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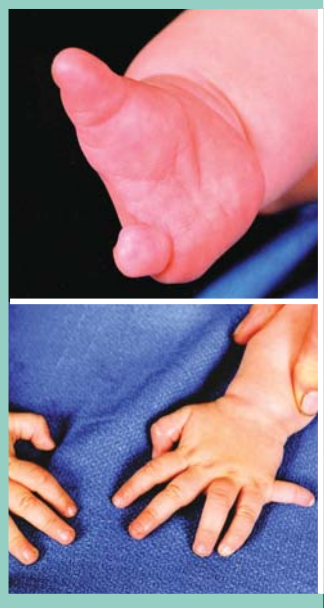
The essence of medical practice

Upper extremity congenital anomalies



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EDITORIAL

Dear Doctor,

Happy New Year!

Welcome to this New Year Issue of Info Medicus.
In this issue, we have feature on "Upper extremity congenital anomalies" as a review article. Congenital abnormalities of the upper extremity are abnormalities that are present at birth. It can affect any part of the body and can affect the body in different ways. Some congenital anomalies affect the way a person looks, while others affect the way a part of the body works. The most common is webbing of the fingers.

We hope this article will be infomative and useful reading for you.

You will also find an article on "Differentiation and diagnosis of tremor" in clinician's corner. Tremor is an involuntary, rhythmic, oscillatory movement of a body part. It is the most common movement disorder encountered in clinical practice. The diagnosis of tremor is based on clinical information obtained from a thorough history and physical examination, hope you will like this article as well.

In this issue we also emphasized on "Management of renal colic" in clinical review. This review includes current guidelines, and other peer reviewed evidence to provide a background on presentation, investigation and medical & surgical management of patients with renal colic.

We have presented a case on "Bulla in the lung" in case review. Several factors have been proposed for the formation of giant bullae, including congenital pulmonary and vascular malformations and smoking. Patients often receive adequate treatment only at an advanced stage.

Besides these, regular sections are presented as usual.

We welcome your feedback regarding "Info Medicus". Your valuable feedback will assist us to better meet your needs and to improve this service.

On behalf of the editorial board, we wish you all a very blissful, healthy and successful life in the year ahead.

Thanks and best regards,

(Dr. S. M. Saidur Rahman)
Medical Services Manager

(Dr. Rumana Dowla)
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REVIEW ARTICLE

Upper extremity congenital anomalies

Congenital anomalies affect 1% to 2% of newborns, and approximately 10% of those children have upper extremity abnormalities. The anomalies require an accurate diagnosis and communication of relevant information to the family. The era of managed care has changed the patterns of referral of children with limb anomalies. Certain upper extremity anomalies occur in isolation, whereas others are associated with systemic conditions. These associated disorders often take precedence over the limb anomaly and must be assessed with appropriate diagnostic testing.

Classification

There are numerous systems for classification of upper limb anomalies on the basis of embryology, teratologic sequencing, and/or anatomy. Each proposal had merit at the time of its inception, although many systems became outdated as our understanding of limb development and genetics was expanded. Embryologic classification defines the defect according to the malformation during limb development. Teratologic sequencing grades congenital anomalies according to the severity of expression. Varying degrees of damage and expression within the limb lead to variable phenotypes. Classifying or ranking congenital anomalies according to their severity of expression is popular because the extent of the pathology often determines function and provides a basis on which to guide treatment. An example of teratologic sequencing is classification of thumb hypoplasia into five types of increasing severity. Anatomical classification schemes provide useful descriptive analyses and often offer therapeutic guidelines for treatment based on pathoanatomy. The most widely accepted classification of congenital limb anomalies was proposed by Frantz and O'Rahilly and presented by Swanson. This system defines the anomalies according to the embryonic failure during development and relies on the clinical diagnosis for categorization. Each limb malformation is classified according to the most predominant anomaly and is placed into one of seven categories (Table I). Different clinical presentations of similar categories of embryonic failures are explained by varying degrees of damage within the organization of the limb mesenchyme.

Table I

Embryologic classification

- **I. Failure of formation of parts**
  - A. Transverse deficiencies
  - B. Longitudinal deficiencies
    - 1. Phocomelia
    - 2. Radial
    - 3. Central
    - 4. Ulnar
- **II. Failure of differentiation**
  - A. Synostosis
  - B. Radial head dislocation
  - C. Symphalangism
  - D. Syndactyly
  - E. Contracture
  - 1. Soft tissue
    - a. Arthrogryposis
    - b. Pterygium
    - c. Trigger
    - d. Absent extensor tendons
    - e. Hypoplastic thumb
    - f. Clasped thumb
    - g. Retroflexible thumb
    - h. Camptodactyly
    - i. Windblown hand
  - 2. Skeletal
    - a. Clinodactyly
    - b. Kirner deformity
    - c. Delta bone
- **III. Duplication**
  - A. Thumb
  - B. Triphalangism/hyperphalangism
  - C. Polydactyly
  - D. Mirror hand
- **IV. Overgrowth**
  - A. Limb
  - B. Macrodactyly
- **V. Undergrowth**
- **VI. Congenital constriction band syndrome**
- **VII. Generalized skeletal abnormalities**

Radial deficiency

One of the classic anomalies associated with systemic conditions is radial deficiency, which affects the preaxial border of the limb. The degree of preaxial deficiency can range from mild thumb hypoplasia to complete absence of the radius (Table II). Irrespective of the degree of expression, all forms warrant systemic evaluation for syndromes or associations. Holt Oram syndrome, thrombocytopenia absent radius syndrome, VACTERL association (vertebral abnormalities, anal atresia, cardiac abnormalities, tracheoesophageal fistula, esophageal atresia, renal defects, radial dysplasia, and lower limb abnormalities), and Fanconi anemia are the primary concerns. The principal systems involved in these syndromes are cardiac, renal, and hematologic. Children with VACTERL association can also have vertebral, tracheoesophageal, and anal problems (Table III). The heart is usually evaluated with auscultation and echocardiography. The kidneys are examined with ultrasound, and the platelet status is assessed with a blood-cell count and peripheral blood smear. The most devastating associated condition is Fanconi anemia. As children with Fanconi anemia do not have signs of bone marrow failure at birth, the diagnosis is not apparent initially. The median age of onset of aplastic anemia is approximately seven years, and the majority of children experience symptoms between the ages of three and twelve years. However, a chromosomal challenge test that detects the disease prior to the onset of bone-marrow failure is available. In this assay, a sample of the lymphocytes i s subjected to diepoxybutane or mitomycin C, which causes chromosomes within Fanconi anemia cells to break and rearrange. In contrast, lymphocytes from unaffected children are stable when treated with these agents. Since bone-marrow transplantation is the only cure for Fanconi anemia, this prefatory diagnosis is crucial for the child and affected family. Early diagnosis provides ample time to search for a suitable bone-marrow donor or for the parents to consider pre-implantation genetic diagnosis.

Table II

Global classification of radial longitudinal deficiency

Type	Thumb anomaly	Carpal anomaly*	Distal part of radius	Proximal part of radius
N	Absence or hypoplasia	Normal	Normal	Normal
O	Absence or hypoplasia	Absence, hypoplasia, or coalition	Normal	Normal, radioulnar synostosis, or radial head dislocation
1	Absence or hypoplasia	Absence, hypoplasia, or coalition	>2 mm shorter than ulna	Normal, radioulnar synostosis, or radial head dislocation
2	Absence or hypoplasia	Absence, hypoplasia, or coalition	Hypoplasia	Hypoplasia
3	Absence or hypoplasia	Absence, hypoplasia, or coalition	Absence of physis	Variable hypoplasia
4	Absence or hypoplasia	Absence, hypoplasia, or coalition	Absence	Absence

**Carpal anomaly indicates hypoplasia of, coalition of, absence of, or bipartite carpal bones. Hypoplasia and absence are more common on the radial side of the carpus, and coalition is more frequent on the ulnar side. Radiographs must be made when the child is older than eight years to allow for ossification of the carpal bones*

Table III

Syndromes associated with radial deficiency

Syndrome	Characteristics
Holt-Oram	Heart defects, most commonly cardiac septal defects
Thrombocytopenia-absent-radius syndrome	Thrombocytopenia present at birth but improves over time
VACTERL	Vertebral abnormalities, anal atresia, cardiac abnormalities, tracheoesophageal fistula, esophageal atresia, renal defects, radial dysplasia, lower-limb abnormalities
Fanconi anemia	Aplastic anemia not present at birth; develops at about 6 yr of age. Fatal without bonemarrow transplant. Chromosomal challenge test now available for early diagnosis

Ulnar deficiency

Ulnar deficiency is four to ten times less common than radial deficiency. Most importantly, unlike radial deficiencies, ulnar deficiencies are not associated with systemic conditions. Ulnar deficiencies are, however, associated with other musculoskeletal abnormalities that warrant a careful physical examination supplemented by radiographs (Fig. 1). An accurate diagnosis of ulnar deficiency obviates the need for the kind of extensive workup for systemic disorders that is indicated for patients with radial deficiency. At first consideration, one would expect a clear difference between radial and ulnar deficiencies. This distinction, however, is not always evident for multiple reasons. The remaining radius assumes characteristics similar to an ulna and can be fused with the distal part of the humerus (radiohumeral fusion). This prevents identification of the proximal part of the radius and results in a bone that resembles an ulna. As another confounding factor, the hand can have radial sided anomalies ranging from a narrow web space to an absent thumb (Table IV). These radial sided deficiencies do not transform an ulnar anomaly into a radial deficiency that warrants systemic evaluation. A careful clinical examination and scrutiny of the radiographs differentiates the two entities. Serial evaluations are occasionally necessary in equivocal cases.

Fig 1: A three year old child with ulnar deficiency of the right upper extremity. The elbow is fused, and the hand has two fingers and no thumb. An examination for other musculoskeletal abnormalities revealed congenital scoliosis secondary to a hemivertebra.

