



# A Case Study of Skeletal Dysplasia Inheritance and Maternal/Fetal Health from a Middle Woodland Context at the Elizabeth Site (11PK512), Illinois

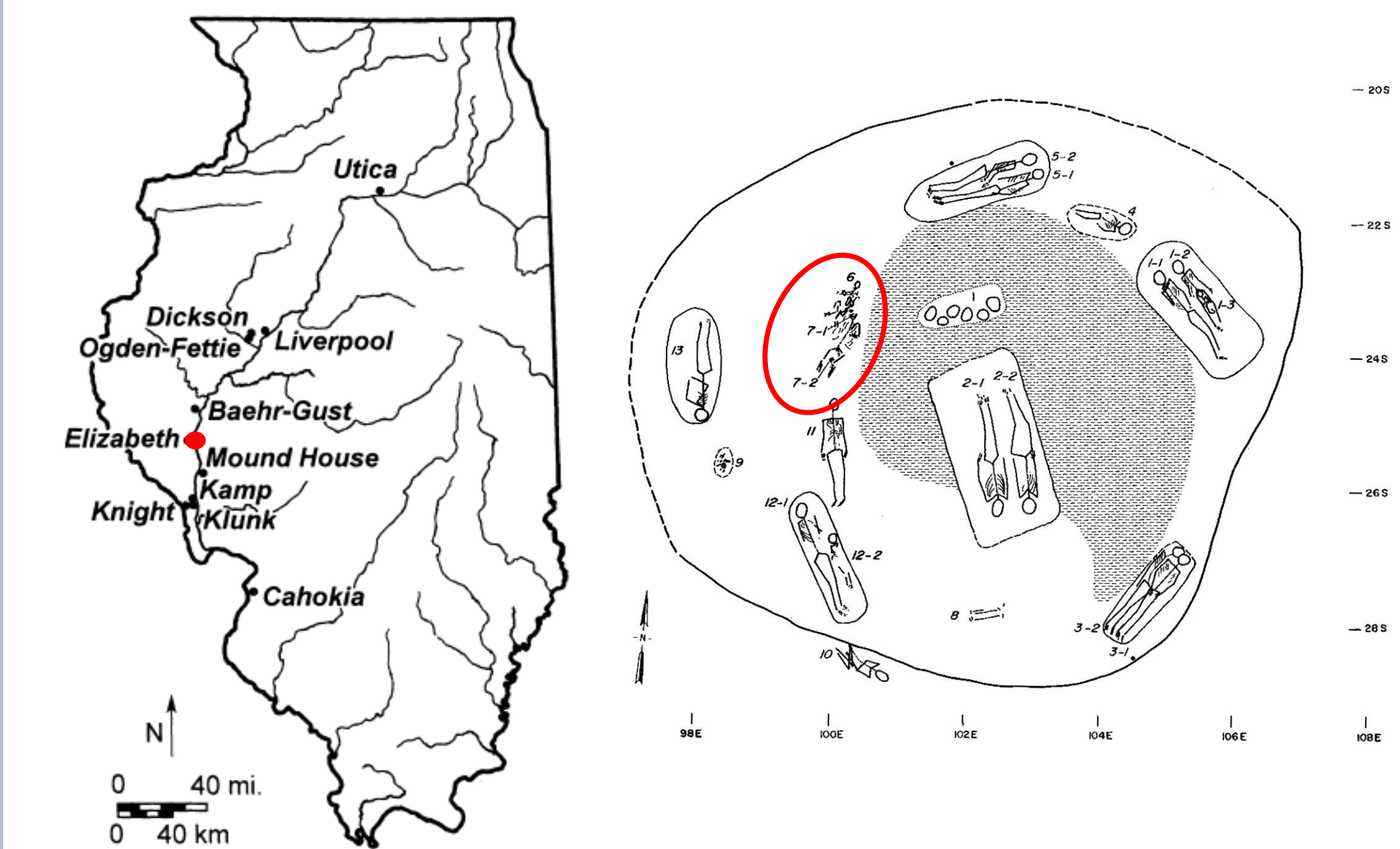
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## INTRODUCTION

