

Rett Syndrome and Osteoporosis

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Definitions

Osteopenia: Refers to bone DXA value between 0.0 and -1.5 standard deviation below the mean.

Osteoporosis: Refers to bone DXA value less than -2.5 standard deviation below the mean.

Any prior “fragility” fracture = osteoporosis, irrespective of BMD

Osteoporosis Modern Definition: A systemic skeletal disease characterized by low bone mass and micro architectural deterioration of bone tissue with a consequent increase in bone fragility and susceptibility to fracture.

BONE QUALITY

21

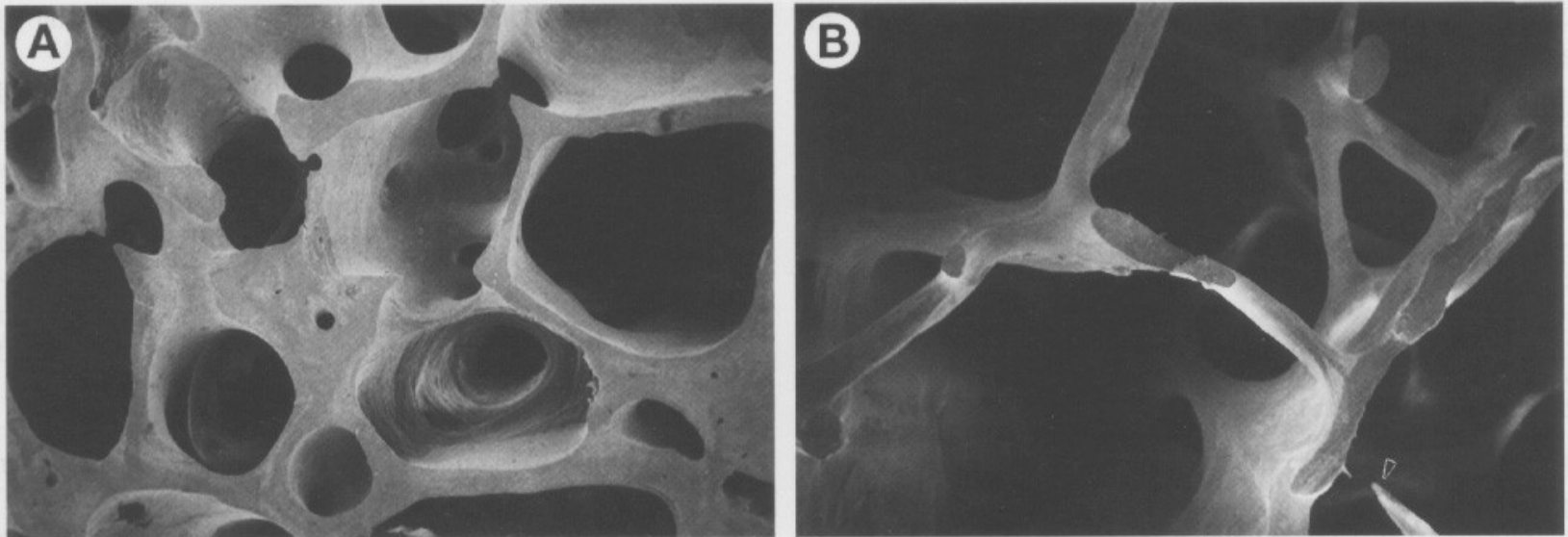
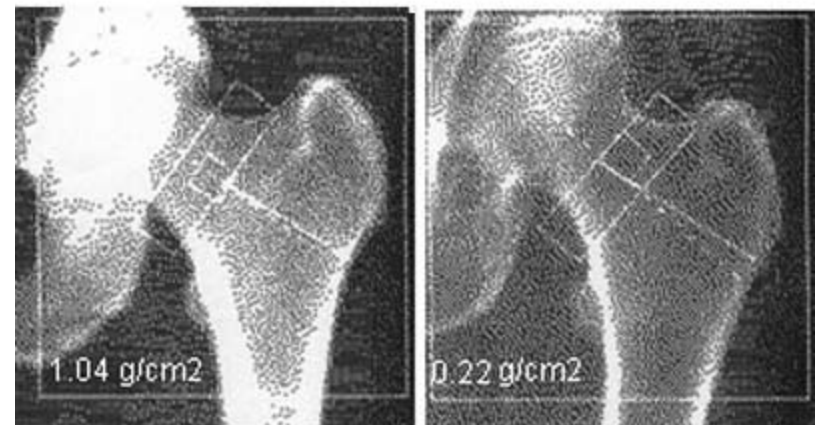
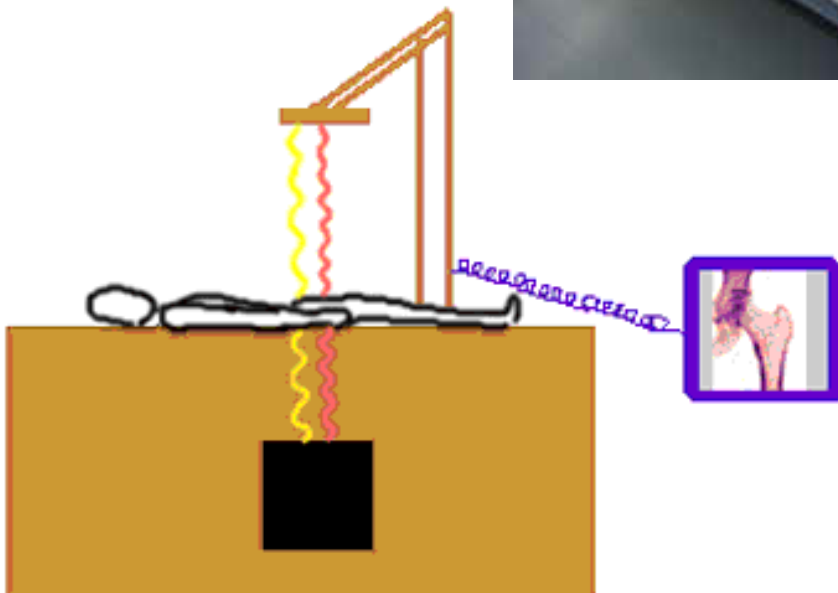