Table IV

Classification of ulnar deficiency according to abnormality of first web space

Type	Grade	Characteristics
A	Normal	Normal first web space and normal thumb
B	Mild	Mild deficiency of first web space and mild thumb hypoplasia with intact opposition and extrinsic tendon function
C	Moderate to severe	Moderate to severe deficiency of first web space and similar thumb hypoplasia with malrotation into plane of digits, loss of opposition, and dysfunction of extrinsic tendons
D	Absent	Absence of thumb

Central deficiencies

Cleft hand results from a longitudinal deficiency of the central rays of the hand (index, long, and ring fingers). During development, the differentiation of these rays occurs at a different time than the differentiation of the radial and ulnar rays (thumb and small finger). There are two types of cleft hand, typical and atypical, and they possess separate features and require discrimination from each other (Table V). In fact, there are major differences

Table V

Characteristics of cleft hand

	Typical cleft hand	Atypical cleft hand
Clinical features		
Involvement	Bilateral	Unilateral
Inheritance	Familial	Spontaneous
Syndactyly†	Common	Rare
Associated with Poland syndrome	No	Yes
Associated with cleft lip, cleft palate	Yes	No
Anatomical findings		
Arterial supply	Ring finger may have 3 digital arteries	Vestigial supply to central digits
Tendon	Dual tendons to ring finger common	Minimal
Skeleton	Hypertrophy adjacent to cleft	Hypoplasia
Classification	Failure of differentiation or abnormal number of digits	Failure of formation

†Syndactyly is considered failure of differentiation and may indicate a parallel mode of occurrence

between typical and atypical cleft hand (a form of symbrachydactyly) that may warrant placement of the two types into different categories of embryologic malformation. These findings suggest that typical cleft hand may result from fusion of, rather than absence of, digital rays. In contrast, atypical cleft hand is caused by necrosis of mesenchymal tissue and the body's attempt to regenerate deficient structures.

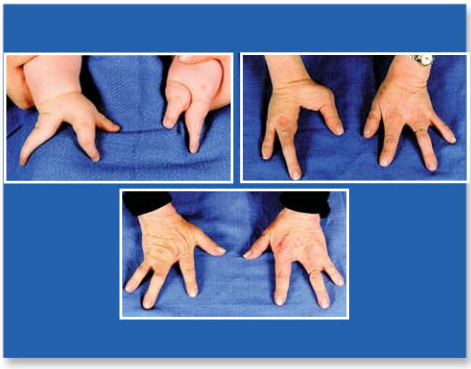


Fig 2: Three generations of typical central deficiency with different degrees of expression. Both of the grandmother's hands (bottom) and the mother's left hand (center) are missing only the long finger. The mother's right hand and both of the infant's hands are also missing the adjacent index and/or ring finger

The typical cleft hand has a v-shaped defect with varying degrees of long ray absence; most commonly the phalanges are missing, and the metacarpals are present. This type of central deficiency is often bilateral and is usually inherited as an X linked dominant trait with incomplete penetrance (Fig. 2). There may be syndactyly of the ring small or thumb index web space, and associated foot involvement is common. Typical central deficiency is also associated with cleft lip and palate. Atypical cleft hand is a form of symbrachydactyly that involves the central three digits (index, long, and ring fingers). Symbrachydactyly is a spectrum of hand deficiencies ranging from short fingers (brachydactyly) that may be connected (syndactyly) to the absence of the central three fingers to complete absence of the digits, similar to a transverse deficiency. In atypical cleft hand, the index, long, and ring fingers are absent while the metacarpals are present. This deficiency creates a u-shaped cleft instead of the v-shaped configuration associated with typical cleft hand, as seen in Fig. 2. An atypical cleft hand is not associated with systemic conditions and is not inherited. The deficiency is usually unilateral, sporadic, and not associated with foot involvement. All forms of symbrachydactyly (including atypical cleft hand) can be associated with Poland syndrome, which is characterized by an ipsilateral chest wall deficiency (Figs. 3-A and 3-B).

Fig 3-A: A two year old child with Poland syndrome. The right upper extremity has an atypical cleft hand, a form of symbrachydactyly

Both symbrachydactyly and Poland syndrome are attributed to a deficient blood supply to the developing limb. A more proximal arterial insufficiency of, for example, the axillary or subclavian artery, results in Poland syndrome, whereas a distal deficiency affects the apical ectodermal ridge and results in symbrachydactyly. The relationship between symbrachydactyly and Poland syndrome requires careful examination of the chest wall. Absence of the sternocostal portion of the pectoralis major muscle is the most common chest wall finding, although absence of additional muscles and breast underdevelopment can also be found. Considerable chest or breast asymmetry may require referral to a plastic surgeon after adolescence. Interestingly, the amount of hand deficiency does not correlate with the degree of chest wall abnormality. Therefore, even when a patient presents with mild symbrachydactyly, a complete evaluation of the integrity of the chest wall is required.

Transverse deficiencies

Congenital transverse deficiency is defined according to the last remaining bone segment. A short below the elbow amputation is the most common transverse deficiency of the upper extremity. The residual limb is usually well cushioned, and rudimentary nubbins or dimpling can be found on the end. These anomalies are usually unilateral, sporadic in occur occurrence, and rarely associated with other anomalies. The diagnosis should not be confused with amniotic disruption sequence (also known as constriction band syndrome), which is the result of entrapment of developing embryonic tissue by an amniotic band. This can manifest as amputation of a part and most commonly affects the digits (of the hands or feet). The diagnosis of amniotic disruption sequence requires the presence of a constriction band either affecting the involved extremity or elsewhere.

A less common level of transverse deficiency is through the hand or metacarpals. This deficiency creates considerable unilateral impairment that can be lessened by prosthetic fitting and/or advanced surgical procedures, such as toe to hand transfers. The long residual limb, however, dissuades the child from accepting a prosthesis and promotes the use of this limb as a sensate helper. The ultimate goal is restoration of prehension to allow independent usage. Advances in pediatric microsurgery have offered reasons to be optimistic concerning this lofty goal. Single and multiple toe to hand transfers are being applied to congenital and traumatic amputations at this level. The procedure requires

considerable expertise and careful postoperative monitoring. Successful toe to hand transfers achieve some form of prehensile activity and augment function. Phocomelia represents a longitudinal failure of formation with an absent intervening segment of the extremity (intercalary aplasia). The missing segment can be the arm or forearm, or both, with the hand attached directly to the shoulder. This deformity is uncommon, with the exception of the markedly increased prevalence (60%) that occurred in association with thalidomide taken during the first trimester of pregnancy. Surgery is rarely indicated and prosthetic fitting is very beneficial, especially for patients with bilateral involvement. Prosthetic fitting can be difficult, however, because of the extreme shortening of the limb.

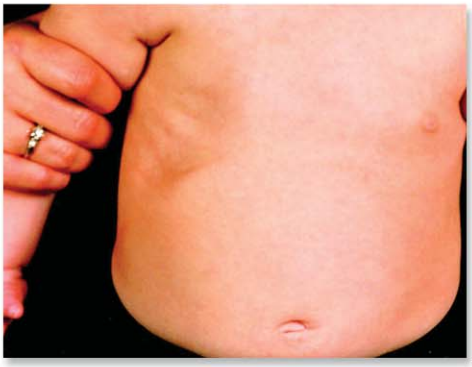


Fig 3-B: A two year old child with Poland syndrome. The chest wall shows absence of the sternocostal portion of the pectoralis major muscle and breast underdevelopment

Syndactyly

Syndactyly is defined as an abnormal interconnection between adjacent digits and is described according to the magnitude and extent of the linkage. The interconnection may encompass the entire length of the adjacent digits (complete) or it may discontinue proximal to the fingertip (incomplete). The syndactyly may involve only skin and fibrous tissue (simple) or include bone (complex). Syndactyly that occurs with other anomalies (e.g., Apert syndrome) is referred to as complicated syndactyly. Syndactyly is a common congenital anomaly, with an incidence of approximately two or three per 10,000 live births, and it tends to occur in families. Inheritable syndactylism is associated with genetic defects involving particular candidate regions on the second chromosome. The mode of transmission is considered to be autosomal dominant with variable expressivity and incomplete penetrance. This terminology signifies familial propagation, although the syndactyly may skip a generation and not be present in full form (variable phenotype). Familial syndactyly is associated with syndactyly of the second and third toes but not with systemic conditions. Syndactyly can also occur sporadically, and this form is also not ordinarily associated with systemic conditions. Complicated syndactyly is a broad category that encompasses many difficult forms of abnormal web space connection and osseous abnormalities. Many of these cases are associated with a syndrome, most notably Poland syndrome (symbrachydactyly), constriction bands, or acrocephalosyndactyly (also known as Apert syndrome). As is the case with atypical cleft hand, syndactyly can occur with Poland syndrome. The mildest hand anomaly associated with Poland syndrome is a small hand with incomplete, simple syndactyly. Amniotic disruption sequence results in digital amputation with or without pseudosyndactyly. This type of syndactyly has discrete characteristics that differ from those of inheritable or sporadic cases. First, the fingers may be truncated secondary to band constriction and amputation. Second, a small cleft resides where the normal web space formed prior to entanglement with the amniotic band. Third, formation of a constriction band around the lower extremity and/or clubfoot deformity is common. Apert syndrome or acrocephalosyndactyly is rare and represents a constellation of congenital anomalies that necessitates a multidisciplinary approach to management. These physically and mentally challenged children require care from various subspecialists, including neurosurgeons to monitor the cranial deformities, maxillofacial surgeons to manage the midfacial anomalies, otolaryngologists to address the potential airway obstruction, and hand surgeons to treat the upperextremity anomalies. Depending on the position of the thumb and the overall appearance, the hands have been described as resembling a spade, spoon, mitten, rosebud, or hoof (Fig. 4).

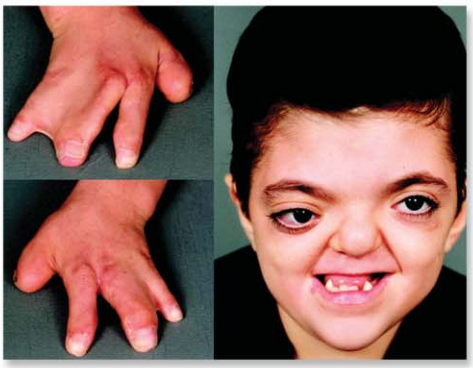


Fig. 4: A five year old child with typical facial features of Apert syndrome and bilateral hand syndactyly after separation of the index long web space. Clinodactyly of both thumbs is typical of Apert syndrome

The most severe forms incorporate the thumb into the complex syndactyly, forming a broad conjoined nail (synonychia) and osseous fusion between the radial four digits. The congested palm is cup-shaped and laden with deep crevasses. Correction of syndactyly is a formidable task that requires multiple procedures to obtain digital independence. Early recognition of the Apert hand is necessary to guide referral and to achieve a multidisciplinary approach to the management of these impaired children.

