is in junior high school and is the manager of the football team. Also, he is a fair bowler, for which he utilizes a special attachment to his prosthesis.

Unilateral, monodigital, ulnar hemimelia with a normal contralateral upper limb is not as serious a handicap. The patient shown in figure 9 was fitted at two years of age with a modified standard below-elbow prosthesis. At the age of four years, the patient and her mother were dissatisfied with the function afforded, because of limited elbow motion. (The Z-plasty at the cubital fossa offered little additional motion.) The child received an elbow disarticulation and was subsequently fitted with a standard elbow-disarticulation prosthesis with a medially placed outside-locking elbow. At the time of writing, she is 18 years of age, ready to enter college, and is considered a very good prosthesis-wearer.

The patient in figure 10 was seen in 1964 at 15 years of age; she has a monodigital, left-sided, ulnar hemimelia. Her degree of radiohumeral flexion was more severe than that of the girl in figure 9. This patient was not particularly concerned with the cosmetic effect (and still is not). She was fitted with a prosthesis that encased the acutely flexed forearm within the humeral socket. The anterior, or ventral, wall of the socket was then fenestrated and a lever was attached.
to the elbow-locking cable, which permitted her to use the single digit to operate the elbow locking/unlocking mechanism. At this writing, she is in her second year in college and now wears a mechanical hand with a cosmetic glove. The upper arm is usually covered by a fluffy-sleeved blouse.

To summarize, there are four approaches to treatment of the monodigital hand: opponens post; below-elbow prosthetic fitting; elbow-disarticulation prosthetic fitting, encasing the forearm in the humeral socket; or no fitting, which is the least recommended procedure.

Rotational deformities occasionally are seen in which there may be up to 180 degrees of medial rotation of the forearm on the humerus. The hand rests at the side of the thorax, pointing dorsally. One patient was seen at eight months of age (see fig. 11). There were three digits in the left hand with soft-tissue syndactyly. She received a derotation osteotomy of the humerus at the age of four years, and a fair result was ob-

| TABLE 1. FREQUENCY OF ABSENCE OF DIGITS IN ULNAR DEFICIENCIES |
|----------------------|------|------|------|------|------|
| Digit                | 1    | 2    | 3    | 4    | 5    |
| Kanavel (69 cases)   | 4    | 8    | 18   | 29   | 32   |
| Prantz/O’Rahilly (26 cases) | 0   | 10   | 14   | 29   | 20   |

| TABLE 2. DIGITAL ABSENCE IN ULNAR HEMIMELIA, 26 CASES |
|----------------------|------|------|------|------|------|
| Digit                | Unilateral | Bilateral |
|                      | Right | Left |
| 1                    | 8     | 2     |
| 2                    | 3     | 2     |
| 3                    | 9     | 0     |
| 4                    | 1     | 1     |
| 5                    | 0     | 1     |
|                      | 21    | 5     |

| TABLE 3. SURGICAL AND NONSURGICAL MANAGEMENT OF ULNAR HEMIMELIA, 26 CASES |
|----------------------|------|------|------|------|------|
| Non surgical       | Cases | Surgical       | Cases |
| No fitting         | 5     | Syndactyly release | 2     |
| Opponens post      | 1     | Elbow Z-plasty   | 2     |
| Below-elbow fitting| 7     | Elbow disarticulation | 2     |
| Above-elbow fitting| 5     | Humeral derotation osteotomy | 2     |
|                    | 18    |                   | 8     |
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tained. Unfortunately, she was lost to clinic follow-up shortly after surgery.

Dislocation of the radiohumeral joint is rare. One such patient was first seen at four years of age. He has five digits on the left hand and four on the right. There were no fingernails. It is of interest to note that this boy has distinctive prints on both the palmar and dorsal surfaces of his fingers. His radiohumeral joint anatomically is nonexistent (see fig. 5). The intrinsic muscles of the hands are weakened, and the wrists are unstable. The forearms and hands have been encased in a half-sleeve of plastic attached to crutches (he also has bilateral amelia of the legs). He is now 18 years old and attends a trade school.

Incomplete ulnar hemimelia occurred twice in this series. The proximal portion of the ulna is present, thus affording a normally appearing elbow joint with an excellent range of motion (see fig. 6). That child was seen at four years of age and fitted with a standard below-elbow prosthesis, which she is currently wearing.

Syndactyly was encountered four times in 26 cases. Two cases have been corrected surgically.—Charles H. Frantz, M.D.

PATHOGENESIS

The term "hemimelia" (hemimelie) was introduced in 1836-37 by Isidore Geoffroy Saint-Hilaire (19), who also introduced the term "teratology". In 1877, Verneuil proposed subdivision of "ectromelia" into longitudinal and transverse varieties (21). In addition to absence of the distal half (two of the four segments) of a limb, it became clear that, in some cases, only one side of the distal half was affected, and such instances were named (after the defective portion) "radial," "ulnar," "tibial," and "fibular" hemimelia. By 1903, a further distinction, that between terminal and intercalary varieties of hemimelia, had been made (7). Finally, in 1951, O'Rahilly suggested the term "paraxial hemimelia" for the longitudinal variety, because either the preaxial or postaxial side of the limb is involved in such cases.

It is not proposed to discuss here either the terminological basis (19) or the teratogenesis (11) of limb malformations in general, as these aspects have been considered recently elsewhere.

Ulnar hemimelia was first reported in 1683 by Goller (14) and hence is probably the first of the paraxial hemimelias to be identified as such, there being some doubt about the true identity of the case of hemimelia described by Pare in 1573 (13).

Although chronological tables of all the early cases of radial, tibial, and fibular hemimelia are available in the literature, no such list other than the bibliography provided by Rabaud and Hovelacque (21) seems to have been prepared for ulnar hemimelia.