FIG. 1. Scanning electron micrographs of (A) normal and (B) osteoporotic cancellous bone from human iliac crest. Reproduced from J Bone Miner Res 1986;1:15–21 with permission of the American Society for Bone and Mineral Research.⁽²⁰⁾

X-rays: Decreased Bone Mass



Measuring Bone Mass: DXA



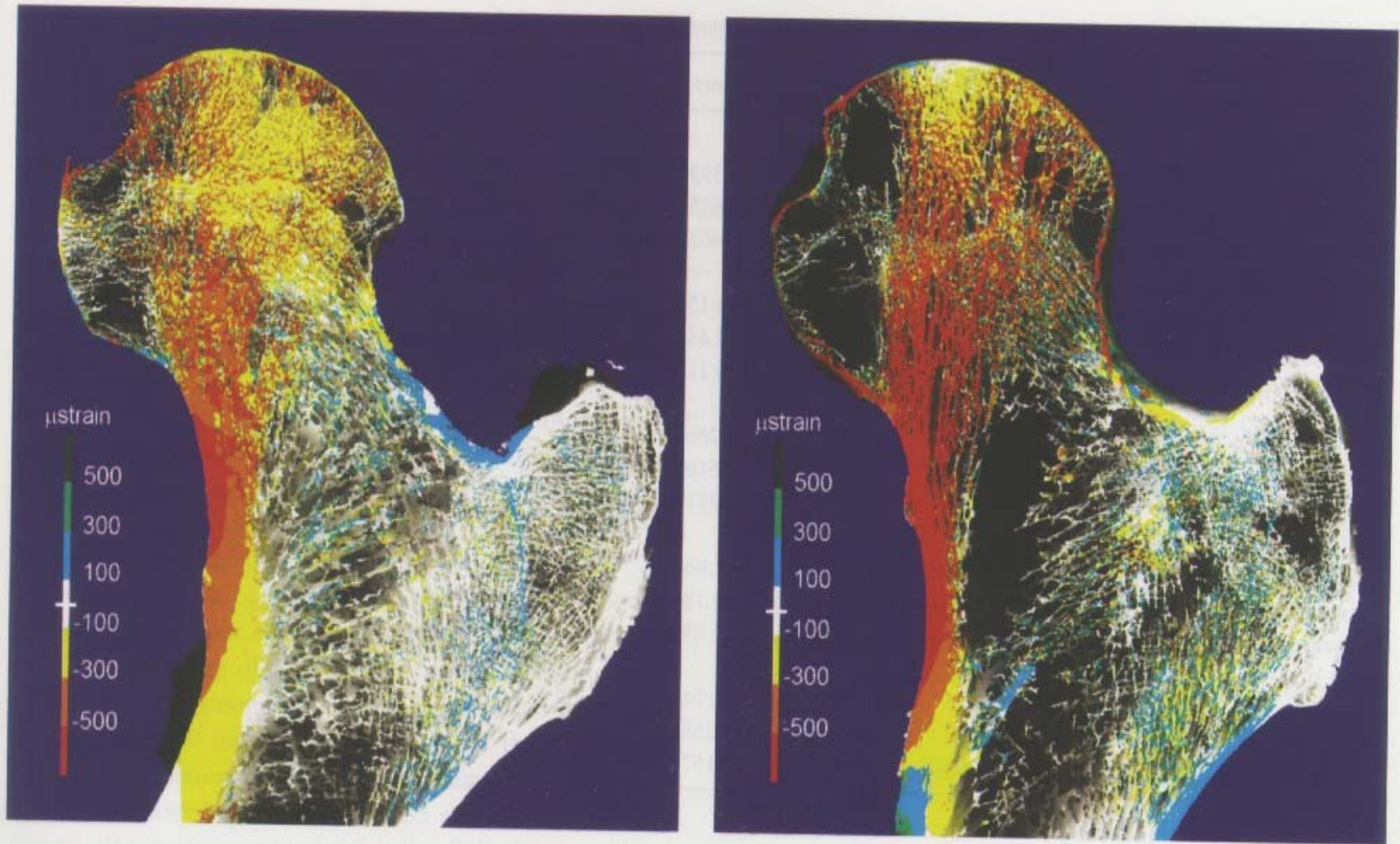


FIG. 3. Calculated distribution of the principal strain component with the largest magnitude in the (A) healthy and the (B) osteoporotic femurs for a physiological joint force representing the stance-phase of walking. Yellow-to-red represent increasing compressive strains; blue-to-green, increasing tensile strains.

DXA SCAN in Rett Syndrome

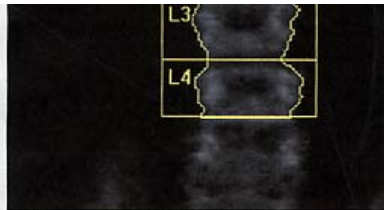


Image not for diagnostic use
k = 1.144, d0 = 49.5
52 x 88

DXA Results Summary:

Region	Area (cm ²)	BMC (g)	BMD (g/cm ³)	T - Score	PR (%)	Z - Score	AM (%)
L1	7.07	2.50	0.353	-7.1	38	-2.4	67
L2	6.68	2.81	0.421	-7.6	41	-2.7	71
L3	7.07	2.76	0.390	-8.8	37	-3.7	62
L4	6.55	2.82	0.430	-8.0	41	-2.7	69
Total	27.38	10.89	0.398	-8.7	39	-3.2	67