Polydactyly

Polydactyly can occur on the preaxial (radial) and the postaxial (ulnar) side of the limb. Preaxial polydactyly is more common in whites, and postaxial polydactyly is more common in blacks. Postaxial polydactyly in a white individual is uncommon and is often indicative of an underlying syndrome (e.g., chondroectodermal dysplasia or Ellis-van Creveld syndrome) (Fig. 5). Postaxial polydactyly is frequently inherited in an autosomal dominant pattern, but it has a variable penetrance pattern. The supernumerary digit is either well developed (type A) or rudimentary and pedunculated (type B). A small nubbin or scrawny postaxial element (type B) can be safely removed by tying the base with a suture in the nursery. The digit will turn gangrenous and fall from the hand. A residual bump or nubbin is the most common complication. A large or nearly normal digit (type A) requires operative ablation. The extra digit is removed, and any important functional parts (e.g., the ulnar collateral ligament and the abductor digiti quinti) are transferred to the adjacent finger. Even though preaxial thumb duplication demonstrates a racial predilection toward whites, most cases are unilateral, sporadic, and not associated with systemic problems. Further subclassification depends on the degree of skeletal replication. The most common type of preaxial polydactyly (about 50% [fifty-three] of 100 cases) involves duplicated proximal and distal phalanges that share a common articulation with a bifid metacarpal head. Treatment often requires use of portions of each component, so called "spare parts," to construct a properly aligned and functional thumb. Central polydactyly is an extra digit within the hand and not along its borders. The central polydactyly may be hidden within a concomitant syndactyly (i.e., synpolydactyly). Identification requires careful examination supplemented by radiographic verification. A particular form of central polydactyly (ring finger duplication) combined with syndactyly has a familial propagation and has been linked to a mutation of the HOXD13 gene on chromosome two.

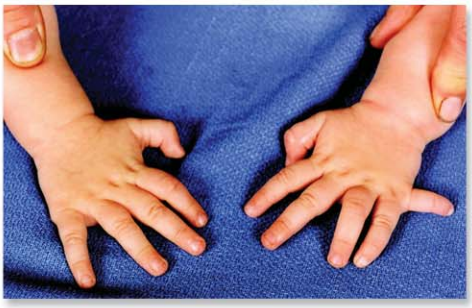


Fig 5: A one year old white child with left postaxial polydactyly. Systemic workup for associated syndromes is warranted in this situation

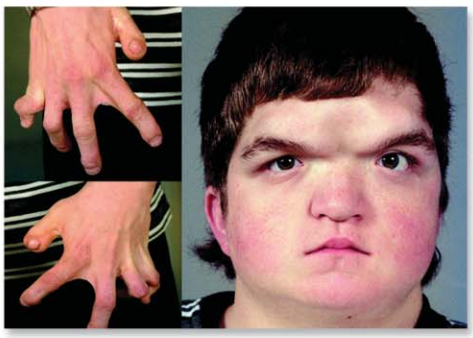


Fig 6: A sixteen year old boy with otopalatodigital syndromeand bilateral hand camptodactyly. Flexion deformities of all fingers are present; they are most severe in the ring and small digits

Camptodactyly

Camptodactyly is a painless flexion contracture of the proximal interphalangeal joint of the small finger that is usually gradually progressive. There is no intra articular or periarticular swelling. The metacarpophalangeal and distal interphalangeal joints are not affected, although compensatory deformities of these joints may develop. Camptodactyly is believed to occur in <1% of the population, although most cases are asymptomatic and many affected individuals do not seek medical attention. Camptodactyly is bilateral in approximately two thirds of patients, although the degree of contracture is usually not symmetrical. Other digits can be affected, although the prevalence decreases toward the radial side of the hand. Camptodactyly has been divided into three categories. A type I deformity is the most common form and becomes apparent during infancy. The deformity is usually an isolated finding and is limited to the small finger. This congenital form affects males and females equally. A type II deformity has similar clinical features, although it is not apparent until preadolescence. This acquired form of camptodactyly develops between the ages of seven and eleven years and affects females more often than it does males. It usually does not improve spontaneously, and it may progress to a severe flexion deformity. During the growth spurt of adolescence, the flexion deformity of the proximal interphalangeal joint progresses and can reach 90°. A type III deformity is often severe, usually involves multiple digits of both extremities, and is associated with a variety of syndromes (most commonly arthrogryposis). The extent of the involvement of the two hands is often asymmetric. This syndromic camptodactyly can occur in conjunction with craniofacial disorders, short stature, and chromosomal abnormalities (Table VI and Fig. 6).

Table VI

Generalized conditions associated with camptodactyly	
Craniofacial disorders	Orofaciodigital syndrome, craniocarpotarsal dystrophy (Freeman-Sheldon syndrome), oculodentodigital dysplasia, otopalatodigital syndrome
Chromosomal disorders	Trisomy 13-15
Short stature	Camptomelic dysplasia I, mucopolysaccharidosis, facial-digital-genital (Aarskog-Scott syndrome)
Others	Osteo-onychodysostosis (Turner-Kieser syndrome), cerebrohepatorenal (Zellweger syndrome)

Recognition of syndromic camptodactyly is usually straightforward, as numerous digits are involved and the condition is associated with facial distortion and limb contractures. Many other disorders present with a flexion deformity of the fingers, and these diagnoses must be considered during the evaluation of a patient with suspected camptodactyly (Table VII). A thorough history and physical examination can exclude the majority of these etiologies. Finger contractures associated with syndactyly, central deficiencies, or brachydactyly are not regarded as camptodactyly.

Table VII

Differential diagnosis of flexion deformities of the fingers

Diagnosis	Distinguishing feature
Mulitple pterygium syndrome	Multiple pterygia, usually includes knee and elbow
Arthrogryposis	Multiple joint involvement, waxy skin and underdeveloped musculature, ulnar deviation of digits
Symphalangism	No active or passive joint motion, absence of skin creases
Boutonnière deformity	History of trauma and pain, joint swelling, reciprocal hyperextension of distal interphalangeal joint
Beal syndrome	Congenital contractural arachnodactyly, kyphoscoliosis, external ear deformities, flexion contractures of proximal interphalangeal joint, elbows, knees
Marfan syndrome	Arachnodactyly without flexion contractures, loose ligaments, eye problems, dissecting aortic aneurysms
Juvenile palmar fibromatosis (mimics Dupuytren disease)	Metacarpophalangeal joint involvement, characteristic skin changes with nodules adherent to dermis
Trigger fingers	Metacarpophalangeal joint involvement, palpable click on finger extension
Inflammatory arthritis	Widespread joint involvement, swelling about joints or tendons

Clinodactyly

Clinodactyly is more common than camptodactyly but is less problematic. The abnormal deviation is in the coronal or radioulnar plane. Clinodactyly typically affects the middle phalanx of the small finger and produces an angulation of the distal interphalangeal joint. The deviation is usually in a radial direction. A deviation of <10° is so common, it may be considered normal. On occasion, clinodactyly can involve several digits and is usually related to one or more delta shaped middle phalanges. The deformity is usually fixed, and there is no intra articular or periarticular swelling. Clinodactyly can be inherited and is considered to be an autosomal dominant trait with variable expressivity and incomplete penetrance. Familial clinodactyly is usually not associated with systemic conditions. However, clinodactyly is a physical finding of many genetic syndromes and chromosomal abnormalities, most notably Down syndrome, in which the prevalence is between 35% and 79%4. Thumb clinodactyly is a prominent feature of Apert syndrome, Rubinstein-Taybi syndrome, diastrophic dwarfism, and triphalangeal thumbs (Fig. 4).

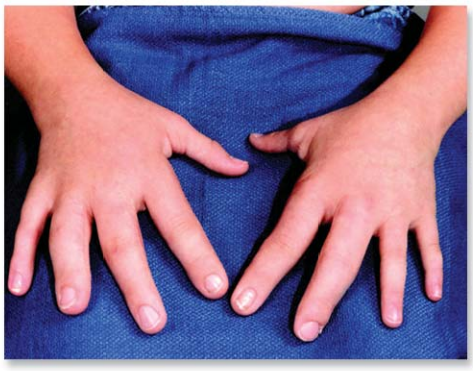


Fig. 7: A fourteen year old girl with Klippel Trenaunay-Weber syndrome and bilateral macrodactyly. The right hand has enlargement of the index, long, and ring fingers. The left hand has macrodactyly of the index and long fingers

Macrodactyly

Macrodactyly represents overgrowth of all structures of the involved digit and is different from an isolated enlargement of the bone (e.g., an enchondroma) or vessels (e.g., a hemangioma). This disfiguring condition can affect one digit or multiple fingers. The radial fingers are more commonly involved. Macrodactyly is usually an isolated abnormality, but it can occur with neurofibromatosis or Klippel-Trenaunay-Weber syndrome (limb hypertrophy, hemangiomas, and varicose veins) (Fig. 7). The etiology remains unknown, and both static and progressive forms have been observed. Static macrodactyly consists of an enlarged digit that is present at birth and grows proportionately over time. Progressive macrodactyly is more common and begins in childhood. The involved digit or digits increase in size throughout growth and stiffen during enlargement. Progressive growth persists until physeal closure occurs with skeletal maturity.

Synostosis

Synostosis is a generic term that indicates an osseous union between bones that are normally separate. Synostosis can occur as an independent entity or as part of a more general condition (e.g., complex syndactyly or ulnar deficiency). Synostosis most commonly involves the elbow and is usually not associated with systemic conditions. Radioulnar synostosis can be isolated or associated with a radial head dislocation. As is the case with camptodactyly and clinodactyly, radioulnar synostosis can be one of the physical findings a variety of syndromes, including trisomy (13 or 21) and fetal alcohol syndrome. These syndromes present numerous problems that are more compelling than the absence of forearm rotation, and this is usually the reason for a delayed diagnosis of radioulnar synostosis. In addition, shoulder and wrist motion can compensate for the lack of forearm rotation during many childhood daily activities. Even in a healthy child, a delay in presentation is common until the child begins engaging in more complex daily activities, such as catching a ball or eating soup. A careful examination is necessary to identify a lack of forearm rotation, particularly in the presence of compensatory intercarpal rotation. Mild degrees of fixed pronation or supination are well tolerated and require no treatment. Extremes of position create functional handicaps and may require a rotational osteotomy through the fusion mass to place the hand in a more functional position. Synostosis can also occur in other parts of the upper extremity. A synostosis of the radiohumeral joint can occur as part of ulnar deficiency (Fig. 1). In the hand, metacarpal transverse synostosis occurs most commonly between the ring and small fingers and is frequently bilateral (in 60% to 80% of patients).

Thumb hypoplasia

There are various grades of thumb hypoplasia, which occurs most commonly as part of radial deficiency. The underdeveloped thumb has been classified into five types to guide treatment recommendations (Table VIII).