Among the hemimelias involving one of the four bones of the third limb segment, or "zygopodium" (forearm and leg), the ulnar type occurs the least. It differs from the others also in that a partial deficiency is more commonly found than complete absence. However, it resembles radial, tibial, and fibular hemimelia in that it is more frequently unilateral, more commonly seen on the right side, and more often observed in the male (17). Of particular interest are those cases in which thorough dissection has been possible (23).

Several additional cases of ulnar hemimelia have been reported in the literature during the past two decades. The higher incidence of unilaterality and of right-sided involvement has been confirmed (9).

It is important to appreciate that the hemimelias may occur as isolated anomalies, or they may, as shown in this paper, be associated with other malformations. Ulnar hemimelia, for example, is sometimes a component of a sporadic syndrome that includes femoral and fibular defects (8). The cause of the "FFU" (femur, fibula, ulna) syndrome is unknown; such factors as parental age and thalidomide have been ruled out, and familial occurrence has not been observed.

A striking example of familial occurrence in several generations was recounted to Roberts (22) by a patient with ulnar hemimelia. Partial ulnar hemimelia of the intercalary type, together with hypoplasia of the thumbs and fibular hemimelia, has more
recently been described and illustrated in two brothers (25). A different condition, ulnofibular dysplasia, characterized by shortening of the ulna and fibula, was found to be inherited as an autosomal dominant (20).

Ulnar hemimelia accompanied by Polydactyly is not unknown (25), and the coexistence of Polydactyly and a long-bone deficiency in the same limb has been noted previously (e.g., heptadactyly and tibial hemimelia) (18). In such cases, it has been suggested that this seeming paradox of excess associated with deficiency may perhaps result from an excessive outgrowth, which occurs relatively late in the early embryonic period, "involved only the digital area, and attracts some of the tissue immediately proximal to the area of excess outgrowth" (26).

In the human, the hand appears in mesenchyme at about 41 postovulatory days (stage 17), so that it may be expected that Polydactyly would be observable by about six weeks after fertilization. Indeed, an example of this as an isolated anomaly has been described (16).

What are generally termed "fusions" of skeletal elements—that is, the occurrence as a single structure of something that is usually composed of two or more elements—may be found either as an isolated anomaly or in association with other disturbances. Carpal and tarsal fusions, for example, are not infrequent in the paraxial hemimelias, and, as emphasized in this paper, ulnar hemimelia may include humeroradial fusion. Normally, of course, certain bony fusions, such as those between the epiphyses and their diaphyses and between the neural arches and their centra, are of constant occurrence. Even in areas where synovial cavities might be expected, however, fusions are not infrequent, such as symphalangia between the middle and distal phalanges of the little toe. The histological development of phalangeal fusion has been studied in detail (2,4), and it is of interest to note that carpal and tarsal fusions have been observed in both the embryonic and the fetal period (4). That such fusions arise early during embryonic development as an absence of joint cavitation (17) is also suggested by studies of experimentally paralyzed chick embryos, in which articular cavities do not form (1,15). The cartilaginous skeletal elements, which are at first united by mesenchyme, become, under these conditions, joined together by fibrous tissue or by cartilage. In other words, fusion takes place across the presumptive joint regions.

That hemimelia occurs at a very early stage of embryonic life is indicated by the important, but neglected, observations of Hovelacque and Noel (5) on a strain of mice presenting tibial hemimelia. It was found that "the first manifestations of the anomaly are disclosed at a very early stage of development. They can be detected in embryos when the undifferentiated blastema begins to undergo change." In the tibial zone of the blastema, a "fibrous tract" appeared, and was connected to the fibula by the interosseous membrane. In some of these embryos, cartilaginous nodules developed in the area (especially proximally) where the tibia would normally form. Such nodules were in direct continuity with the fibrous tract; both constituted a unit that represented the tibia. The vascularization of the limbs was entirely normal. It was concluded (21) that "the tibia is never completely absent despite appearances; one can always find a trace of the element though it may be represented by only a nodule of pinhead size." There is no reason to believe that the above statements would not apply equally to the other types of paraxial hemimelia.

To return to the human—the mesenchymal femur, tibia, and fibula appear at about 41 postovulatory days (stage 17), and the humerus, radius, and ulna appear at about 37 postovulatory days (stage 16). In other words, it may be expected that, in the light of the French workers' observations, paraxial hemimelia could be detected in the human before six weeks after fertilization.

Prior to the first appearance of these specific skeletal elements, a sensitive period for teratogenic agents exists, as have been shown by correlations between the time of ingestion of thalidomide during pregnancy and the types of resultant anomalies (12). Thus, tibial defects occurred
mostly when ingestion began before the 46th menstrual day (perhaps about 32 post-ovulatory days). In one illustrated case, ingestion that commenced at 46 menstrual days resulted in bilateral radial hemimelia and malformations of the femur and tibia.

Finally, it may be mentioned that ulnar hemimelia has been found sporadically in various animals, such as the pig (23, 24). It has also been produced experimentally by the inclusion of large doses of acetazolamide (a carbonic anhydrase inhibitor) in the diet of rats during pregnancy (10). Of particular interest in these experiments is the circumstance that the ulnar hemimelia was practically restricted to the right side of the body.—Ronan O’Rahilly, M.D.

SUMMARY

The management of 26 cases of ulnar hemimelia has been discussed. This deficit is seen 1:8 less frequently than radial hemimelia. Bilaterality was present in 23% of the cases. Prior to determining the plan of treatment, a complete functional analysis should be carried out. Most of these children do not need surgery and may be treated by prosthetic fitting only. The pathogenesis of paraxial hemimelia and the embryogenesis of associated conditions, such as Polydactyly and joint fusions, are discussed.

REFERENCES