



Self-actualized Perceptions of X-Linked Hypophosphatemia Suggest a Pro-active role for Patient-Support Networks in Managing Patients with this Rare Metabolic Bone Disorder

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Abstract from 1994

X-Linked hypophosphatemic rickets: a disease often unknown to affected patients. Michael J. Econs, Gregory P. Samsa, Michael Manger, Marc K. Drezner, John R. Feussner. Duke University Medical Center, Durham, NC 27710, USA. *Bone and Mineral* 24: (1994) 17-24.

X-Linked hypophosphatemic rickets (XLH) is an X-linked dominant disorder that is secondary to renal phosphate wasting. Affected individuals frequently present the following characteristics: short stature, lower-extremity deformity, bone pain, dental abscesses, enthesopathy, rickets, and osteomalacia. Since the disorder is characterized by evident phenotypic abnormalities, we hypothesized that there would be a high degree of knowledge about the disease in affected kindreds. Thus, we constructed a six-page, self-administered questionnaire to determine whether family members are, in fact, aware of their disease and properly diagnosed and treated. We also designed the survey to determine rates of symptoms thought to be associated with rickets/osteomalacia in a population with a lower referral bias than is usually seen in tertiary care centers. We administered the questionnaire to 234 study subjects (57 affected) who were members of one of three large kindreds. Although 62% of affected individuals knew that they had some problem with their bones, only 22.6% were told by a physician that they had rickets or osteomalacia. This apparent lack of awareness occurred in spite of 61.1% of affected subjects complaining of bone or joint problems to their personal physician. Indeed, of those patients who had persistent complaints, only 34.5% were told they had rickets or osteomalacia. Only one patient was taking phosphate and vitamin D. The spectrum of symptoms evident in affected subjects compared with normals included: dental abscesses (54.5% vs 13.0%, P<0.001), bone pain (45.5% vs 28.2%, P=0.027), back pain (51.8% vs 35.1%, P=0.036), joint stiffness (48.2% vs 16.8%, p<0.001), joint pain (55.4% vs 31.1%, P=0.003), weakness (25.0% vs 10.7%, P=0.023), and hearing loss (28.6% vs 9.8%, P=0.002). Surprisingly, although affected individuals complained of many symptoms due to XLH, they fractured bones less frequently than controls (20% vs 38.1%, P=0.018). Our data demonstrate that, despite the presence of disease in family members, few affected subjects knew that they had XLH. Although the presence of symptoms did increase knowledge of disease status, only one-third of symptomatic individuals knew of their diagnosis.