Total BMD CV 1.0%, ACF = 1.040, BCF = 1.016, TH = 4.979

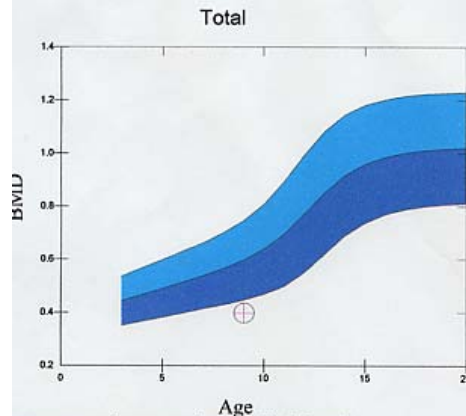
WHO Classification: Osteoporosis

Fracture Risk: High

**FOR RESEARCH PURPOSES ONLY
NOT TO BE USED FOR CLINICAL CARE**

Physician's Comment:

[Handwritten signature]



reference curve and scores matched to Pediatric Female

cc: 2004 ASBMR Abstract #SU112

HOLOGI

DXA: Lateral distal femur site: sensitivity

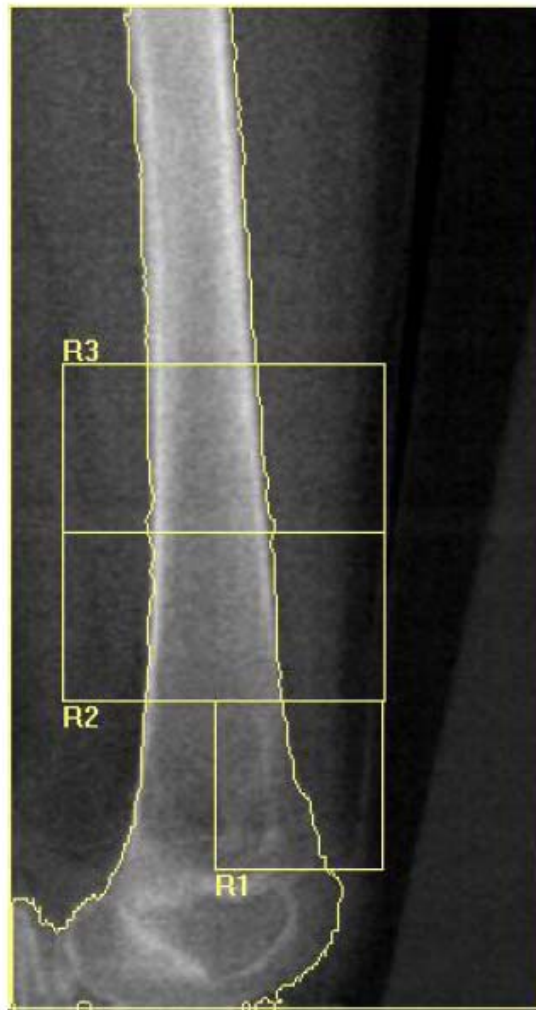
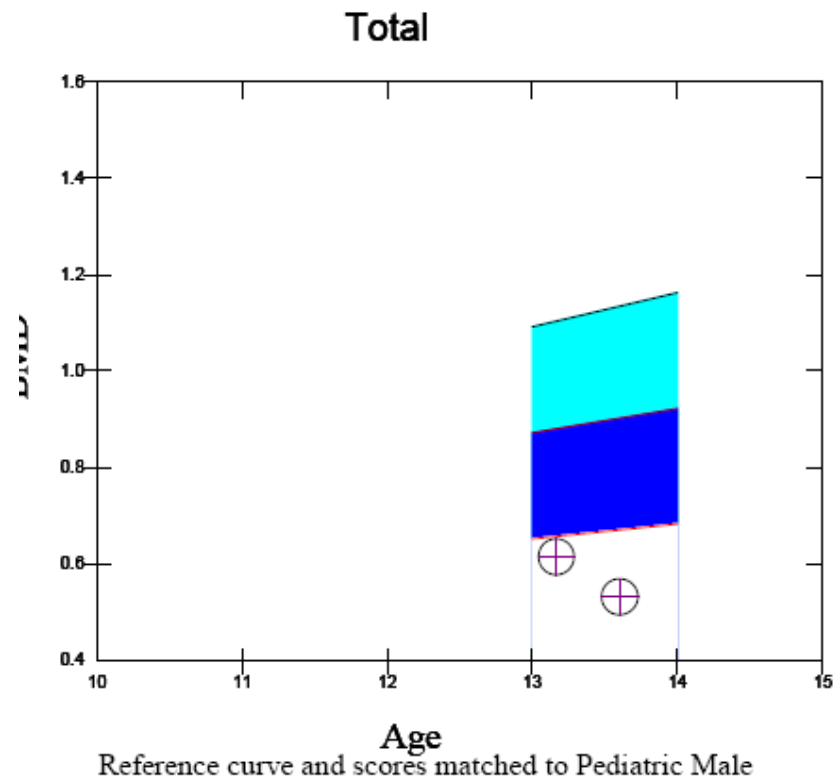
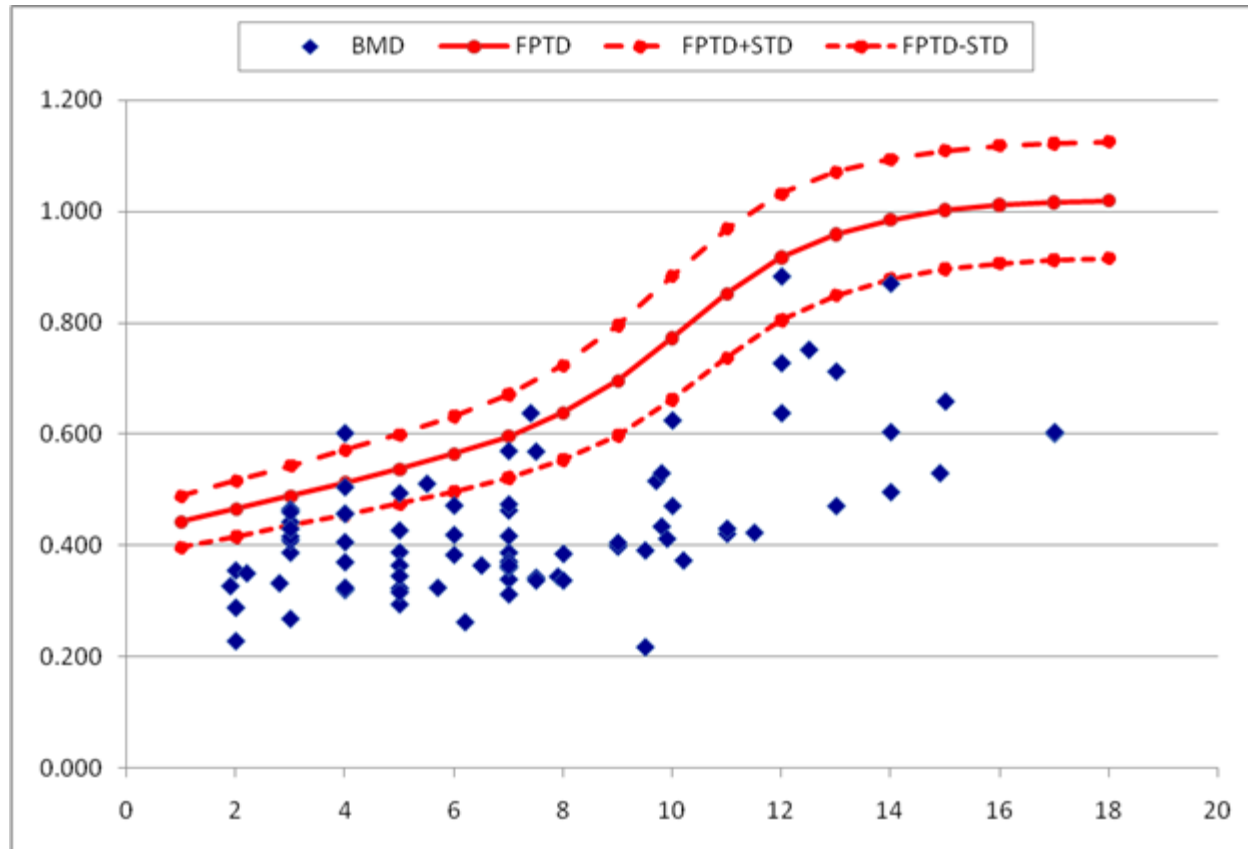


