THE RADIOGRAPHIC MANIFESTATIONS OF THE SKELETAL DYSPLASIAS

A Bit of “Little”
THE RADIOGRAPHIC FIRST APPROACH

TERMINOLOGY/METHODOLOGY:

- PROPORTIONATE/DISPROPORTIONATE SHORT STATURE
- RHIZOMELIC; MESOMELIC; ACROMELIC; MICROMELIA

NEXT: A QUICK CURSERY LOOK AT EPIPHYSES/METAPHYSSES ETC. FOR ...

EPIPHYSEAL DYSPLASIA, METAPHYSEAL DYSPLASIA, DIAPHYSEAL DYSPLASIA; SPONDYLO- … COMBINATIONS

NOTE: THERE ARE MORE THAN 450 WELL DESCRIBED SKELETAL DYSPLASIAS, OF THESE OVER 300 HAVE THE MUTATION/GENE IDENTIFIED.
THEN,
EVERY BONE IN THE SKELETAL SURVEY MUST BE INDIVIDUALLY ANALYSED LOOKING FOR POSSIBLE DIAGNOSTIC/CHARACTERISTIC FEATURES
CLASSIFICATIONS

- RADIOGRAPHIC [ORIGINAL] vs
- MOLECULAR [RECENT]
- [LUMPERS vs SPLITTERS]
IT IS VERY IMPORTANT FOR US NOW, IN ORDER TO BE HELPFUL TO THE CLINICIAN TO BE AWARE OF THE MOLECULAR & RADIOGRAPHIC GROUPS
WITH THE ADVENT OF MOLECULAR/GENE TESTING, AS ONE ENTITY IS MOLECULARLY DEFINED, ONE CAN PRESUME OTHER ENTITIES THAT HAVE SIMILAR RADIOGRAPHIC FINDINGS COULD SUGGEST ALLELIC SKELETAL DYSPLASIAS!

MOLECULAR ENTITIES IN POINT:

- TRPV4-OPATHIES
- TYPE II COLLAGEN-OPATHIES
BECAUSE THERE ARE OFTEN SO MANY RADIOGRAPHIC ABNORMALITIES WE NEED TO ANALYSE SUCCESSFULLY W/ KEY TERMS

SO LET US BEGIN...

SOME COMMON SKELETAL DYSPLASIAS
 THANATOPHORIC DYSPLASIA I
  MOST COMMON LETHAL SKELETAL DYSPLASIA
 THANATOPHORIC DYSPLASIA II
 ACHONDROPLASIA
  MOST COMMON NON LETHAL SKELETAL DYSPLASIA
 HYPOCHONDROPLASIA
THAN I & THAN II HAVE DIFFERENT FGFR 3 MUTATIONS
THAN 1

- KEY DIAGNOSTIC TERMS
- FRENCH TELEPHONE RECEIVER FEMURS
- H, u, n SHAPED VERTEBRAE/PEDICLES
U-shaped T-SPINE
H-shaped upper L-SPINE
VERTEBRAE IN THE AP PROJECTION
MICROMELIA
SEVERE
PLATYSPONDYLY
ANTERIOR ROUNDED PLATYSP. VERTEBRAL BODIES
“OLD FASHIONED” TELEPHONE RECEIVER FEMUR
THAN 2

- STRAIGHT, MICROMELIC FEMURS / MEDIAL SPIKE
- KLEEBLATTSCHADEL [CLOVERLEAF SKULL]
KLEEBLATTSCHADEL
CLOVERLEAF SKULL
RATHER
STRAIGHT
FEMURS

MEDIAL SPIKE

“FADE –OFF”
PROXIMAL
FEMUR
ACHONDROGENESIS 2
HYPOCHONDROGENESIS
SPONDYLO-EPIPHYSEAL DYSPLASIA CONGENITAE [SEDC]
SEMD-STRUDWICK
KNIEST DYSPLASIA
STICKLER DYSPLASIA
FEW OTHERS, VERY RARE

NOTE: AS A GROUP THEY HAVE OVERLAPPING FEATURES
ACHONDROGENESIS 2

- Severest form [lethal] of COL II defect
- Pronounced vertebral ossification defects:
  - Absent, hypoplasia, platyspondyly
- Absent epiphyseal equivalent: [pubic, ischial, talus/calcaneal] ossification [at 24-26 weeks gestation plus]
- True micromelia
- Occipital ossification defect
NEAR TERM FETUS

HYPOPLASTIC VERTEBRAL BODIES
NO SIGNIFICANT ISCHIAL & PUBLIC OSSIFICATION
PROPORTIONATE SHORTENING W/ LARGER HANDS/FEET
LARGE POSTERIOR OCCIPITAL OSSIFICATION DEFECT
Obviously even more radiographic absences & micromelia w/o skull [membranous bone] involvement
SPONDYLO–EPIPHYSEAL DYSPLASIA CONGENITA  [SEDC]

- OVOID OFTEN HYPOPLASTIC VERTEBRAL BODIES
- OCCIPITAL OSSIFICATION DEFECT
- FLAT ACETABULAR ROOF
- ABSENT EPIPHYSEAL, EPIPHYSEAL EQUIVALENT OSSIFICATION:
  - PUBIC BONES, TALUS/CALCANEOUS
N’L SKULL/FACE

