Brachydactyly, Macrodactyly, Amniotic band syndrome
Brachydactyly
Brachydactyly

- Short finger where all the elements of digital skeleton are present but one or more reduced in size
Brachydactyly

- Brachytelophalangy: distal
- Brachymesophalangy: middle
- Brachybasophalangy: proximal
- Brachymetacarpia: metacarpal
Epidemiology

- Commonly as a feature of congenital hand anomalies
- Rarely the dominant feature, inherited anomaly
- Noncongenital – injury to growth plate from trauma, infection, frostbite
Pathology

Ch2q          Brachydactyly E
Ch9q22         Brachydactyly B
Indian hedgehog gene       Brachydactyly A1
Growth differentiation factor 5      Brachydactyly C
Cartilage derived morphogenetic protein
Modified Bell classification

- Type A
  - A1 Farabee
  - A2 Mohr-Wreidt
  - A3 Bauer – clinodactyly
  - A4 Temtamy
  - A5 Bass
- Type B Mackinder
- Type C Drinkwater
- Type D Breitenbecher (Stub thumb)
- Type E Bell
- Other
• Middle phalanges short, rudimentary or fused with distal phalanges
• Short proximal phalanx thumb and big toe
A2 – Mohr- Wriedt

- Rarest
- Short index finger and 2\textsuperscript{nd} toe
A3 – Bauer (clinodactyly)

• Short middle phalanx with ulnar drift
A4 – Temtamy

- Middle phalanges of IF, SF
- Radial deviation of distal phalanges
- Feet often involved
A5 – Bass

- Absent middle phalanges, hypoplastic nails & toes
Type B Mackinder

- Hypoplasia of distal phalanges IF-SF
- Absent fingernails
- Feet involved but less severe
- Occasional syndactyly
Type C Drinkwater

- Middle phalanges IF and MF short
- Middle phalanx SF triangular and short
- Ulnar deviation IF
- Hyperphalangism of IF and MF
- RF uninvolved and stands out as longest digit
Type D Breitenbecher (stub thumb)

- Distal phalanx of thumb
- Base is broader than head of prox phalanx
- Early closure of growth plate
Type E Bell

- Short metacarpals, normal length phalanges
- +/- metatarsal shortening
Clinical features