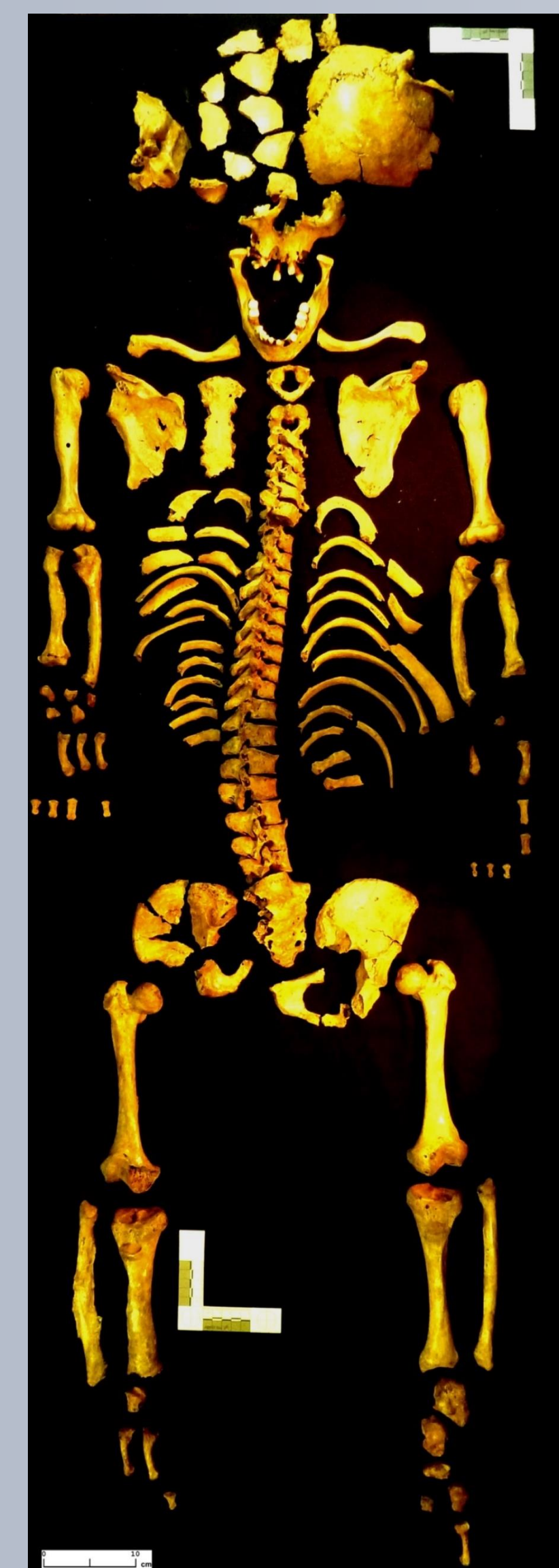
Two individuals, an adult (EZ 3-7-1) and a fetus (EZ 3-7-2), were excavated from Mound 3 at the Elizabeth site (11PK512) in Pike County, Illinois, by the Center for American Archeology Contract Archeology Program and the Northwestern University Archeological Field Schools in 1980. A radiocarbon analysis of the bone from EZ 3-7-1 reported a calibrated range of AD 132-388, with the calibrated median date of AD 268, indicating a Middle Woodland context (King et al., 2011).



Adapted from Charles et al., 1988 & Van Nest et al., 2001

Following a paleopathological evaluation, it was determined that EZ 3-7-1 had a combined skeletal dysplasia of achondroplasia and Leri-Weill Dyschondrosteosis, as well as pervasive abnormal bone addition. The skeletal remains of EZ 3-7-2 were disturbed by postmortem taphonomic processes, but were found situated in breech position on the sacrum of EZ 3-7-1, suggesting that EZ 3-7-1 was pregnant at the time of death, or perhaps died during childbirth. This poster presents the examination of the fetal remains, which revealed: the near full term development of EZ 3-7-2; abnormal, active, woven bone addition on the ulnae, femora, and tibiae; and abnormal morphological development of the long bones and cranial elements recovered. This study explores inheritance of skeletal dysplasias, systemic physiological disruption in fetuses, and the possible role of fetuses and neonates as indicators of maternal health in the bioarchaeological record.

