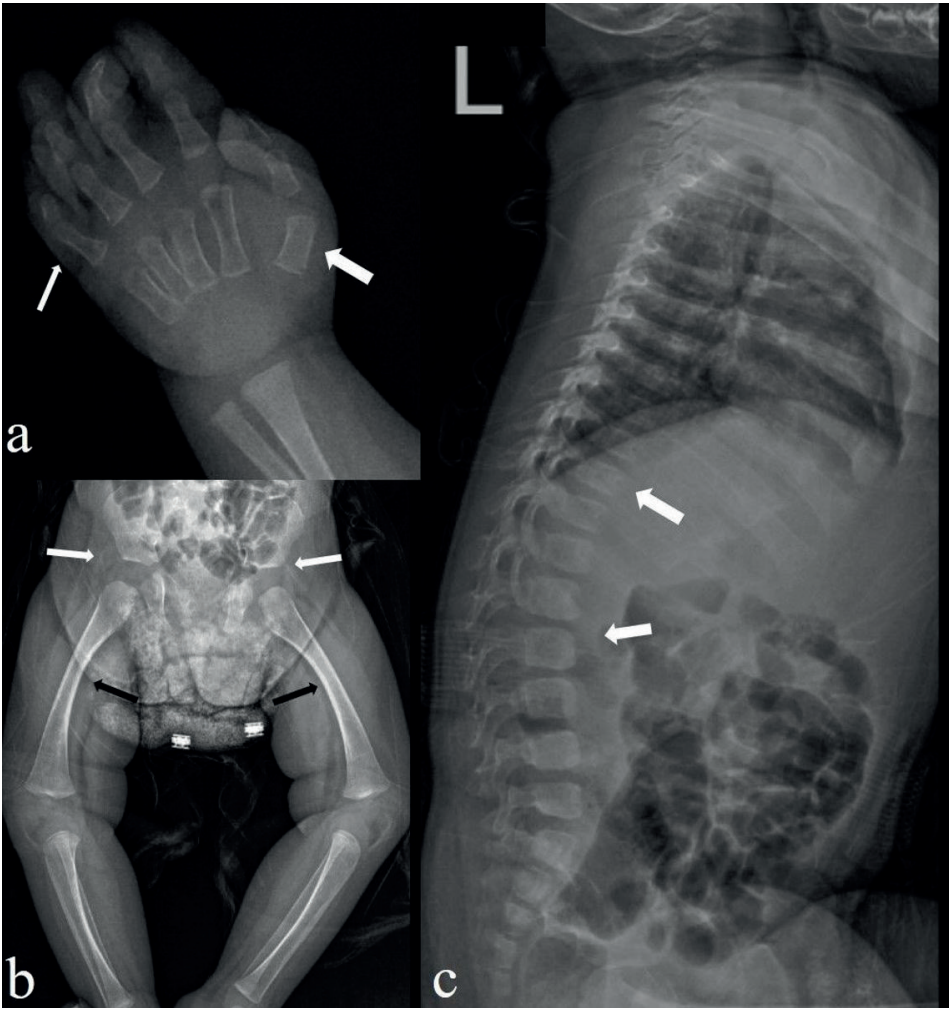


Supplementary Table S1. Results of metabolic investigations.

Tests	8th month	References
Organic acids screen, random urine	Non spesific	
Amino acids, random urine	Non spesific	
Amino acids, plasma	Non spesific	
Carnitine and acylcarnitines, dried blood spot	Non spesific	
Biotinidase activity	5.14 nmol/mL/min	4.2 – 12.8 nmol/mL/min
Lactate	1.61 mmol/L	0.5 – 2.2 mmol/L
Ammonia	56.9 µmol/L	16 – 60 µmol/L



Supplementary Fig. S1. **a.** An anteroposterior hand radiograph reveals clinodactyly (thin white arrow) and a short, thick first metacarpal bone (thick white arrow). **b.** Pelvic radiograph showing the pelvis is elevated and small, with bilateral horizontal and shallow acetabula (white arrows). Delayed epiphyseal ossification is observed in the femoral epiphyses, and bowing of the femurs is present on both sides (black arrows). **c.** A lateral thoracolumbar radiograph reveals mild thoracolumbar kyphosis (white arrows). Posterior scalloping of the lumbar vertebrae is observed, along with mild spondyloepiphyseal dysplasia.