• Severity of digital shortening variable

• MC phalanx – middle

• MC digits – small and index
<table>
<thead>
<tr>
<th>Table 40-3</th>
<th>SYNDROMES ASSOCIATED WITH BRACHYDACTYLY</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Generalized Brachydactyly</strong></td>
<td><strong>D Brachydactyly (Stub Thumb Brachydactyly)</strong></td>
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<tr>
<td>Acradynplasia</td>
<td>Tabatznik's syndrome</td>
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<tr>
<td>Hypochondroplasia</td>
<td>Rubinstein-Taybi syndrome</td>
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<tr>
<td>Diastrophic dwarfism</td>
<td>Robinow's syndrome</td>
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<tr>
<td>Metachondroplasia</td>
<td></td>
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<tr>
<td>Multiple epiphyseal dysplasia</td>
<td></td>
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<tr>
<td>Spondyloepiphyseal dysplasia</td>
<td></td>
</tr>
<tr>
<td>Metaphyseal dysostosis</td>
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<tr>
<td>Peripheral dysostosis</td>
<td></td>
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<tr>
<td>Dyschondroplasia</td>
<td></td>
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<tr>
<td>Hereditary multiple exostosis</td>
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<tr>
<td>Ollier's disease</td>
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<tr>
<td>Weill-Marchesani syndrome</td>
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<tr>
<td>Ellis-van Creveld syndrome</td>
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<tr>
<td>Orofaciodigital syndrome</td>
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<tr>
<td><strong>Underdevelopment of Distal Phalanges</strong></td>
<td><strong>Brachydactyly with Metacarpal Shortening</strong></td>
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<tr>
<td>Pyknodysostosis</td>
<td>Turner's syndrome</td>
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<tr>
<td>Cleidocranial dysostosis</td>
<td>Albright's hereditary osteodystrophy</td>
</tr>
<tr>
<td>Fanconi's anemia</td>
<td>Pseudohyoparathyroidism</td>
</tr>
<tr>
<td>Progeria</td>
<td>Pseudopseudohyoparathyroidism</td>
</tr>
<tr>
<td>Larsen's syndrome</td>
<td><strong>Brachydactyly with Short First Metacarpal</strong></td>
</tr>
<tr>
<td>Kent's syndrome</td>
<td>Holt-Oram syndrome</td>
</tr>
<tr>
<td>Pulger's syndrome</td>
<td>Fanconi's anemia</td>
</tr>
<tr>
<td>Coffin-Siris syndrome</td>
<td>Progressive myositis ossificans</td>
</tr>
<tr>
<td><strong>A3 Brachydactyly (Short Middle Phalanx of Little Finger)</strong></td>
<td><strong>Brachydactyly with Polydactyly</strong></td>
</tr>
<tr>
<td>Down syndrome</td>
<td>Ellis-van Creveld syndrome</td>
</tr>
<tr>
<td>Poly X syndrome</td>
<td>Orofaciodigital syndrome</td>
</tr>
<tr>
<td>Russell-Silver syndrome</td>
<td><strong>Brachydactyly with Syndactyly</strong></td>
</tr>
<tr>
<td>Coffin-Siris syndrome</td>
<td>Cornelia de Lange syndrome</td>
</tr>
<tr>
<td>Orofaciodigital syndrome (types I and II)</td>
<td>Apert's syndrome</td>
</tr>
<tr>
<td>Otopalatodigital syndrome</td>
<td><strong>Brachydactyly with Cone-Shaped Epiphyses</strong></td>
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<tr>
<td>Thrombocytopenia-absent radius syndrome</td>
<td>Trichohiphahalangeal syndrome</td>
</tr>
<tr>
<td>Noonan's syndrome</td>
<td>Langer-Giedion syndrome</td>
</tr>
<tr>
<td><strong>Miscellaneous</strong></td>
<td>Acrodyostosis</td>
</tr>
<tr>
<td>Du Pan's syndrome</td>
<td><strong>Miscellaneous</strong></td>
</tr>
<tr>
<td>Hand-foot-uterus syndrome</td>
<td></td>
</tr>
</tbody>
</table>
Management

• Lengthening of shortened digit
  – Immediate with bone grafting
  – Gradual distraction
Distraction osteogenesis

- Concomitant soft tissue lengthening
- Up to 15 mm without need for graft
- Time consuming, requires cooperation of patients, parents
- Ex-fix for up to 4 months for consolidation
Management

• Short phalanges
  – Usually avoid surgery
    • Function preserved
    • Lengthening difficult w/ stiff, contracted digits
  – Immediate lengthening if associated deformity (angulation)
Management

• Short metacarpal
  – Distraction lengthening
    • Transverse osteotomy, wound closed
    • Distraction begins 4-7 days
    • .25-.5 mm twice a day
    • Frame removed when consolidation of metacarpal

  – Immediate lengthening
Buck-Gramcko
Macrodactyly
Macrodactyly

• Disproportionately large digit noted at birth or within first years
• Uncommon, usually sporadic
• Sometimes a feature of neurofibromatosis
Pathology

- Etiology unknown
  - Abnormal nerve supply > unimpeded growth
  - Increased blood supply
  - Abnormal humeral mechanism stim growth

** MC type associated with fatty infiltration and enlargement of nerve

*Nerve territory oriented macrodactyly*
- **Palmar** aspect more affected than dorsal
- **Distal** more enlarged than proximal
- Tendon normal but **sheath** enlarged
- Thickened skin, fatty infiltration of nerve w/ endo and perineural fibrosis
- **Exostoses**
Growth rate

- **Static** – (macrodactyly simplex congenita)
  - Present at birth
  - In proportion during child’s growth

- **Progressive** – (macrodactyly dystrophia lipomatosa progressiva)
  - More common
  - Not apparent until 2 y/o
  - Disproportionate and rapid growth
  - Involves palm (swelling of median nerve) by 3-4 y/o
### Classification of Macrodactyly