OCCIPITAL OSSIFICATION DEFECT

CERVICAL VERTEBRAL HYPOPLASIA

TERM INFANT
ABSENT
CERVICAL/SACRAL
VERTEBRAL
OSSIFICATION
OVOID LUMBAR VERTEBRAL BODIES

MODERATELY SHORT RIBS
ABSENT PUBIC & EPIPHYSEAL KNEE OSSIFICATION

TERM INFANT
TINY TALO/CALCANEAL OSSIFICATION

TERM INFANT
Sulfation Disorder Group

- Diastrophic Dysplasia
- Achondrogenesis 1B
- Atelectosteoogenesis 2
- Med [Ar] Multilayered Patella, Brachydactyly, Clubbed Feet
- SEMD, Omani Type [Very Rare]
- SEMD, Pakistani Type [Very Rare]
DIASTROPHIC DYSPLASIA

- OVOID/SMALL 1ST METACARPAL
- ACCESSORY CARPAL CENTERS
- TWISTED METATARSALS
- EAR PINNA OSSIFICATION
- CERVICAL KYPHOSIS
- DOUBLE LAYERED MANUBRIUM/PATELLA
DIFFICULT DX IN NEW BORN

NO SPECIFIC CHANGES ON

THIS RADIOGRAPH
NEED HAND/FOOT RADIOGRAPHS

HYPOPLASTIC 1ST METACARPAL W/O A HITCHHIKER THUMB
CLUB FEET
TWISTED METATARSALS
HITCH HIKER GREAT TOE
SHORT RIB [WITH / WITHOUT POLYDACTYLY] GROUP [SRP]

- SRP 1-3
- SRP 2-BEEMER [W/o POLYDACTYLY]
- ATD [JEUNE] DYSPLASIA
- ELLIS-VAN CREVELD [EVC] DYSPLASIA

MOLECULARLY QUITE COMPLICATED

*NEED TO MAKE A SPECIFIC DX
EXTREMELY SHORT HORIZONTAL RIBS
WITH/WITHOUT POLYDACTYLY
IRREGULAR/POINTED/SPURRED METAPHYSICAL ENDS OF LONG BONES [ESPECIALLY FEMURS]
VERY SHORT RIBS

METAPHYSICAL SPURS ON BOTH ENDS OF FEMURS
SAME AS SRP 1-3 EXCEPT:

- ROUND METAPHYSICAL BONE ENDS
  W/O SPURS
AGAIN, VERY SHORT RIBS

ROUND FEMORAL METAPHYSSES
MODERATELY SHORT RIBS
TRIDENT ACETABULUM
NARROW SACROSCIATIC NOTCHES
HANDS:
- METAPHYSEAL WIDENING/CUPPING LATER DEVELOPING INTO CONE EPIPHYSES

*PROX FEMORAL SCLEROSIS/CUPPING AT A LATER AGE
MODERATELY SHORT RIBS
TRIDENT [3-PRONGED] ACETABULUM
PROXIMAL PHALANGEAL CUPPING

MIDDLE PHALANX HYPOPLASIA, “TOMB-STONE” APPEARANCE
4 YO

CUPPING, CONE-SHAPED EPIPHYSSES
4 YO

TRIDENT
ACETABULUM

PROX. FEMORAL
METAPHYSICAL
SCLEROSIS
MULTIPLE EPIPHYSEAL DYSPLASIA [MED] GROUP*

- PSEUDOACHONDROPLASIA
  - AD; COMP GENE
- MED RIBBING/FAIRBANKS [MILD/SEVERE]
  - GENES:
    - COL 9A 1, 2, 3- AD
    - MATRILLIN- AD
    - COMP- AD
    - DIASTROPHIC DYSPLASIA GENE- AR

*COMPLICATED GROUP [VERTEBRAL BODY CHANGES ALSO PRESENT] THE GROUP ALL TOGETHER IS QUITE COMMON
NOTE:

THE AD FORMS OF MED DO NOT HAVE TRUE BRACHYDACTYLY
PELVIS/HIPS: ACETABULAR ROOF HYPOPLASIA, MINI-EPIPHYSSES

SPINE: SUPERIOR/INFERIOR ROUNDED VERTEBRAE, MIDDLE TONGUE

KNEES: SMALL EPIPHYSSES, METAPHYSICAL WIDENING & EXCESSIVE SLANTING/SLOPING

HANDS: BRACHYDACTYLY, VERY SMALL EPIPHYSSES, METAPHYSICAL WIDENING/CUPPING, SMALL HYPOPLASTIC CARPAL BONES
MINI-EPIPHYSES

OSSIFICATION
DEFECT OF
ACETABULAR
ROOF
ROUNDED VERT. BODIES

ANTERIOR TONGUE PROJECTION
VERY SMALL EPIPHYSEAL OSSIFICATIONS
KNEE METAPHYSEAL SLOPING
IN CONCLUSION

I HOPE THIS TALK WILL HELP YOU TO ANALYZE SKELETAL DYSPLASIAS TO HELP THE CLINICIAN TO A CORRECT DX. AT LEAST TO PLACE THE PATIENT IN THE CORRECT GROUP OF DISORDERS
MAJOR REFERENCES

- TAYBI & LACHMAN’S RADIOLOGY OF SYNDROMES, METABOLIC DISORDERS & SKELETAL DYSPLASIAS
  RS LACHMAN, 5TH EDITION MOSBY/ELSEVIER 2007

- BONE DYSPLASIAS, SPRANGER ET AL, 2ND EDITION OXFORD PRESS 2002
REFERALS TO THE
INTERNATL SKELETAL DYSPLASIA
REGISTRY

SAMANTHA ALON
REGISTRY COORDINATOR

UCLA - OHRC

PHONE : (310) 825 8998
FAX : (310) 206 5266

E-MAIL : SAlon@mednet.UCLA.edu