Table VIII

Thumb deficiency classification

Type	Findings	Treatment
I	Minor generalized hypoplasia	Augmentation
II	Absence of intrinsic thenar muscles Narrowing of first web space Ulnar collateral ligament insufficiency	Opponensplasty Release of first web space Ulnar collateral ligament reconstruction
III	Similar findings to type II plus extrinsic muscle and tendon abnormalities Skeletal deficiency A: Stable carpometacarpal joint B: Unstable carpometacarpal joint	 A: Reconstruction B: Pollicization
IV	Pouce flottant or floating thumb	Pollicization
V	Absence	Pollicization

Overview

Discoveries surrounding embryogenesis, limb formation, and the human genome are directly affecting our ability to detect and manage limb anomalies. Currently, only a small aggregate of limb anomalies has been mapped to specific chromosomal segments and even fewer have been mapped to the molecular level, but the number is rapidly increasing. Further identification of an abnormal gene or set of genes will expand the role of clinical geneticists and increase the availability and clinical applicability of genetic testing for limb malformations. Detection of abnormalities is also being enhanced by high resolution ultrasound examination of the developing fetus. The identification of fetal anomalies has resulted in the field of in utero surgery. This approach is being used for relief of renal obstructions, correction of diaphragmatic hernias, and closure of myelomeningoceles. As is true of many innovative procedures, the surgery is currently high risk, but advances in investigation and technique will decrease its morbidity. Separation of a syndactyly and release of a constricting amniotic band are just a couple of possible future applications of this technique. Advances in detection and surgery will result in parallel social and ethical conflicts. These issues will require scrutiny and careful consideration with regard to use and misuse of these innovative techniques. Such steps are necessary to ensure appropriate application of modern strategies for the diagnosis and treatment of anomalous conditions.

Reference: J Bone Joint Surg Am. 2003;85:1564-1576

HEALTH NEWS

Cheap colour test picks up HIV

A cheap test which could detect even low levels of viruses and some cancers has been developed by UK researchers. The designers from Imperial College London say the device could lead to more widespread testing for HIV and other diseases in parts of the world where other methods are unaffordable. The test can be configured to a unique signature of a disease or virus - such as a protein found on the surface of the HIV. The colour of a liquid changes to give either a positive or negative result. If that marker is present it changes the course of a chemical reaction. The final result is blue if the marker is there, red if the marker is not. The researchers say this allows the results to be detected with "the naked eye". The researchers told that this method should be used when the presence of a target molecule at ultra low concentration could improve the diagnosis of disease. It is important to detect some molecules at ultra low concentrations to test cancer recurrence after tumour removal. It can also help with diagnosing HIV infected patients whose viral load is too low to be detected with current methods. Early testing showed the presence of markers of HIV and prostate cancer could be detected. However, trials on a much larger scale will be needed before it could be used clinically. The researchers expect their design will cost 10 times less than current tests which could pave the way for more widespread use of HIV testing in poorer parts of the world.

Exercising in 70s 'may stop brain shrinkage'

Exercising in 70s may stop brain from shrinking and showing the signs of ageing linked to dementia, say experts from Edinburgh University. Brain scans of 638 people past the age of retirement showed those who were most physically active had less brain shrinkage over a three year period. The study found no real brain size benefit from mentally challenging activities, such as reading a book, or other pastimes such as socializing with friends and family. When the researchers examined the brain's white matter - the wiring that transmits messages round the brain. They found that the people over the age of 70 who were more physically active had fewer damaged areas than those who did little exercise. And they had more grey matter - the parts of the brain where the messages originate. Experts already know that our brains tend to shrink as we age and that this shrinkage is linked to poorer memory and thinking. Exercise increases blood flow to the brain, delivering oxygen and nutrients to brain cells, which may be important. Or it may be that as people's brains shrink, they become less inclined to exercise. Regardless of why, experts say the findings are good news because exercise is an easy thing to do to boost health. This research re-emphasizes that it really is never too late to benefit from exercise, so whether it's a brisk walk to the shops, gardening or competing in a fun run it is crucial that, those of us who can, get active as we grow older.

Now, video game to teach kids anger management

Children with serious anger problems can be helped by a simple video game that hones their ability to regulate their emotions, a pilot study at Boston Children's Hospital has found. The connections between the brain's executive control centers and emotional centers are weak in children's with severe anger problems. Researchers at Boston Children's Hospital have developed a PC game called "RAGE Control" that teaches kids that have anger problems how to manage those emotions. Players are hooked up to a fingertip heart rate monitor while they play, and the heart rate is displayed on the screen. The game is a space shooter; the aim is to shoot the enemy ships and leave the friendly ships alone. If the player's heart rate gets too high, the controls lock and the player are unable to continue shooting until their heart rate comes down again. The aim is to teach children how to remain calm in stressful situations. Researchers at Boston Children's Hospital led by a study were 37 children age 9 to 17 divided in two groups and compared the two groups. The first group, consisting of 19 of the children, was given only standard therapy - cognitive behavioural therapy, relaxation exercises and social skills training. The second group of 18 children was given the same, but their therapy session ended with 15 minutes of playing RAGE control. After daily sessions over a 5 day period, the group that played the game was significantly better at managing their heart rate and had decreased anger scores in intensity, frequency and expression. The control group, on the other hand, showed no significant change. The research team plans another clinical trial to test whether letting children take RAGE Control home, to play with parents and siblings will increase its effect.

Reference: bbc.co.uk

Differentiation and diagnosis of tremor

Tremor is an involuntary, rhythmic, oscillatory movement of a body part. It is the most common movement disorder encountered in clinical practice. There is no diagnostic standard to distinguish among common types of tremor, which can make the evaluation challenging. However, establishing the underlying cause is important because prognosis and specific treatment plans vary considerably. History and physical examination can provide a great deal of certainty in diagnosis. The diagnosis of tremor is based on clinical information obtained from a thorough history and physical examination. For particularly difficult cases, single photon emission computed tomography to visualize the integrity of the dopaminergic pathways in the brain may be useful to diagnose Parkinson disease.

Classification

Tremors are classified as either resting or action (Table I). A rest tremor occurs in a body part that is relaxed and completely supported against gravity (e.g., when resting an arm on a chair). It is typically enhanced by mental stress (e.g., counting backward) or movement of another body part (e.g., walking), and diminished by voluntary movement of the affected body part. Most tremors are action tremors, which occur with voluntary contraction of a muscle. Action tremors can be further subdivided into postural, isometric, and kinetic tremors. A postural tremor is present while maintaining a position against gravity. An isometric tremor occurs with muscle contraction against a rigid stationary object (e.g., when making a fist). A kinetic tremor is associated with any voluntary movement and includes intention tremor, which is produced with target directed movement.

Table I

Broad classification of tremor

Tremor type	Description
Action	Occurs with voluntary contraction of muscle Includes postural, isometric, and kinetic tremors
Postural	Occurs when the body part is voluntarily maintained against gravity. Includes essential, physiologic, cerebellar, dystonic, and drug induced tremors
Kinetic	Occurs with any form of voluntary movement. Includes classic essential, cerebellar, dystonic, and drug induced tremors
Intention	Subtype of kinetic tremor amplified as the target is reached. Presence of this type of tremor implies that there is a disturbance of the cerebellum or its pathways
Rest	Occurs in a body part that is relaxed and completely supported against gravity. Most commonly caused by parkinsonism, but may also occur in severe essential tremor

Essential tremor

The most common pathologic tremor is essential tremor. In one half of cases, it is transmitted in an autosomal dominant fashion, and it affects 0.4 to 6 percent of the population. Careful history reveals that patients with essential tremor have it in early adulthood (or sooner), but most patients do not seek help for it until 70 years of age because of its progressive nature. Despite being sometimes called "benign essential tremor," essential tremor often causes severe social embarrassment, and up to 25 percent of those afflicted retire early or modify their career path. Essential tremor is an action tremor, usually postural, but kinetic and even sporadic rest tremors have also been described. It is most obvious in the wrists and hands when patients hold their arms in front of themselves (resisting gravity); however, essential tremor can also affect the head, lower extremities, and voice. It is generally bilateral, is present with a variety of tasks, and interferes with activities of daily living. Diagnostic criteria have been proposed, but none have been accepted universally. Persons with essential tremor typically have no other neurologic findings; therefore, it is often considered a diagnosis of exclusion. If the tremor responds to a therapeutic trial of alcohol consumption (two drinks per day), the diagnosis of essential tremor is assured.

Parkinsonism

Parkinsonism is a clinical syndrome characterized by tremor, bradykinesia, rigidity, and postural instability. Many patients will also have micrographia,

shuffling gait, masked facies, and an abnormal heel to toe test. Causes of parkinsonism include brainstem infarction, multiple system atrophy, and medications that block or deplete dopamine, such as methyldopa, metoclopramide (Reglan), haloperidol, and risperidone (Risperdal). Idiopathic Parkinson disease is a chronic neurodegenerative disorder; its prevalence increases with age. It is the most common cause of parkinsonism. More than 70 percent of patients with Parkinson disease have tremor as the presenting feature. The classic parkinsonian tremor begins as a low frequency, pill rolling motion of the fingers, progressing to forearm pronation/supination and elbow flexion/extension. It is typically asymmetric, occurs at rest, and becomes less prominent with voluntary movement. Although rest tremor is one of the diagnostic criteria for Parkinson disease, most patients exhibit a combination of action and rest tremors.

Enhanced physiologic tremor

A physiologic tremor is present in all persons. It is a low amplitude, high frequency tremor at rest and during action that is not reported as symptomatic. This tremor can be enhanced by anxiety, stress, and certain medications and metabolic conditions. Patients with a tremor that comes and goes with anxiety, medication use, caffeine intake, or fatigue do not need further testing.

Drug and metabolic induced tremors

Dozens of medications can cause or exacerbate tremor (Table II). Patients with new onset tremor should have a comprehensive medication review with specific attention to medications (prescribed and over the counter) started proximal to the onset of tremor.

Medications particularly prone to inducing or exacerbating tremor are those that stimulate the sympathetic nervous system (e.g., amphetamines, terbutaline, pseudoephedrine) and psychoactive medications (e.g., tricyclic antidepressants, haloperidol, fluoxetine. When medication review reveals a likely culprit, a trial off of this medication should be attempted. Metabolic causes of tremor are varied. Initial workup of tremor may include blood testing for hepatic encephalopathy, hypocalcemia, hypoglycemia, hyponatremia, hypomagnesemia, hyperthyroidism, hyperparathyroidism, and vitamin B_{12} deficiency.

