

OSTEOLOGY - Birth Defects & Malformation

Powerpoint presentation (*not available online*)

Teratogenesis: The embryonic period (weeks 1-8) is a critical time with respect to external factors (drugs, disease, toxins) that harm (insult) developing fetuses.

Variation in Vertebral #

- deviation from 7C:12T:5L:5S
- *Lumbarization* of S1
- *Sacralization* of L5

Supernumerary Ribs - cervical ribs cause problems with brachial plexus & subclavian vessels

Variation in Appendicular Skeleton

- *Polydactyly*: supernumerary digits (radial/fibular side)
- *Syndactyly*: fusion of digits (webbing)
- *Meromelia*: absence of portions of limb
- *Amelia*: absence of limbs

Talipes equinovarus (*club-foot*)

- distorted ankle/foot due to limb position *in utero*

Mandibulofacial dysostosis - (MFD)

Family of gross malformations (dysplasias) of face/throat

a. *Microstomia, Microtia, Anotia, Hemifacial microsomia:*

- Gross malformations of the jaw/ear

b. *Treacher Collins syndrome:*

- Dysplasias of ear, maxilla/mandible

c. *Pierre Robin syndrome:*

- Dysplasias of ear/eye, mandible, cleft palate

Anencephalus - Hypoplasia of brain/stem, normal face

Hydrocephalus - accumulation of fluid (CSF) expands the fetal brain. Bone material is limited so that vault is thin/brittle with predictable deformations of the face.

Cleft Palate & Facial clefts - facial bones fail to fuse together correctly. Errors may be slight (cleft palate) or may present clinically as dramatic facial clefts.

Spina bifida - varying degrees of vertebral arch defects

Craniosynostosis - premature sutural fusion

- e.g., *Scaphocephaly & Cranioostenosis*

Growth errors = Pituitary gland (*sella tursica*) GH

- *Hyperpituitarism*: Pituitary giants: (proportional)
= excessive growth hormone during puberty
- *Hypopituitarism*: Pituitary dwarfs: (proportional)
= insufficient growth hormone during puberty
- *Acromegaly*: (over-growth - disproportionate)
= excessive growth hormone **AFTER** puberty

Achondroplasia - achondroplastic dwarfism (congenital)

- cartilages in braincase/long bones do not grow correctly
- premature fusion of epi-/diaphyses, floor of braincase
= short bones, prominent forehead, retracted nasal bridge

Maintenance of bone mass:

Calcification: deposition of calcium (not 'bone!')

Ossification: generation of bone matrix that is **then** calcified

- 1) **Exercise** maintains bone structure and mass
- 2) **Dietary** calcium, phosphate, Vitamins A & C
- 3) **Vitamin D** (steroid): (calcium+phosphate absorption)
 - Synthesis starts w/ UV light (skin) completed in kidney
- 4) **Hormones**:
 - a. *Parathyroid* - increases calcium levels in the blood
 - b. *Calcitonin* - decreases calcium levels in the blood
 - c. *Thyroxine* - stimulates bone growth
 - d. *Estrogen/Testosterone* - retard growth of epiphyses

Degenerative bone diseases - bone-matrix degrades:
kyphosis, scoliosis, compression fractures

Osteopenia : @ 30-40 yr. osteoblasts (hormonal) become less active, bone becomes thin/weak

Osteoporosis: when bone mass decreases to the point where normal function is compromised

Osteopathic scoliosis = abnormal spinal curvature

Scurvy - Vit. C-def.: weak/brittle bone – *Telescoped bones*

Rickets = Vit. D-def.: weak/flexible bone - *Bow-legged*

Gout = *Hyperuricemia* (metabolic disorder)

- urate crystals cause inflammation which destroys joints

Osteophyte - artifact of trauma/healing:

- bone spurs in the annulus fibrosae of the spine

- within CT of injured joints, synovial sheaths & bursae

Types of fractures

1. Closed/Open: completely internal/projects through skin
2. Simple/Compound - single/multiple regions
3. (non)Displaced - orientation of broken ends
4. Transverse - clean break across shaft of bone
5. Spiral - twisting break that spirals along shaft
6. Greenstick - incomplete fracture (children)
7. Epiphyseal - fracture along epiphyseal plate/line
8. Comminuted - telescoping of multiple bone fragments
9. Compression - crushed along long axis (vertebrae)
10. "Hip fractures"

Fracture repair

1. Bleeding - Hematoma (clot) seals off injured area
2. Osteoclasts/-blasts rapidly produce daughter cells
3. Bone calluses bridge gaps between fragments
4. Dead bone fragments digested away by osteoclasts
5. Osteoclasts/-blasts remodel site for 4-12 months

The following is supplemental reading for your own information. It is not officially assigned reading. However, these topics would be considered fair game for extra credit (bonus) questions.