## EZ 3-7-1: ADULT FEMALE



EZ 3-7-1

### Combined Skeletal Dysplasia of Achondroplasia and Leri-Weill Dyschondrosteosis

- Short stature
- Disproportionally large, bulbous, brachycephalic skull and a small foramen magnum
- Reduced interpedicle distance, spinal stenosis, lumbar lordosis, postural kyphosis
- Incomplete elbow extension, Madelung's deformity, and Cubitus Valgus
- Mesomelia of the upper limb
- Rhizomelia of the lower limb

### Abnormal Periosteal Addition

- Woven bone in various stages of activity
  - Vault and face, the ribs, clavicles, scapulae, radii, ulnae, femora, tibiae, fibulae, and the right 2nd and 3rd metatarsals
- Extensive sclerotic addition on right tibia and fibula (from fracture?)
- Osteomyelitis, primary periostitis, or treponematosi

## EZ 3-7-2: PERINATE

### Age Estimation

- Petromastoid and squamotypanic parts of temporal not fused: less than 1 year (Scheuer & Black, 2000)
- Calcified Deciduous Incisor: older than 3 fetal months (Schuurs, 2013)
- Bone Measurements: 30 to 36+ fetal weeks (Fazekas and Kosa, 1978)

Selected Measurements	Avg. (mm)	Age (fetal wks)
Occip Basilaris Max Width	11.10	30-34
<b>Occip Basilaris Sagittal Length</b>	<b>12.12</b>	<b>38</b>
L Occip Lateralis Max Length	17.72	32
L Occip Lateralis Max Width	11.35	34-40
L Temporal Petrosa Length	29.17	32-34
L Temporal Petrosa Width	14.09	30-32
<b>Sphenoid Body Length</b>	<b>8.28</b>	<b>28-30</b>
<b>Sphenoid Body Width</b>	<b>10.88</b>	<b>22-24</b>
C1 Max Length Arch	12.02	32-38
C2 Max Length Arch	12.87	30-32
L Radius	41.18	32-34
L Ulna	48.37	32-34
<b>L Femur Length</b>	<b>49.31</b>	<b>28-30</b>
<b>R Femur Length</b>	<b>49.90</b>	<b>28-30</b>
R Femur Distal Width	14.64	32-34
L Tibia	45.21	30-32
L Ischium Max Length	11.53	30-34
L Ischium Max Width	7.75	30-32
<b>R Ilium Max Length</b>	<b>23.83</b>	<b>28-32</b>
<b>R Ilium Max Width</b>	<b>16.95</b>	<b>24-26</b>