Table II

Selected medications and substances that may exacerbate tremor

- Amiodarone
- Amphetamines
- Atorvastatin
- Beta-adrenergic agonists
- Caffeine
- Carbamazepine
- Corticosteroids
- Cyclosporine
- Epinephrine
- Fluoxetine
- Haloperidol
- Hypoglycemic agents
- Lithium
- Metoclopramide
- Methylphenidate
- Pseudoephedrine
- Terbutaline
- Theophylline
- Thyroid hormones
- Tricyclic antidepressants
- Valproic acid
- Verapamil

Cerebellar tremor

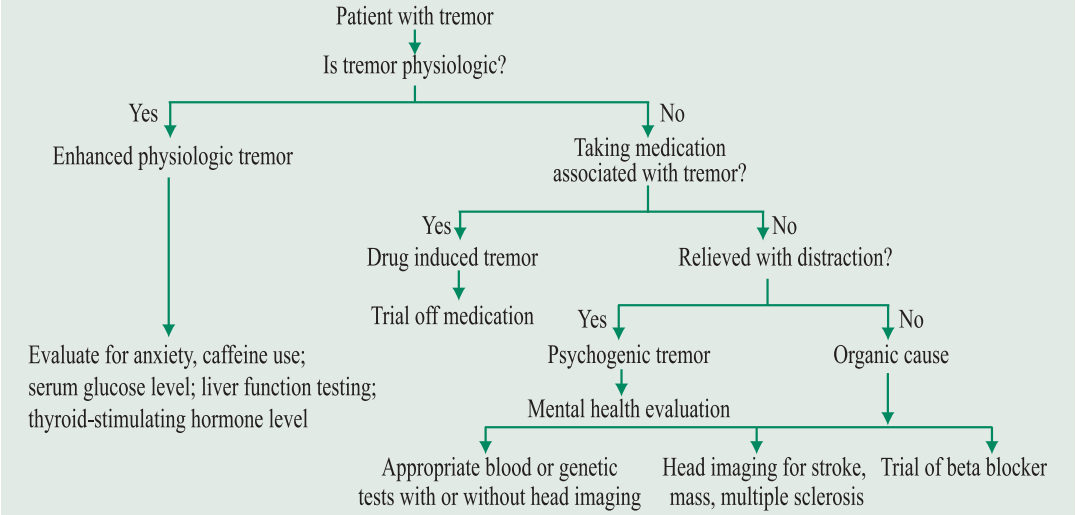
The classic cerebellar tremor presents as a disabling, low frequency, slow intention or postural tremor, and is typically caused by multiple sclerosis with cerebellar plaques, stroke, or brainstem tumors. Other neurologic signs include dysmetria (overshoot on finger to nose testing), dyssynergia (abnormal heel to shin testing and/or atraxia), and hypotonia.

Psychogenic tremor

Differentiation of organic from psychogenic tremor can be difficult. Features consistent with psychogenic tremor are abrupt onset, spontaneous remission, changing tremor characteristics, and extinction with distraction (Table III). Often, there is an associated stressful life event. Based on clinical experience, the prevalence of psychogenic tremor is thought to be high, but there are no precise estimates.

Table III

Diagnosis of tremor



Dystonic tremor

Dystonic tremor is a rare tremor found in 0.03 percent of the population. It typically occurs in patients younger than 50 years. The tremor is usually irregular and jerky, and certain hand or arm positions will extinguish the tremor. Other signs of dystonia (e.g., abnormal flexion of the wrists) are usually present.

Wilson disease

Wilson disease is a rare, autosomal recessive disorder that manifests in persons 5 to 40 years of age, sometimes with a "wing-beating" tremor. Serum ceruloplasmin level and 24 hour urinary copper excretion should be considered in young patients presenting with tremor to exclude this potentially life threatening disease.

Diagnostic approach

The diagnosis of tremor is based on clinical information obtained from a thorough history and physical examination. Although there is overlap and variability among the individual tremor syndromes, the intrinsic features of the tremor usually provide key diagnostic clues (Table III & Table IV).

Table IV

Features of common tremor syndromes

Tremor syndrome	Clinical features	Diagnostic tests	Treatment
Cerebellar tremor	Intention or postural tremor; ipsilateral involvement to lesion; abnormal finger-to-nose test; imbalance; abnormal heel-to-shin test; hypotonia	Head computed tomography or magnetic resonance imaging	Treat underlying cause, deep brain stimulation
Enhanced physiologic tremor	Postural tremor; low amplitude; use of exacerbating medication	Serum glucose level, thyroid-stimulating hormone level, liver function testing, patient history to evaluate for anxiety and caffeine use	Treat underlying cause, reassurance
Essential tremor	Postural tremor; symmetric; involves hands, wrists, lower extremities, head, or voice; family history; improvement with alcohol	No specific test; complete blood count, thyroid-stimulating hormone level, serum chemistry profile may rule out other disease	Propranolol, primidone
Parkinsonian tremor	Rest tremor; asymmetric; involves distal extremities; decreases with voluntary movement; bradykinesia, postural instability, and rigidity	No specific test; positron emission tomography or single-photon emission computed tomography for atypical presentation	Dopamine agonists, anticholinergics
Psychogenic tremor	Abrupt onset; spontaneous remission; extinction with distraction; changing tremor characteristics	Careful history	Mental health counseling

Reference: Am Fam Physician. 2011; 83(6):697-702

CASE REVIEW

Bulla in the lung

A 32 year old man presented with thoracic pain radiating to the left arm and upper dorsum, shortness of breath, and palpitations. He had had upper back tension for 6 months. Medical history was unremarkable apart from moderate nicotine use (two pack years). Echocardiography, electrocardiography, and laboratory tests were unremarkable, excluding a cardiac event. CT of the chest after chest radiography showed a large bulla of 16 cm diameter in the right hemithorax (Fig. 1). The Physicians did not detect radiological evidence of underlying pulmonary disease. The bulla wall was unremarkable and no structures were seen within the bulla. They suspected a congenital bulla and resected it to eliminate the risk of rupture with potentially life threatening pneumothorax. The air filled specimen was stapled and excised from the middle lobe (Fig. 2), which largely preserved the lobe. Histopathological examination showed disseminated bronchioalveolar carcinoma in the bulla wall which reached the resection borders. A careful second analysis of the preoperative CT of the chest did not show evidence of malignancy. MRI of the brain and CT of the abdomen were done to stage the disease, neither of which showed metastases. The Physicians then did a complete middle lobe resection including systematic mediastinal lymphadenectomy because the initial resection showed microscopic residual tumour. Full histopathological investigation showed five foci of bronchoalveolar carcinoma (minimally invasive, predominantly adeno carcinoma with lepidic pattern, thyroid transcription factor 1 positive, 2-5 mm diameter) and one well differentiated adenocarcinoma of 3 mm diameter. The tumour was negative for epidermal growth factor receptor mutation and adjuvant chemotherapy (four cycles of cisplatin with pemetrexed) was given. At last follow up, there was no evidence of tumour recurrence and the patient was in good clinical condition. Several factors have been proposed for the formation of giant bullae, including congenital pulmonary and vascular malformations (potentially accompanied by the progressive trapping of air in a check valve mechanism) and smoking. A review of over 34 cases of giant bulla with lung cancer found that all patients were male, had a mean age of 53 years, were current or former heavy smokers, and displayed various histopathological subtypes. Clinical signs for large bullae might include dyspnoea (due to compression of pulmonary tissue), palpitations, and chest pain as in this patient, or recurring infections. However, many giant bullae are clinically silent. Diagnosis with chest radiography and CT is straightforward, whereas detection of associated malignancy might be more challenging, especially if tumour growth is diffuse. Patients often receive adequate treatment only at an advanced stage. Radiographic patterns suggestive of a possible malignant process include nodular opacities within or next to the bulla, partial or multifocal thickening of the bulla wall, and secondary signs of the bulla (pneumothorax, changing diameter, fluid retention). Patients with giant bullae should receive surgical bulla resection. If resection is not possible, they should have regular follow up. Resection not only prevents potential acute complications such as pneumothorax, but also can exclude associated occult lung cancer by careful pathological examination of the resection specimen even in the absence of radiological signs for malignancy.

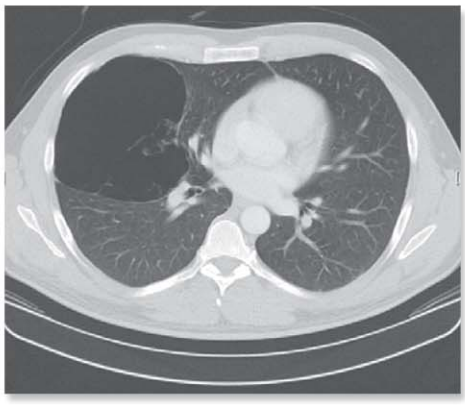


Fig 1: CT of the chest showing a large bulla of 16 cm diameter in the right hemithorax

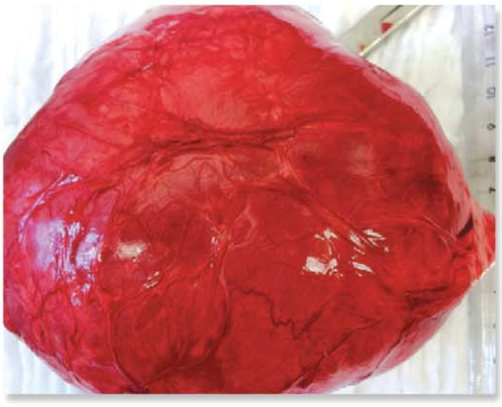
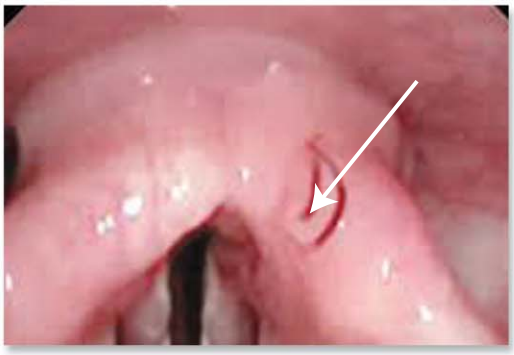


Fig 2: The resected specimen showing the air filled bulla

Reference: The Lancet October 6, 2012, Vol. 380:1280

IMAGES IN CLINICAL MEDICINE

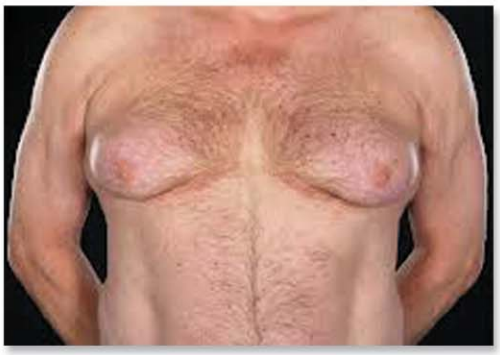
A Laryngeal fluke



A 25 year old woman presented with 1 week history of an itchy throat and cough. She reported having consumed raw carp 3 weeks earlier. The physical examination revealed hyperemia of the oral mucosa. Fiber optic laryngoscopy showed a living parasite measuring 2 mm by 4 mm on the surface of the left arytenoid laryngeal mucosa, with its head attached to the mucosa by suckers. The organism was removed with the use of a fiber clamp and was identified as an adult liver fluke (*Clonorchis sinensis*). Testing for anti *C. sinensis* IgG antibody was positive. The results of liver function testing were normal. The patient's symptoms rapidly resolved after removal of the parasite. Liver fluke infection is a zoonotic disease that is mainly caused by consumption of raw freshwater fish and shrimp throughout Southeast Asia. Liver flukes can cause disorders of the bile duct, gallbladder, liver, and rarely pharynx. Morphologic and pathological analyses distinguish adult liver flukes from *Gnathostoma spinigerum*, *Gongylonema pulchrum*, and *Paragonimus westermani*. The patient was treated with praziquantel for 2 days. Recovery was achieved over a 16 month follow up.