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252 x 220



DXA Bone Mineral Density Measurements



Calcium and Vitamin D Supplements

- TUMS: Calcium carbonate 500
- OSCAL: Calcium Carbonate
- CALTRATE: Calcium carbonate 600 mg
- CITRACAL: Calcium citrate 500-600
- D added: 400-500 international units
- THESE ARE ABSORBED TO THE SAME EXTENT

This policy has been revised by the [policy posted on February 1, 2006.](#)

POLICY STATEMENT

PEDIATRICS Vol. 104 No. 5 November 1999, pp. 1152-1157

AMERICAN ACADEMY OF PEDIATRICS:

Calcium Requirements of Infants, Children, and Adolescents

Committee on Nutrition

Few data are available about the calcium requirements of children before puberty. Calcium retention is relatively low in toddlers and slowly increases as puberty approaches. Most available data indicate that calcium intake levels of about 800 mg/d are associated with adequate bone mineral accumulation in prepubertal children. The benefits of greater levels of intake in this age group have been studied inadequately.^{[20,32](#)} One study found a benefit of calcium supplements to children as young as 6 years of age.

The available data support recent recommendations for calcium intakes of 1200 to 1500 mg/d beginning during the preteen years and continuing throughout adolescence as recommended by the National Institutes of Health Consensus Conference^{[2](#)} and the National Academy of Sciences.

Vitamin D Supplements

- Measure Vitamin D in blood tests: value should be over 32 ng per ml
- Vitamin D helps calcium absorption from the stomach
- Vitamin D helps form bone mineral and so helps strengthen bone:
- Adults vitamin deficient get “osteomalacia” and high parathyroid levels
- Vitamin D reduces hip fractures