### Abnormal Morphology

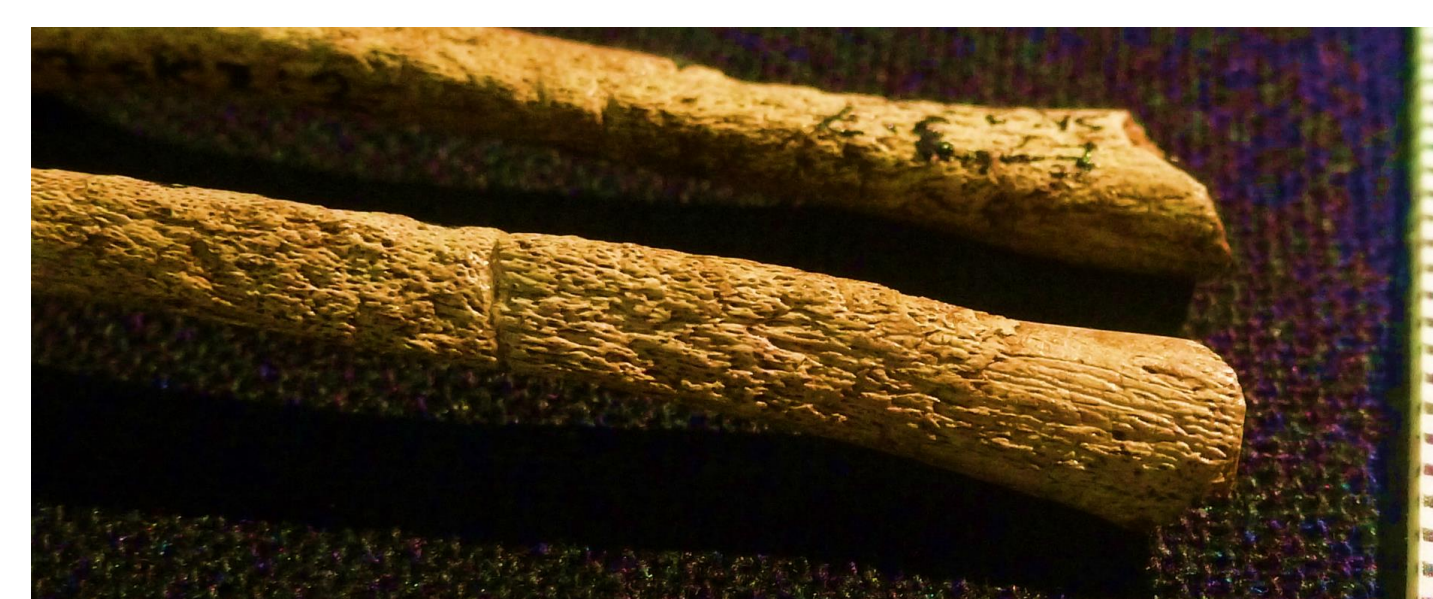
- Proximal metaphyses and epiphyseal surface of femora
- Wide femora metaphyses
- Rhizomelia of the femora
- Pars basilaris of the occipital
- Body of the sphenoid
- Left ilium



Femoral Comparison

### Abnormal Periosteal Reaction

- Porous, slightly striated, and active bone addition
  - primarily on anterior surface of long bones
- Bone porosity