Reference: N. Engl. J. Med. September 27, 2012:367; 13

Gynecomastia induced by prostate cancer treatment



A 60 year old man presented to the breast clinic with gynecomastia. A high grade, advanced prostate cancer (T3N1M0; Gleason score, 9 [grade 4 plus grade 5]) had been diagnosed 4 years earlier, and the patient was treated with radiotherapy and adjuvant hormonal therapy. Hormonal treatment consisted of an androgen receptor inhibitor (bicalutamide at a dose of 150 mg daily) for 2.5 years. Six months after the initiation of treatment, progressive enlargement of the breasts developed, with pain bilaterally. After therapy was discontinued, the pain resolved; however, the size of the breasts remained static and caused the patient considerable social embarrassment. Examination of the breasts did not reveal any suspicious abnormality. Surgical correction was performed. Gynecomastia occurs in up to 80% of patients who receive nonsteroidal antiandrogens (e.g., bicalutamide, flutamide, or nilutamide), usually within the first 6 to 9 months after the initiation of treatment.

Reference: N. Engl. J. Med. September 27, 2012:367; 13

Info Quiz Participants

- Have you selected the correct answer (s) You still have time to put your entry submission together for Info Quiz Prize
- The closing date for entries is 15 February 2013
- We look forward to receiving your winning entry

Info Quiz Answers
October-December 2012

1. d	2. a	3. a	4. c	5. b
6. d	7. a	8. b	9. a	10. a

CLINICAL REVIEW

Management of renal colic

Urinary stone disease is increasingly prevalent, with a lifetime risk of about 12% in men and 6% in women. Age of onset of a first stone episode for men rises from their 20s and peaks at age 40-60 years, with an incidence of three cases per 1000 population per year. Women appear to peak a little younger in their late 20s. The male to female ratio is narrowing, with one study showing a reduction from 1.7:1 to 1.3:1 over a five year period. Presentation varies according to geographical and seasonal factors, with higher incidences in warmer climates and during the summer months.

What are urinary stones?

Urinary stones are formed by the aggregation of crystals with a non crystalline protein (matrix) component. These crystals clump together to form a stone and then move when they reach a certain size and pass down the ureter, frequently causing colic symptoms. Eighty per cent of stones contain calcium, most commonly in the form of calcium oxalate (60%). Calcium phosphate accounts for 20% of stones, with uric acid forming approximately 7%, although this uric acid proportion may rise in obese patients. Another 7% are infection stones containing magnesium ammonium phosphate. Bladder stones usually have a different cause, often as a result of bladder outflow obstruction.

Who gets urinary stones?

The incidence of stones is higher in warmer climates, owing to a combination of dehydration and sun exposure (vitamin D). Obesity is also a risk factor, with large epidemiological studies showing both high body mass index and weight as independent risk factors for stones. There is a 2.5 times greater risk if a patient has a family history of stone disease. This increase is probably a genetic predisposition but may also be due to similar environmental factors such as dehydration and diet. Any anatomical abnormality of the urinary tract (such as a horseshoe kidney) indicates a higher risk of stone formation, as well as several medical disorders such as primary hyperparathyroidism, renal tubular acidosis, myeloproliferative disorders, all chronic diarrhoeal conditions (for example, Crohn's disease), and gout. Occupations involving work in a hot environment (for example, kitchen workers) are also at risk due to dehydration. Previous stone formation is a risk factor, with a 30-40% chance of forming a second stone within five years of the initial episode. Both observational studies and a randomised trial (compared with control) have shown the importance of fluid intake patients producing less than 1 L of urine per day are at highest risk of stone formation, while producing 2 L of urine per day substantially reduces the risk of stone episodes.

What is renal colic?

Renal colic describes the pain arising from obstruction of the ureter, although ureteric colic would be a more accurate term. The pain is caused by spasm of the ureter around the stone, causing obstruction and distension of the ureter, pelvicalyceal system, and renal capsule. Although the most common cause is a stone, the term "renal colic" actually refers to a collection of symptoms attributed to the kidney and ureter. There are other intrinsic or occasionally extrinsic causes such as lymphadenopathy, although extrinsic causes tend to present with milder and more chronic discomfort. Other common intrinsic causes are blood clots (from upper tract bleeding) or sloughed renal papilla (which can occur in sickle cell disease, diabetes, or long term use of analgesics).

What are the symptoms of renal colic?

The classic presentation of renal colic is the sudden onset of severe loin pain (in the costovertebral angle, lateral to the sacrospinus muscle, and beneath the 12th rib), often described as akin to labour pains. Depending on the site of obstruction, the pain will radiate to the flank, groin, and testes or labia majora. This pain can be a useful method of judging the level of obstruction. If a stone is at the vesico-ureteric junction (VUJ), the patient may often complain of strangury (the urgent desire to pass urine with poor volumes, urinary frequency, and straining) due to irritation of the detrusor muscle from the stone. Nausea with vomiting is common. The pain is a colic, and thus comes in waves of varying intensity. Patients will often have completely pain free spells between attacks. Furthermore, they are often restless and cannot get comfortable, by contrast with peritonitic conditions in which patients remain still. Visible haematuria may occur, but in these cases it is important to ensure that the pain is not secondary to a clot as a result of other upper tract pathology. If concomitant urinary infection is present, the patient could complain of fevers and sweats. The Table I lists possible differential diagnosis.

Table I

Differential diagnosis of renal colic	
Differential	**Features in history and examination**
Pyelonephritis	Fever and tender kidney (obstruction with sepsis is an emergency; if obstruction is suspected, immediate imaging is required)
Musculoskeletal pain	Worse with movement
Appendicitis	Tenderness or peritonism in right iliac fossa
Cholecystitis	Worse with eating fatty foods, tenderness in right upper quadrant
Diverticulitis	Associated bowel symptoms, usually tender in left iliac fossa
Leaking abdominal aortic aneurysm	Older age, vascular risk factors
Testicular torsion	Tender testis on examination
Gynaecological problems (for example, ovarian pathology, ruptured ectopic pregnancy)	Younger age, pelvic pain

What investigations are needed?

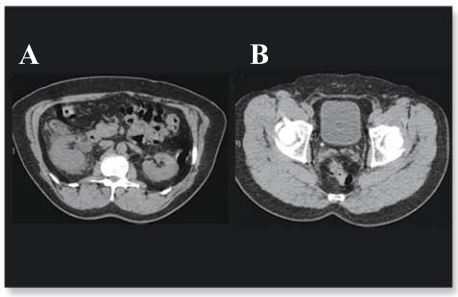


Fig. 1: Representative axial slices of the kidneys, ureter, and bladder for renal colic, using NCCT. (A) Normal contralateral (left) kidney, with hydronephrosis of the right kidney and peri-nephric fat stranding indicating obstruction. (B) Distal ureteric stone (4 mm in diameter) with peri-uretericoedema (rim sign) differentiating it from a phlebolith; the dilated ureter could also be traced down to the stone

Urine investigations

Expert guidelines from the British Association of Urological Surgeons (BAUS) and College of Emergency Medicine state that all patients should have a urine dipstick documented. However, the sensitivity of haematuria in patients with ureteric stones is about 90%, and 40% of patients presenting with acute flank pain and haematuria do not have urolithiasis. Expert opinion suggests that the diagnosis and decision on whether to perform imaging should not be based solely on the presence or absence of haematuria. The presence of leucocytes and nitrites would support a diagnosis of urine infection, and the expert guidelines state that a midstream urine sample should be sent for culture.

Blood investigations

Expert guidelines state that determining full blood counts (for white cell counts) and renal function should be considered, which is mandatory in patients with pyrexia or a single functioning kidney. Patients with proven stone formation should have basic metabolic studies measuring serum calcium and urate.

Imaging

Most patients assessed in the emergency department will proceed to immediate imaging for a definitive diagnosis and management plan. However, if the pain has already resolved, there is no sepsis, and the patient has a normal contralateral kidney, immediate imaging is not mandatory and urgent imaging and review can be organised. The exact timing of this approach will depend on local availability, but it is advantageous to make a firm diagnosis and management plan. In the only trial that analysed this, researchers arranged imaging within two to three weeks, although other expert consensus opinion suggests a timeframe of seven days.

Non contrast CT

Non contrast CT (NCCT) (Fig. 1) has become the imaging method of choice for investigating acute flank pain. Several studies have shown consistently better results from NCCT than from intravenous urograms (IVUs; Fig. 2) (NCCT sensitivity 94-100% and specificity 92-100% v IVU 51-87% and 92-100%, respectively).

Radiation doses for NCCT can be reduced to similar levels as IVU by using a low dose protocol while maintaining diagnostic accuracy. NCCT has other benefits over IVUs, including speed of the test, detection of other pathology, and eliminating risks of nephrotoxicity or of allergic or anaphylactic reactions from the intravenous contrast. Guidelines from the BAUS, European Association of Urology (EAU), and American Urological Association recommend NCCT as the definitive investigation. If the stone is visible on the scout film, a plain radiograph of the kidneys, ureter, and bladder is not required, although such a radiograph would be needed if the stone was not visible, to assess visibility for directing the modality of follow up imaging.