Vitamin D Supplements

Liquid Drisdol or ergocalciferol: 8000 IU/cc

Capsules: 50,000 IU weekly to restock stores

Tablets: 400-1000 IU daily

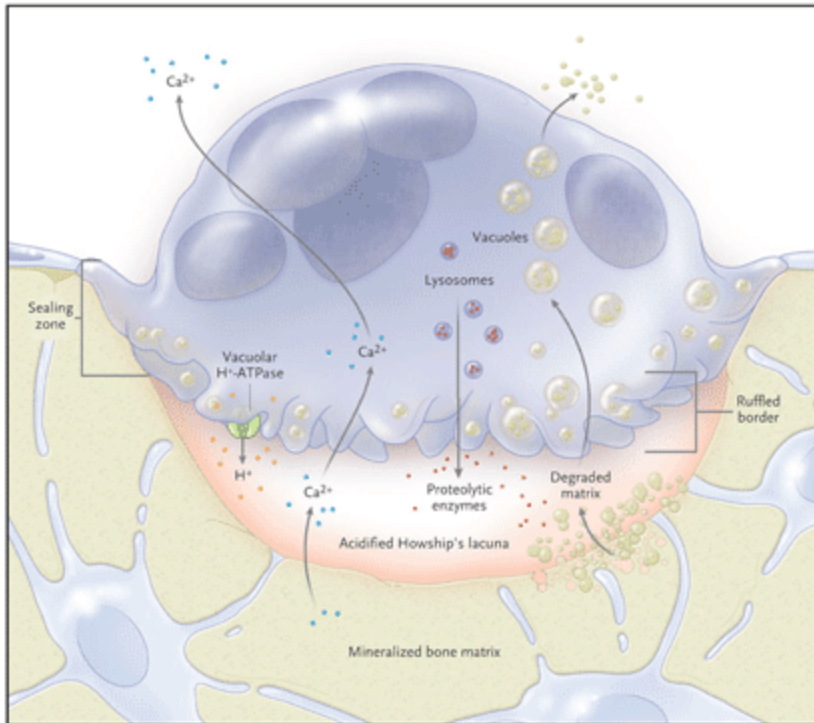
Recommended Vitamin D Daily Intake*

Age RDA (Age based)		Weight Recommended	
0-50 yrs	200 IU/day	50 lbs/ 20 kg	600-800 IU/day
51-70 yrs	400 IU/day	90 lbs/ 40 kg	1100-1600 IU/day
70+ yrs	600 IU/day	110 lbs/ 50 kg	1200-2000 IU/day
		150 lbs/ 70 kg +	2000-2800 IU/day

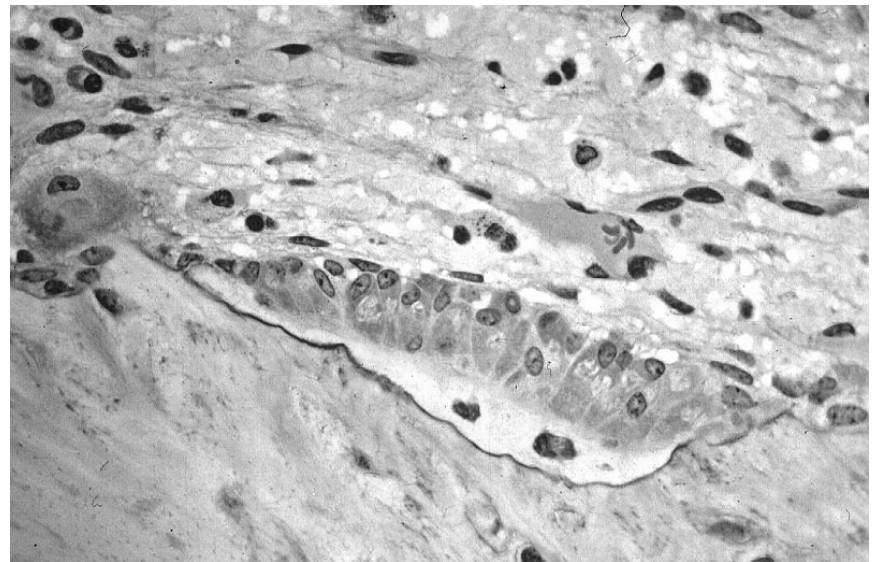
Monitoring of serum vitamin D levels is essential to assure correct dosage at each of these recommended levels (32-75 ng/ml).

*Shapiro JR, McMahon E , Hollis BW. Recommendations Regarding Vitamin D Intake in Osteogenesis Imperfecta. March 6, 2006.

Where is the bone problem in Rett ?