Distal Ulnae of EZ 3-7-2



EZ 3-7-2



Proximal Femoral Metaphyses of EZ 3-7-2



Right Femur of EZ 3-7-2

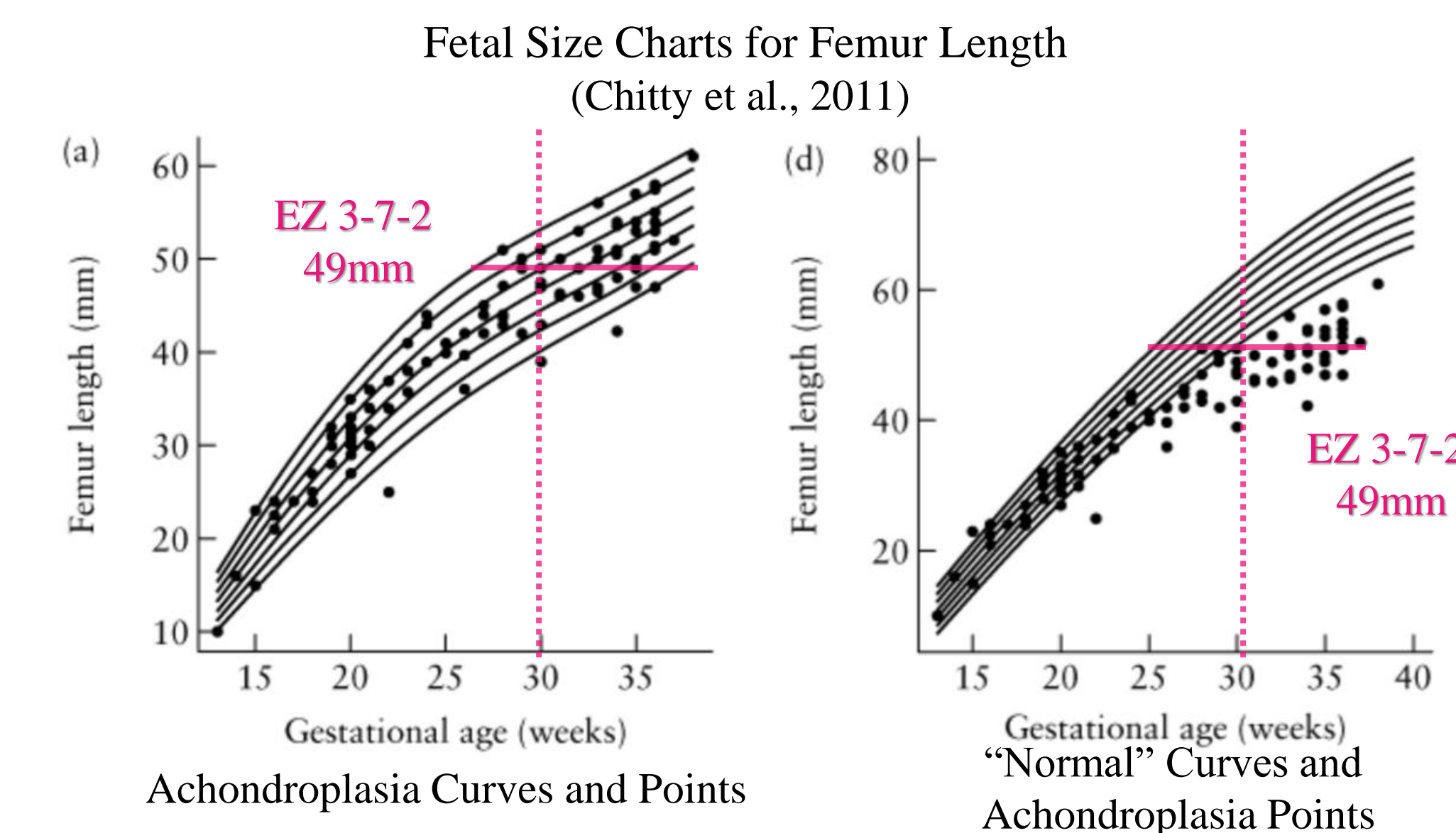


Left Ilium of EZ 3-7-2

## DISCUSSION

### Inheritance of Skeletal Dysplasia

- EZ 3-7-1 would have had the heterozygous form of achondroplasia and the heterozygous form of Langer mesomelic dysplasia (LMD) resulting in Leri-Weill Dyschondrosteosis (LWD).
- If it is assumed that the biological father of EZ 3-7-2 was not achondroplastic, EZ 3-7-2 had a 50% chance of having achondroplasia. If the biological father had achondroplasia, there was a 50% chance EZ 3-7-2 would be heterozygous for achondroplasia and 25% chance for homozygous dominant (and lethal) achondroplasia.
- If the biological father did not have LWD, EZ 3-7-2 had a 50% of having LWD. If the biological father did have LWD, EZ 3-7-2 had a 50% chance of having LWD and a 25% chance of having LMD.
- In comparing the growth curves for achondroplasia and "normal" fetal femur lengths, it becomes more likely that EZ 3-7-2 had an abnormally short femur (rhizomelia), perhaps due to achondroplasia. There is less (or no) visible manifestation (mesomelia) of LWD.



### Prenatal Infection

- Most frequent bacterial intra-uterine infections: *Listeria monocytogenes*, *Treponema pallidum*, *Mycobacterium tuberculosis* and *Campylobacter fetus*. Only *Treponema pallidum* produces osteological indications, but not involving the periosteum in fetuses (Shiple et al., 1921; Pálfi et al., 1992).

### Indication of Maternal Health

- Severe nutritional deficiency of a pregnant woman will directly affect the fetus, influencing maternal and fetal morbidity and mortality (Kinaston et al., 2009).
  - Iron, Vitamin C, and/or Vitamin D deficiency present with porosity and periosteal addition (Brickley and Ives, 2006)
- Maternal estrogenic levels during pregnancy can influence fetal bone tissue development (Magliaccio et al., 1996).
- Maternal obesity (Wu et al., 2012) and prenatal maternal stress (Beydoun and Saftlas, 2008) have negative impacts on fetal growth and development.

## CONCLUSIONS

- EZ 3-7-2 inherited the skeletal dysplasia of achondroplasia and possibly LWD from EZ 3-7-1.
- EZ 3-7-1 did not transplacentally transmit her infection to EZ 3-7-2.
- The abnormal periosteal reaction of EZ 3-7-2 was most likely an indication of poor maternal health, including possible nutritional deficiency, hormonal imbalance, and/or obesity.

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