Ultrasonography

Ultrasonography is a cheap alternative as a primary diagnostic screening tool, particularly in thinner patients. It is good at identifying stones (particularly those >5 mm in diameter) within the pelvicalyceal system.

Patients should be scanned with a full bladder to identify stones at the VUJ. However, stones elsewhere in the ureter (between the pelvi-ureteric junction (PUJ) and VUJ) are unlikely to be seen. Secondary signs, such as dilatation, which may suggest an obstructing stone, improve the test's sensitivity. It is the first line test in pregnancy and children.

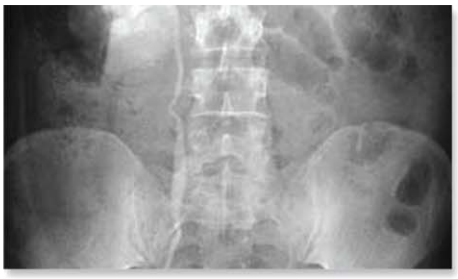


Fig. 2: IVU, 1 h after contrast injection, showing a normal left kidney and dilated right pelvicalyceal system. The ureter is dilated all the way to the lower ureter (where a small calculus was visible on plain imaging). The image should be taken after micturition to allow the ureter to be traced to the bladder

Plain radiography of the kidneys, ureter, and bladder

Plain radiography of the kidneys, ureter, and bladder could be useful, with a sensitivity of 44-77% and specificity of 80-87%. Detection rates of radiography and ultrasonography combined could approach those of CT if in the most experienced hands, but a NCCT will be required if uncertainty remains. The combined approach is certainly reasonable if CT is not immediately available or if radiation dosing needs to be minimised.

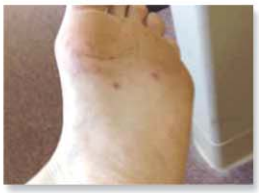
What treatment options are available for the stone?

When to actively treat a stone will depend on size, location, ongoing symptoms, local availability, and patient preference. Most units will conservatively manage patients with stones smaller than 10 mm, controlled pain, normal renal function, and no signs of sepsis. If stones have not passed within four to six weeks, they are unlikely to do so, and very few pass after eight weeks. Patient preference is paramount, especially in situations where commitments necessitate a predictable clinical course or they intend on foreign travel. A patient's intention to travel would be an indication for prompt stone treatment, and patients with ureteric stones should be advised of the risks of developing colic during a flight. If the flight had to be diverted, patients could be liable for medical costs since their insurance policy would probably be invalidated. Indications for initial active treatment of stones are low chance of spontaneous passage, persistent pain, ongoing obstruction, and renal insufficiency. If coexistent infection is present at admission for treatment, renal drainage only should be conducted. The main treatment options are extracorporeal shockwave lithotripsy (ESWL) or ureteroscopy. ESWL is usually an outpatient procedure performed with analgesia or sedation. A shockwave is generated and focused on the stone. The procedure is generally well tolerated but is not available in all urology units, and could require more than one treatment. Ureteroscopy is typically done under general (or spinal) anaesthesia. Usually a rigid or semi-rigid ureteroscope is used, although evidence suggests that flexible ureteroscopy has better clearance rates for upper ureteric stones. The vast majority of stones will be cleared in one treatment, but an indwelling stent may be required for some time afterwards. Both procedures have high success rates for all ureteric stones. Only rarely would other more invasive surgical options be used, such as percutaneous antegrade ureteroscopy (involving direct puncture into the kidney) or laparoscopy, which is usually reserved for large stones (>15 mm), and only if other options have failed or are not suitable.

Reference: BMJ 01 September 2012 Vol. 345: 30-35

DERM DILEMMA

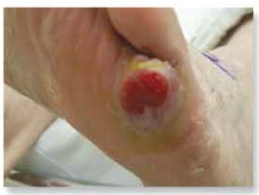
CASE 1



A 23 year old graduate student presents with an eruption on her hands and the soles of her feet that began acutely 3 days prior. She developed a fever, slight loss of appetite, and an oral blister 2 days before the onset of the rash. She has had recent contact with children. Examination reveals scattered macules on her feet and vesiculopapules on her fingers. Slightly erythematous erosion is noted on her tongue. Lymph nodes are non palpable.

What is your diagnosis?

CASE 2



An 83 year old man with a painful growth beneath his left fifth toe is treated by his podiatrist with dressing changes weekly for several weeks with no improvement. Most recently, the lesion has begun to bleed spontaneously. The patient has emphysema and a persistent cough. He denies a history of skin cancer and is a lifelong smoker. Examination reveals a friable, firm, erythematous nodule beneath the fifth toe which is tender to palpation. Serous drainage is also evident. Scattered actinic keratoses of his face and hands are noted as well.

What is your diagnosis?

Answers 21

Reference: Eme. Med. September 2012, Vol. 44, No.6:11-12

CLINICAL METHOD

Emergency pericardiocentesis

Overview

Placement of peripheral intravenous (IV) catheters is a fundamental skill that all health care professionals should possess. Unfortunately, it can be difficult to obtain IV access in some patients, including obese patients, children, and patients who have undergone placement of many IVs or who have a history of difficult IV access. Ultrasound guidance enables visualization of veins that are not apparent on physical examination, resulting in fewer needle sticks, more rapid cannulation, and less discomfort in patients with difficult IV access.1-3 Ultrasound guided IV placement is indicated when it is difficult or impossible to use traditional techniques. There is no contraindication to ultrasound guided IV placement, but it is unnecessary when traditional techniques are sufficient. Health care providers who are proficient in standard placement techniques will find it relatively easy to learn ultrasound guided techniques. There are two common approaches to the position of the transducer during ultrasound guided placement of IV catheters: transverse and longitudinal. The transverse approach is easier to learn, but the longitudinal approach is preferred by many experienced providers because it allows better visualization of the needle. It is best to learn both techniques. Potential cannulation sites are the hand, the antecubital veins of the forearm, and the basilic, cephalic, and brachial veins of the upper arm.

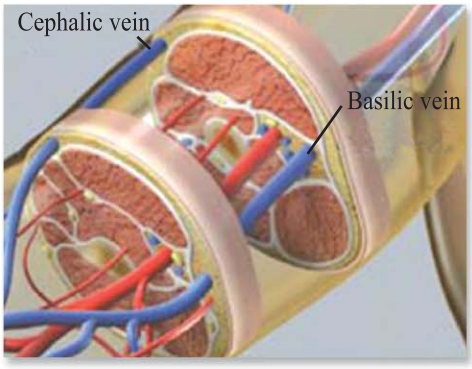


Fig. 1 : Insertion of the spinal needle in the subxiphoid approach

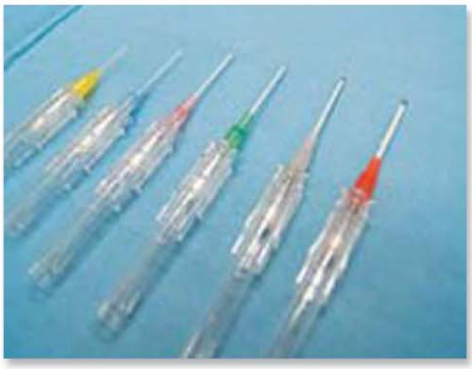


Fig. 2 : Advancement of the needle toward the left shoulder

Preparation

To get started, the physicians should wash hands, adhere to universal precautions, clean the ultrasound transducer with a germicidal solution, place a tourniquet, and apply sterile gel or surgical lubricant to the transducer. Then the physician has to adjust the gain and depth settings of the ultrasound system so that vessels appear black and are in the center of the ultrasound image. Find an appropriate vein by scanning the arm in the transverse orientation, which provides a cross sectional view of the anatomy and allows simultaneous visualization of veins, arteries, and other structures (Fig. 1). Vessels appear as circular structures, and veins are easily distinguished from arteries because veins collapse with compression. When choosing a vein, the physicians have to remember that a vein with a relatively large diameter is more likely to result in successful catheterization. The depth of a vein is not as important as its diameter, but longer IV catheters are needed for veins that lie more than 1 cm from the surface of the skin. Standard IV catheters are 3.2 cm long (Fig. 2). Common sizes range from 24 gauge for newborns to 14 gauge for adult patients with trauma. Angiocatheters that are 6.4 cm long are adequate for cannulating vessels that are a maximum of about 2 cm from the surface of the skin. Specialized catheters may be best for cannulation of deeper veins.

Placement with the transverse technique

The physicians have to make sure that the left side of the transducer correlates with the left side of the ultrasound image. Identify the target vein and note the depth of the vein. Clean the skin with an antiseptic swab. Insert the needle through the skin at a 45 degree angle. Advance the needle toward the vein and look for the needle tip. Since the needle is perpendicular to the ultrasound plane as it passes through the skin, it appears as just a single bright dot with posterior shadowing or a repeating artifact (Fig. 3). By advancing the needle tip, follow it by fanning or tilting the transducer in the direction that the needle is traveling. If the physician cannot identify the needle tip, look for compression and movement of the adjacent soft tissue and tenting of the anterior wall of the vein. Bouncing, or vibrating, the needle will enhance movement of the soft tissue adjacent to the needle and may help to identify the position of the needle tip. The physician has to watch for blood return as an indication that the needle tip has entered the vein. Decrease the angle of the needle, and advance it by an additional 1 or 2 mm to make sure that the entire needle tip and the tip of the catheter are inside the vein. While holding the needle still, advance the catheter over the needle and into the vein. Secure the catheter in the standard fashion and flush with saline.

Placement with the longitudinal technique

The physician has to identify the target vein and surrounding structures using the transverse view, and then rotate the transducer 90 degrees to the longitudinal axis of the vein. It is crucial to know which side of the ultrasound image represents the distal vein, because that is where the needle will enter the image. Anchor the hand on the patient to

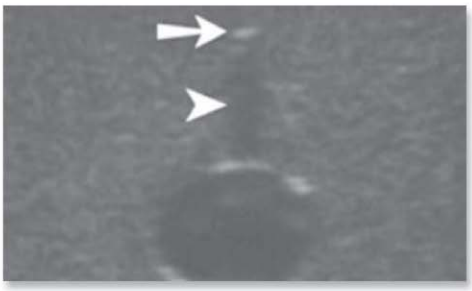


Fig. 3: In this ultrasound image showing the transverse approach to IV placement, the needle tip appears as a single bright dot (arrow); posterior shadowing (arrowhead) helps to identify the tip

keep the transducer completely still during the procedure, so not to lose site of the vein while advancing the needle. Clean the skin with an antiseptic swab. Insert the needle through the skin at precisely the center of the long axis of the transducer, at an angle of 30 degrees, and carefully advance it within the ultrasound plane. The needle should be well visualized in the long axis view (Fig. 3). If the physician can see the vein but not the needle, then the needle must be lateral to the vein. Back up and redirect the needle as necessary. Do not advance the needle unless the physician can visualize the tip. The anterior wall of the vein may become compressed in response to needle contact, so use a quick forward motion to puncture it. Visualize the needle tip inside the vein and watch the catheter as it is advanced off of the needle and into the vein. Secure the catheter and flush with saline.