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Gigantism and lipofibromatosis</td>
</tr>
<tr>
<td></td>
<td>Associated with enlarged, fat-infiltrated nerves within the digit and extending proximally through the carpal tunnel. The most common form.</td>
</tr>
<tr>
<td>II</td>
<td>Gigantism and neurofibromatosis</td>
</tr>
<tr>
<td></td>
<td>Usually occurs in conjunction with plexiform form of neurofibromatosis and is often bilateral. May be osteochondral masses associated with the enlarged skeleton.</td>
</tr>
<tr>
<td>III</td>
<td>Gigantism and digital hyperostosis</td>
</tr>
<tr>
<td></td>
<td>Hyperostotic form with osteochondral periarticular masses developing in infancy. No significant nerve enlargement. Very rare and not hereditary. Nodular and stiff digits, may be other skeletal anomalies.</td>
</tr>
<tr>
<td>IV</td>
<td>Gigantism and hemihypertrophy</td>
</tr>
<tr>
<td></td>
<td>Rare anomaly without known inheritance pattern or etiology. Macrodactyly part of hemihypertrophy. All digits involved, but less severe than type I or II. Deformity marked by intrinsic muscle hypertrophy or abnormal intrinsic anatomy. Presents as flexion contracture, ulnar deviation, and an adducted thumb deformity.</td>
</tr>
</tbody>
</table>
Type I - Lipofibromatosis

- MC type
- Usually unilateral
- Multiple digital enlargement 2-3 x MC than single digit
- Preference for median n. territory
  - IF 37%
  - LF 30%
  - Thumb 18%
  - RF 12%
  - SF 3%
- If all digits eval for hemihypertrophy
- Radial deviation
Lipofibromatosis

- Osseous growth until physeal closure
- Soft tissue growth into adulthood

- Soft tissue swelling palm > compression neuropathy
- Thickening of flexor sheath > triggering
Type II - Neurofibromatosis

• 1:2500-3300 live births

• Neurocutaneous syndrome
  – Six or more café-au-lait spots
  – Fibromatous skin tumors
  – Multiple neurofibromomas on peripheral nerves
Type III - Hyperostosis

- Nerves normal, however follows median nerve territory
- Nodular transverse enlargement
- Bilateral
- Rapid loss of motion secondary to periarticular mass formation ~ puberty
Type IV - Hemihypertrophy

- Enlargement of digits less severe
- Deformities
  - Flexion contractures (MPJ)
  - Ulnar deviation
  - Adduction deformity of thumb
  - Abnormal muscle origins / insertions
- Vascular insufficiency or malformations
- Association w/ renal, adrenal and brain tumors
| **Limiting Growth** | Digital nerve stripping  
|                     | Epiphysiodesis |
| **Digit Reduction** | Soft tissue debulking  
|                     | Skeletal terminalization  
|                     | Reposition nail unit on shortened skeleton  
|                     | Palmar pedicle (Barsky procedure) (Barsky, 1967; Flatt, 1977)  
|                     | Dorsal pedicle (Tsuge procedure) (Tsuge, 1967)  
|                     | Nail island flap (Rosenberg, 1983)  
|                     | Resection of distal portion of nail and pulp  
|                     | Tsuge (Tsuge, 1985)  
|                     | Hoshi (Hoshi, 1973)  
|                     | Fujita (Fujita, 1983)  
|                     | Bertelli (Bertelli, 2001) |
| **Correction of Deviation** | Closing wedge osteotomy (combined with epiphysiodesis as required) |
| **Thumb Macrodactyly** | Metacarpophalangeal arthrodesis |
| **Amputation** | Ray amputation (with transposition of digit for central ray amputation) |
Limiting digital growth

- Ligation of digital arteries
- Compression bandaging
- Stripping digital nerves
- **Epiphysiodesis**
  - When digit is adult length of same sex parent
  - Deviation corrected with closing wedge osteotomy
Reducing the digit