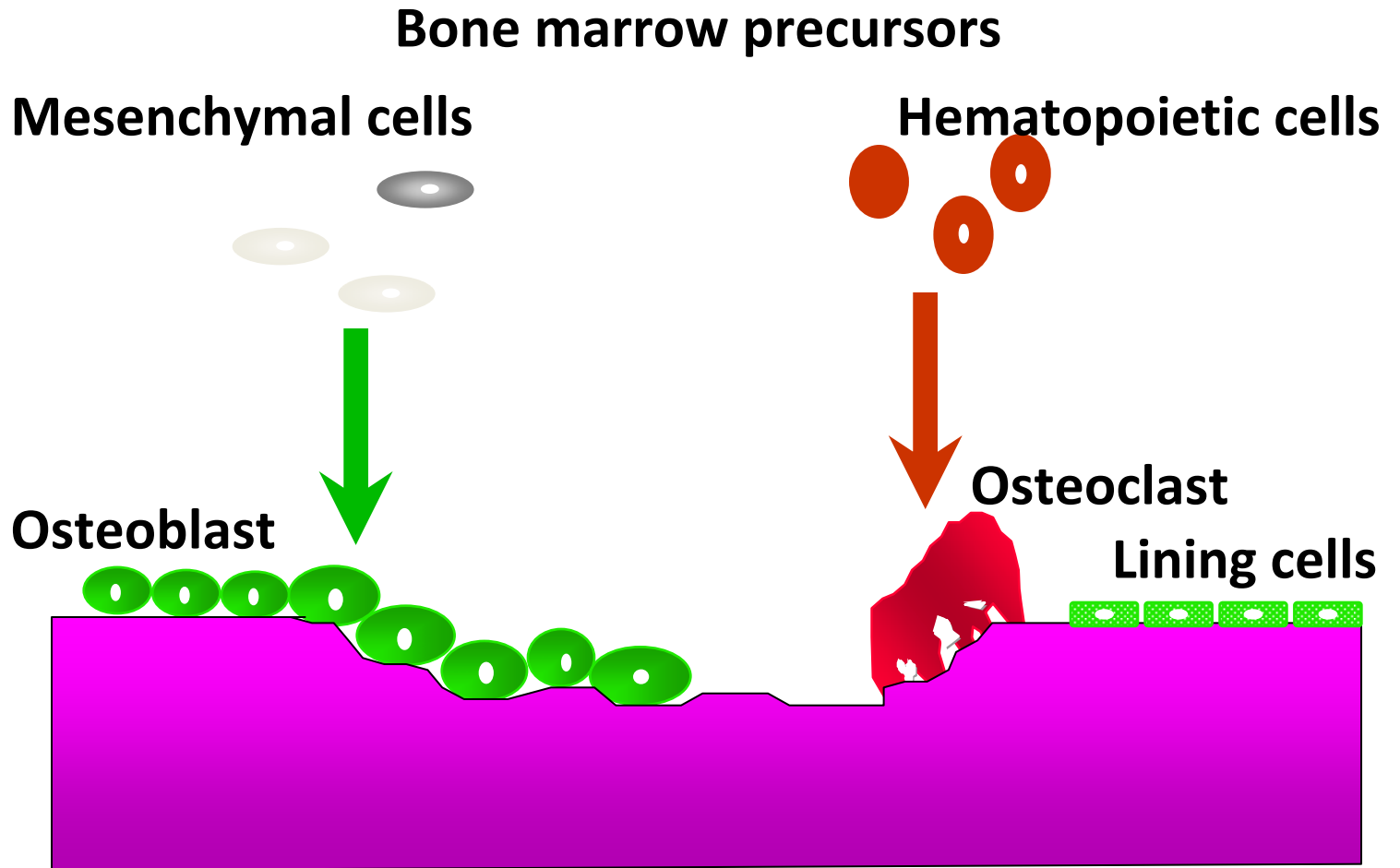


Osteoclast: Bone resorbing cell



Osteoblast : Bone Forming cell

Bone remodeling



Bone Specific Therapy

- Antiresorptive Agents: Bisphosphonates

- Aredia (Pamidronate) IV 3 months
- Boniva (Ibandronate) Oral and IV 3 months
- Reclast (Zoledronic acid) IV 6-12 months

- Bone Forming Agents

- Forteo SC daily for 18 months
- Calcium and Vitamin D