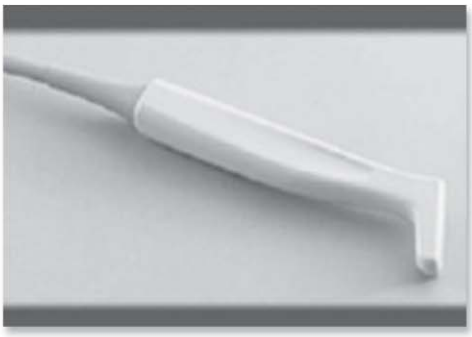


Fig. 4: These linear array transducers have a frequency of 5 to 13 MHz. They are ideal for ultrasound guided vascular access

Complications

Overly deep placement of the needle is a common problem, especially when the transverse technique is used. Centimeters are marked along the edges of all ultrasound images to indicate the depth of scanning in relation to the skin surface. Ergonomics is important and should not be overlooked. The patient's arm should be positioned on a flat horizontal surface, so the ultrasound images and needle movements are both in the vertical plane. Adjust the position and height of the patient and the ultrasound monitor so that the physician can work comfortably, without bending or stooping or having to turn the head to see the ultrasound images. When catheters do not flush well after insertion, consider checking the longitudinal view to diagnose the problem. Transducer selection is important. Linear-array transducers with high frequencies (5 to 14 MHz) are specifically made for ultrasound guided procedures (Fig. 4).

Summary

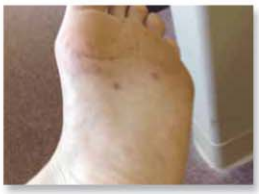
Ultrasound guidance facilitates placement of peripheral IV catheters when standard techniques fail. Learning ultrasound guided techniques is relatively easy, especially for providers who are proficient in the placement of standard IV catheters. Small linear transducers and long IV catheters are ideal for ultrasound guided IV catheter placement.

Reference: N. Engl. J. Med. June 21, 2012:366:15

DERM DILEMMA

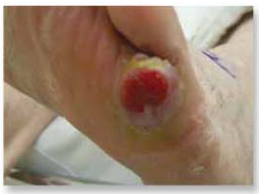
ANSWER

CASE 1



Hand, foot, and mouth disease (HFMD) is a contagious disorder that primarily affects infants and children younger than 10 years. Occurrence in an adult is uncommon. The cause is an enterovirus, usually coxsackievirus A16. A prodrome consisting of fever, malaise, and sore mouth is followed by macules and vesicles arising on the hands and feet. Shallow ulcers dot the buccal mucosa and tongue. The condition is usually self limiting, with spontaneous resolution within a week's time. In 2012, the Centers for Disease Control and Prevention documented more than 60 cases of HFMD with more severe illness, some requiring hospitalization due to dehydration or severe pain. The majority of these patients had recent exposure to children at a school or a child care facility.

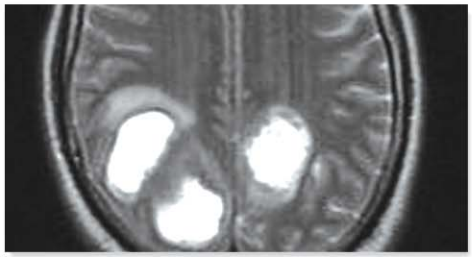
CASE 2



Amelanotic nodular melanoma may mimic both benign and less aggressive malignant lesions, resulting in delayed diagnosis and treatment. The classic ABCDs (asymmetry, borders [irregular], color [variegated], diameter >6 mm) that characterize the most common superficial spreading variant of melanoma are usually absent. Nodular melanomas typically present as rapidly expanding nodules or plaques. Histopathology in this case revealed a deeply invasive Clark level V tumor with a 9.6 mm Breslow thickness. Following lobectomy for unrelated lung cancer, the patient underwent left lateral foot amputation. Sentinel lymph node biopsy was negative and the patient is said to be doing well 18 months after surgery.

CLINICAL FOCUS

Intracranial neoplasms



In the developed world, cerebral tumours account for 2% of deaths at all ages. The majority are metastatic from malignancies outside the nervous system.

Primary and secondary brain tumours

Common sources of metastases from extracranial primary tumours are bronchus, breast and GI tract. Primary intracerebral tumours are classified by their cell of origin and degree of malignancy (Table I). Even when malignant, they do not metastasise outside the nervous system.

Clinical features

- Headache is not invariable
- There may be symptoms of raised intracranial pressure
- Focal deficits are of slow onset and progressive
- The nature of the deficit depends on the site of the tumour
- Seizures are caused by tumour cells infiltrating an area of the cerebral cortex which excites seizure activity

Table I

Primary intracranial tumours

Histopathological type	Common site	Age
Malignant		
Glioma (astrocytoma)	Cerebral hemisphere Cerebellum Brain stem	Adulthood Childhood/adulthood Childhood/young adulthood
Oligodendroglioma	Cerebral hemisphere	Adulthood
Medulloblastoma	Posterior fossa	Childhood
Ependymoma	Posterior fossa	Childhood/adolescence
Cerebral lymphoma	Cerebral hemisphere	Adulthood
Benign		
Meningioma	Cortical dura, parasagittal, Sphenoid ridge, Suprasellar, olfactory groove	Adulthood
Neurofibroma	Acoustic neuroma	Adulthood
Craniopharyngioma	Suprasellar	Childhood/adolescence
Pituitary adenoma	Pituitary fossa	Adulthood
Colloid cyst	Third ventricle	Any age
Pineal tumours	Quadrigeminal cistern	Childhood (teratomas) Young adult (germ cell)

Management

Medical: Dexamethasone (8 mg 12 hourly) lowers intracranial pressure by resolving the reactive oedema. In severely raised intracranial pressure, 16-20 mg of dexamethasone or 200 ml of 20% mannitol may be infused. Seizures may treated with anticonvulsants. Prolactin or growth hormone secreting pituitary tumours may respond to dopamine agonists.

Surgical: Surgery is the mainstay of treatment. Only partial excision may be possible if tumour is inaccessible or if removal will cause unacceptable brain damage. Biopsy should be considered even if tumour cannot be removed (histology has implications for management). Meningiomas and acoustic neuromas offer the best prospects for complete removal. Pituitary adenomas may be removed by a trans-sphenoidal route, avoiding craniotomy.

Radiotherapy and chemotherapy: These have only a marginal effect on cerebral metastases and malignant gliomas in adults. Radiotherapy reduces recurrence of pituitary after surgery. Radiotherapy may be an adjunct to surgery for meningiomas which cannot be completely excised or whose histology suggests an increased tendency to recurrence.

Reference: Davidson's Essentials of Medicine

INFO QUIZ

Jog your memory

Please select the correct answer by (√) against a, b, c, d & e of each questions in the Business Reply Card and send it through our colleagues or mail within 15 February 2013; this will ensure eligibility for the Raffle Draw and the lucky winners will get attractive prizes!

1. **A 50 year old woman presented with a 24 hour history of palpitations. An ECG revealed atrial fibrillation with a ventricular rate of 130 beats per minute. Which drug is most likely to restore sinus rhythm?**
   a. Adenosine
   b. Bisoprolol
   c. Digoxin
   d. Flecainide
   e. Verapamil

2. **A 27 year old woman presented with a right sided thyroid swelling with associated cervical lymphadenopathy. What is the most likely cause?**
   a. Anaplastic carcinoma
   b. Follicular adenoma
   c. Follicular carcinoma
   d. Hashimoto's thyroiditis
   e. Papillary carcinoma

3. **A 32 year old man was treated with combination chemotherapy for testicular cancer. Subsequent investigations confirmed a complete clinical remission. What is the dominant cellular process that explains why this therapy was successful?**
   a. Apoptosis
   b. Differentiation
   c. Mutagenesis
   d. Necrosis
   e. Senescence

4. **A 47 year old woman was being treated with lithium for bipolar affective disorder. On examination, her blood pressure was 168/104 mmHg. What drug is the most appropriate antihypertensive?**
   a. Amlodipine
   b. Bendroflumethiazide
   c. Doxazosin
   d. Losartan
   e. Ramipril

5. **A 37 year old woman with breast cancer had a family history of breast and ovarian cancer. Molecular genetic testing revealed a BRCA1 mutation. What is the normal function of BRCA1?**
   a. Angiogenesis
   b. Apoptosis
   c. Cell adhesion
   d. Promotion of mitosis
   e. Tumour suppression

6. **A 20 year old woman presented 24 hours after taking an overdose of 80 tablets of thyroxine 100 μg. On examination, she was clinically euthyroid. What is the most appropriate treatment?**
   a. β-adrenoceptor blockade
   b. Forced alkaline diuresis
   c. Haemodialysis
   d. No treatment
   e. Stomach washout

7. **The half life of a novel anti obesity drug exhibiting first order kinetics was calculated to be 4 hours. What percentage of the drug will be eliminated 20 hours after ingestion?**
   a. 75%
   b. 80%
   c. 90%
   d. 97%
   e. 100%

8. **On removal of the renal arterial clamp following a donor kidney transplantation, the surgeon noted changes suggestive of hyper acute rejection. Which immunoglobulin is likely to be responsible?**
   a. IgA
   b. IgD
   c. IgE
   d. IgG
   e. IgM

9. **A 46 year old man presented within 1 hour of ingesting 40 tablets of slow release theophylline. What is the most appropriate initial management?**
   a. Activated charcoal
   b. Alkaline diuresis
   c. Gastric lavage
   d. Observation only
   e. Whole bowel irrigation

10. **A 45 year old woman with immune thrombocytopenia failed to respond to corticosteroid therapy. Splenectomy was planned. What is the optimum time for pneumococcal vaccination?**
    a. 1 month after surgery
    b. 1 month before surgery
    c. 1 week after surgery
    d. 1 week before surgery
    e. Perioperatively

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