Epithelium & Development: Development is a dynamic orchestration of cell proliferation, cell/tissue function, and interaction. More cells will die during the morphogenesis (growth of size and shape) of the body than are left over for the adult body! As a generalization, the most important morphogenetic events in the head (whole body for that matter) concern the development and organization of simple sheets of cells — Epithelia. However, epithelial tissues are capable of exhibiting only the most simple morphogenetic constructions: sheet-folding and pocket formation. These limitations place further restrictions on subsequent epithelio-mesodermal interactions, yet they express a wide range of constructions (e.g., including hair, nails, sweat glands, mammary glands, eyeballs, brain, and gut). Spatial competition among these various soft-tissues and incipient musculo-skeletal elements is the primary determinant of shape, position, surface modeling, and orientation of all foramen, nerves, bony elements and organs in the head.

Skull Evolution - Original theories of skull growth (and evolution) conceived a plan by which the vertebrae elongated anteriorly so as to enclose the brain in bone and cartilage. Indeed, Goethe (1791) proposed that the skull was little more than a transformed series of cervical vertebrae. This concept was abandoned in favor of evidence that indicated that the head was a relatively new invention created by neural crest cells as they migrate ahead of the brain and gut tube early in development. More recently, a great deal of genetic/developmental evidence indicates that the occiput is derived from somatic mesoderm and that the hypoglossal nerve and canal are remnants of the primitive first intervertebral foramina and spinal nerve. Goethe was correct in 1791!

Chondrocranium vs. Osteocranium - The chondrocranium is formed of cartilage and acts as a foundation or bracket along which the primary sensory capsules are arranged, i.e., nasal (nose/mouth), optic (eye), and otic (ear) capsules. As the chondrocranium expands and ossifies (endochondral ossification), cartilage "growth plates" are left between the ossifications (synchondroses) which function like the epiphyseal plates found in the long bones of the appendicular skeleton (also endochondral ossification). Growth of these plates extends the basicranium in an anterior-posterior axis and literally forces the facial component of the skull anteriorly and inferiorly away from the brain case (see Achondroplasia). The floor of the braincase (basicranium) is formed around the chondrocranium. The rest of the skull is formed predominantly by bone that is derived by an intramembranous ossification. That is, bone forms within the connective tissue sheets that enclose the important organs of the head, e.g., eyes, pharynx, brain. These intra-membranous bones are usually flattened (squama - squamous) and consist of an inner and outer table sandwiching a spongy matrix of bone (diploe) in between. This diploe is eventually pneumatized to form the various para-nasal sinuses in the head. Early on, the membranous spaces (fontanelles) among the various squama (facial and brain case components) allow for the expansion of the underlying soft tissues. Eventually, the fontanelles decrease in size leaving smaller expansion zones called sutures that will themselves eventually fuse together after growth of the underlying tissues has ended.

"Hand-me-downs" - Our human evolutionary history goes back five million years, long before the likes of Vesalius, Galen, or Netter came up with the absurd convention of the "anatomical position". By understanding evolutionary and developmental mechanisms, we can better understand why certain adult structures aren't particularly well designed or are "wired" or "plumbed" in an indirect manner. Each bit of your anatomy (plumbing, electrical, structural, motor) is placed under great physiological or physical stresses at some point during its development. That is, parts are not built so that they will only work in the adult body, instead, they must function at each stage of development. From this perspective, the adult is left only with that which the embryo found useful. This mechanism can be easily traced through an individual's own development, but provides some rather interesting insights into structures that are "hand-me-downs" from our vertebrate ancestry. For example: If tissue beds or other building materials are somehow left unused, those materials will be borrowed by adjacent regions and incorporated into new structures - i.e., they have been "cannibalized". An example is found in the trapezius muscle that evolved from what had been a hammock of tissue that supported the throat. One key to its ancestry (its migration from the neck, and its new function as an extrinsic shoulder muscle) lies in the fact that the trapezius is innervated by CN XI. Another example can be found in the minute bones (auditory ossicles) in our inner ears...these materials have been recycled from elements that contribute to the jaw joint when you were a very early embryo.