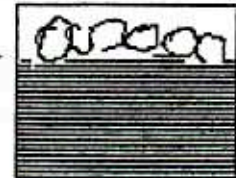


Bone Mass in GSD I, III & IX Patients
Presentation by Dr Cabrera -Abreu

A study by Great Ormond Street doctors in London reported concern about excess calcium appearing in the urine of GSD patients. The question asked is where is this excess calcium coming from?

Bones are composed of three parts:

- a) CELLS
- b) a MATRIX made of the protein collagen
- c) the MINERALS calcium and phosphate.

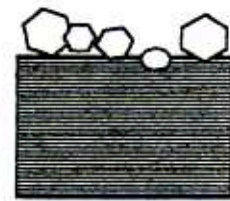


Normal Bone
in "FORMATION"

Bones are not static

but undergo a continuous process of:

bone destruction called "RESORPTION"
followed by
bone reconstrction called "FORMATION".

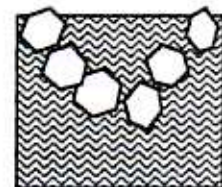


Thus in the "quiet" or steady state "normal" bone density occurs when

"destruction" = "formation"

If destruction is greater than reconstration you get **OSTEROPOROSIS** and thin brittle bones.

Thin Bone in
"RESORPTION"



Thus calcium loss in urine can be an indicator of thinning of the bones of GSD patients.

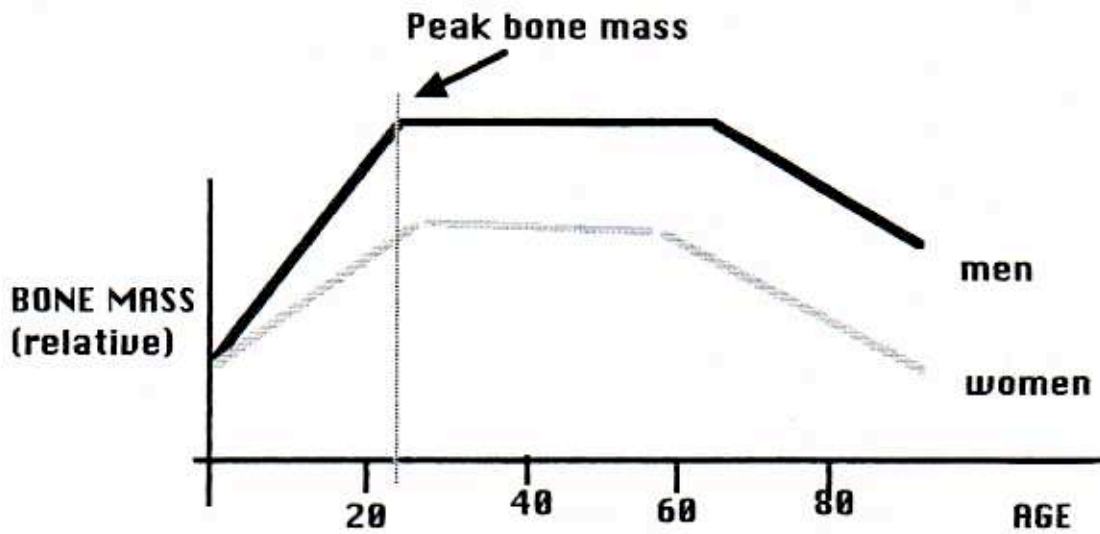
There are two ways to assess thinness of bone.

[Fig. 1] - A hip scan to determine bone mass/density in men and women vs. age.

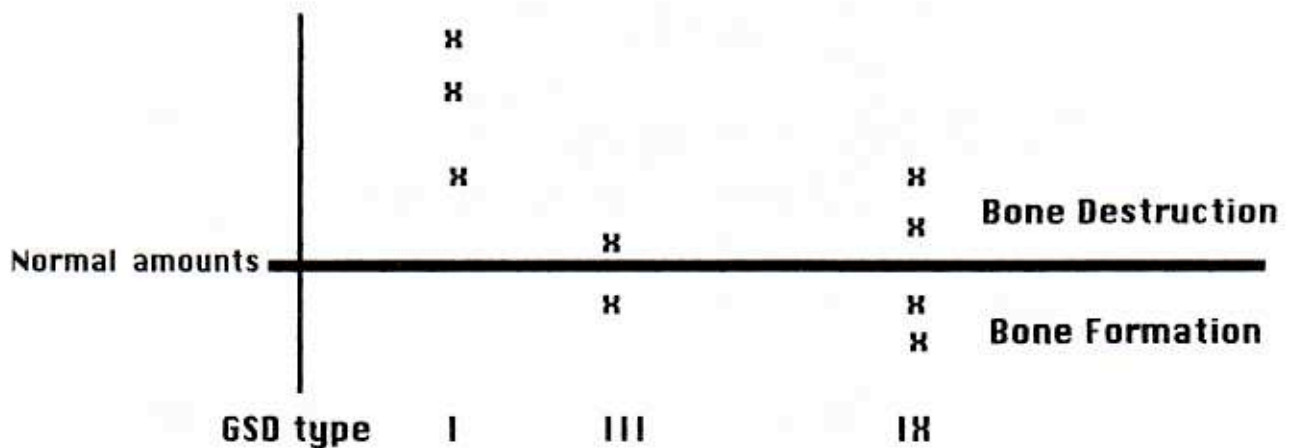
[Fig. 2] - Blood and urine tests.

Here the number of collagen fibres which break down into fragments in the blood can be measured and thus give an indication of bone destruction/formation.

[Fig 1] Hip Scan- Normal range



[Fig.2] Pyr/creatinine as a measure of the protein collagen fragments in urine of GSD patients.



How can we stop the bone thinning?

- n In women HRT (hormone replacement therapy) can be given.
 - n In older men biophosphates are given.
 - n In GSD patients the aim is to maintain and, if possible achieve a higher than normal peak bone mass (which usually occurs at the age of 25- see Fig.1) doses of Calcium and Vitamin D plus regular exercise in childhood and adolescence should all be considered. If the bones are very thin then Bisphosphonates could be given.
- As yet it is not known if the incidence of osteoporosis in some GSD patients is higher than normal or if it just has a more adverse effect