- Debulking, one side at a time
- Midlateral incision
- Neurovascular bundle dissected
- Fat, excess skin excised
- Narrowing of skeleton
  - Burring or longitudinal ostectomy
- Osteotomy to correct deviation or shorten
Reducing the digit

- Preservation of nail with shortening
  - Barsky
  - Flatt
  - Tsuge
  - Rosenberg

- Ray amputation
Macrodactyly management

- **Type I & II**
  - Early debulking w/ nerve stripping
  - Epiphyseal fusion, length reduction when similar size digit of parent
- **Type III**
  - Early excision of masses, palmar plate before joint motion blocked
  - Arthroplasty / arthrodesis
- **Type IV**
  - Night splinting, stretching (prevent contractures)
  - Debulking grossly hypertrophic intrinsics
  - Extensor tendon centralization
Macrodactyly

- Devastating anomaly
- Multiple procedures required with usually unsatisfactory results...
- Consider ray amputation early
Amniotic band syndrome
Amniotic band syndrome

• AKA
  – Amniotic disruption sequence
  – Constriction ring syndrome

• Partial or complete circumferential constrictions around limbs or digits
Amniotic band syndrome

• 1:5000-15,000
• Associated w/ (20-50%)
  – Talipes equinovarus (clubfoot)
  – Cleft lip, palate
  – Hemangioma
  – Meningocele
  – Cranial or cardiac defects
Etiology

• Rupture of amniotic membrane
• Part or all of fetus lies outside amnion, between it and chorionic membrane
  
  < 45 days – abortion
  
  > 45 days – severe limb abnormalities

• Amniotic disruption
  
  – Release of amniotic bands that encircle and strangulate limbs or parts of limbs
Clinical features

• Circumferential or incomplete and can occur anywhere on body
• Proximal structures normal
• Pseudosyndactyly
  – Distal digit fusion
  – Proximal sinus cleft
Clinical features

• Deep structures affected
  – Veins, lymphatics > edema
  – Nerve > motor, sensory defects
  – Bone > skeletal narrowing
<table>
<thead>
<tr>
<th></th>
<th>Classification of Ring Constriction Syndrome</th>
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<tbody>
<tr>
<td>1</td>
<td>Simple constriction rings</td>
</tr>
<tr>
<td>2</td>
<td>Rings accompanied by distal deformity, with or without lymphedema</td>
</tr>
<tr>
<td>3</td>
<td>Rings accompanied by distal fusion: acrosyndactyly</td>
</tr>
<tr>
<td></td>
<td>(a) Type I Tips are joined</td>
</tr>
<tr>
<td></td>
<td>(b) Type II Tips are joined: webs too distal</td>
</tr>
<tr>
<td></td>
<td>(c) Type III Tips are joined: no web. Complex syndactyly with proximal sinus</td>
</tr>
<tr>
<td>4</td>
<td>Amputation</td>
</tr>
</tbody>
</table>
Management

• Simple ring constriction
• Complicated ring constriction
  – Distal edema
  – Two rings close together
  – Rings with deep structural damage
• Ring constriction with syndactyly
• Ring constriction with intrauterine amputation
  – Stump refashioning
  – On-top plasty
  – Free toe transfer
Simple ring constriction

- Excise deep part of constriction
- Z-plasty
Complicated ring constriction

• Distal edema
  – Indicates venous / lymphatic obstruction
  – **Urgent** surgical treatment
  – Z-plasty

• Two rings close together
  – If won’t interfere with Z-plasty do at same time
  – Tx one at time, if distal edema perform second within weeks
Complicated ring constriction

- Rings with deep structural damage
  - [Early] correction to limit neurological damage
  - Explore nerves to evaluate for fascial bands
Ring constriction with intrauterine amputation

- Structures proximal to constriction normal ~ reconstruction
- Stump refashioning
Ring constriction with intrauterine amputation

• On-top plasty
  – Limited role
  – Sacrifice one ray to augment another
  – Usually for thumb reconstruction – such as 2\textsuperscript{nd} metacarpal when IF missing

• Microvascular free-toe transfer
  – Thumb when absent at MP level