

# **Supplementary Appendix**

## Supplementary Appendix Figure 1:

### Skeletal Surveys Showing Changes Between 3 and 4 Years-Of-Age:

**Top Panel:** At age 3 years, there is diploic thickening of the skull. Generalized osteosclerosis includes the carpal and tarsal bones, and especially the epiphyses, and there is cortical thickening. Undertubulation of major long bones accompanies minimal sparing of the sclerosis in the metaphyses.



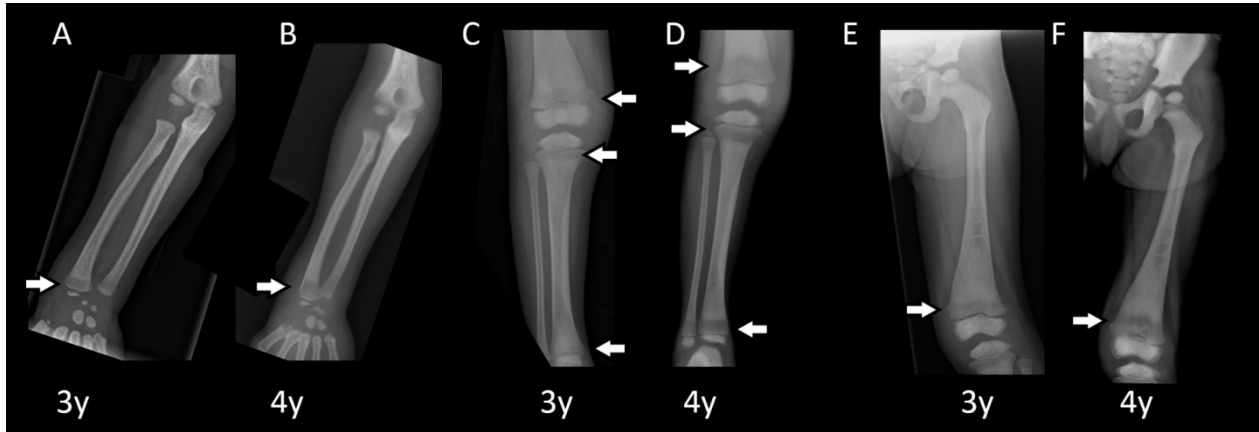
**Bottom Panel:** At age 4 years, generalized osteosclerosis and undertubulation persist, but the metaphyses are no longer osteosclerotic.



## Supplementary Appendix Figure 2:

### Changes Within Metaphyses

At age 3 years (a,c,e), generalized osteosclerosis includes epiphyses but there is some minimal distal metaphyseal sparing (arrows). At age 4 years (b,d,f, respectively), the metaphyseal osteosclerosis has resolved, although undertubulation (widening) of the long bones persists.



At age 3 years (left panel), there is: i) generalized osteosclerosis including especially the femoral epiphyses, with mild sparing of the distal metaphysis; ii) cortical bone thickening; and iii) undertubulation together consistent with osteopetrosis.

At age 3 years 10 months (right panel), metaphyseal lucency has extended and increased, but diaphyseal and epiphyseal osteosclerosis and undertubulation persist. Thus, metaphyseal osteosclerosis has corrected in the new endochondral bone.