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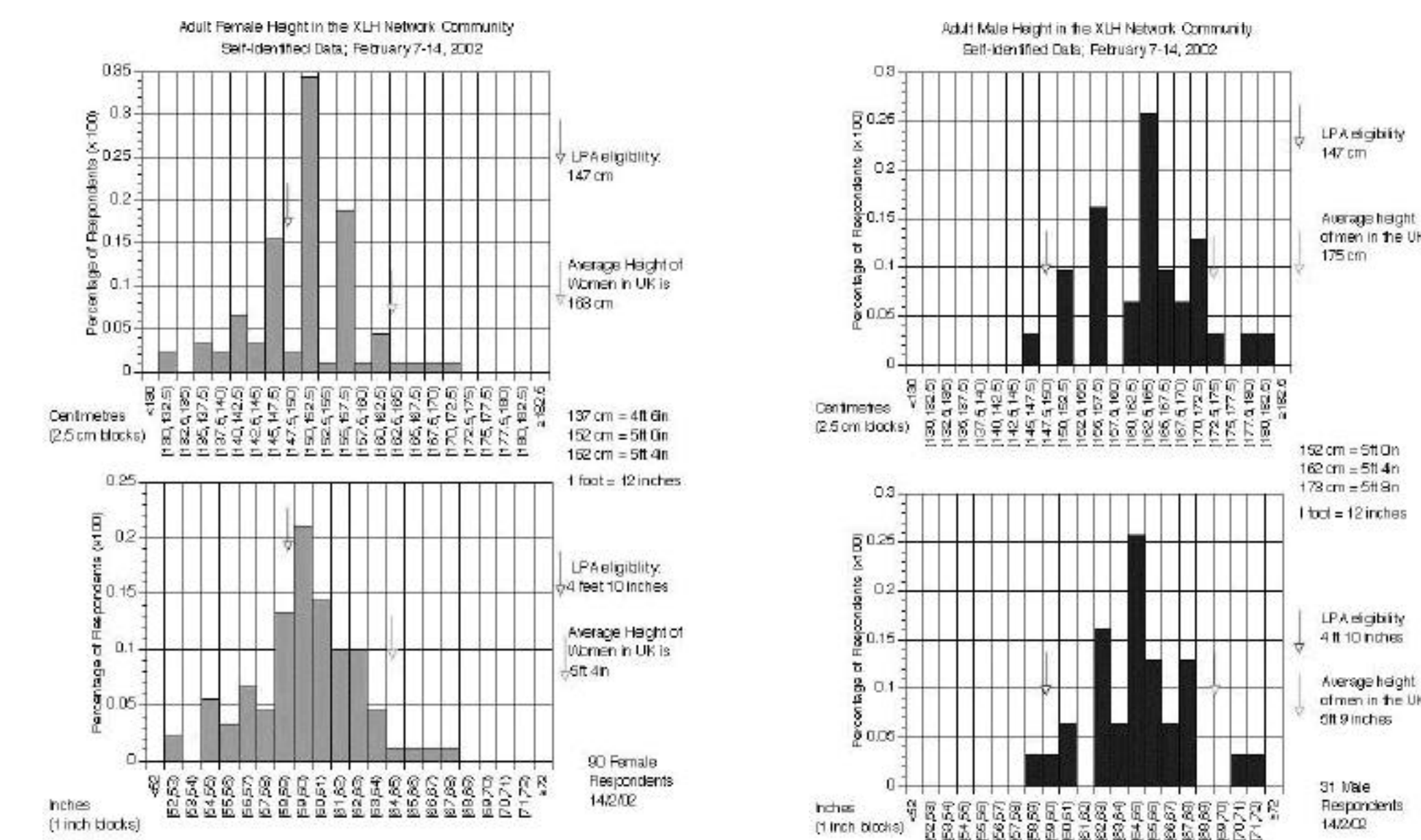
A survey of families with XLH(1) indicated that 1/3 have little comprehension of the ramifications of their disorder. An internet-based patient-support network has since developed, which seeks with timely information to enfranchise and empower people affected by XLH. Enquirers join if a family member is professionally diagnosed with XLH. We asked whether this network (recognizing the potential for bias towards severely affected cases) could help to characterize a population of adults with XLH that is beyond the assessment capacity of major metropolitan medical centers. In a rapid email poll, 121 responses of self-identified adults with XLH were elicited and grouped according to age, gender and height. By contrast, other studies assessed 57(1), 22(2), or 16(3) XLH adults. Average (modal, normal distribution, 30 respondents) male height in the XLH Network poll was 5'5" (UK national average 5'9") while average female height (91 respondents) was 5'0" (UK national average 5'4"). Of the females, 1/3 were at or below 4'8". We observed that profoundly short stature was not reported by respondents aged 18-25 years, though the number in this cohort was low (10 female, 5 male). This poll may reinforce the perception of members that short stature is a manifest consequence of XLH, while subtly emphasizing, to parents of newly diagnosed children, the potential for improved height provided by optimal therapeutic compliance (calcitriol and phosphate) since the 80s. A separate poll revealed that 26 of 49 adult respondents were not taking medication for XLH; clinical observations (2,3) that osteomalacia with bone pain may be alleviated by a return to these medications, are regularly discussed in the XLH Network. Continuing anecdotal presentation of problems associated with XLH in adulthood: spinal stenosis; Meniere's disease; enthesopathies; knee and joint pain; provides clear evidence that XLH is not only a childhood disorder, but can have important ramifications throughout adulthood, which may require a pro-active approach with clinicians.

- Econs M et al (1994) *Bone Miner.* 24:17-24.
- Reid IR et al (1989) *Medicine* 68: 336-352.
- Sullivan W et al (1992) *J. Clin. Endocrinol. Metab.* 75:879-885.

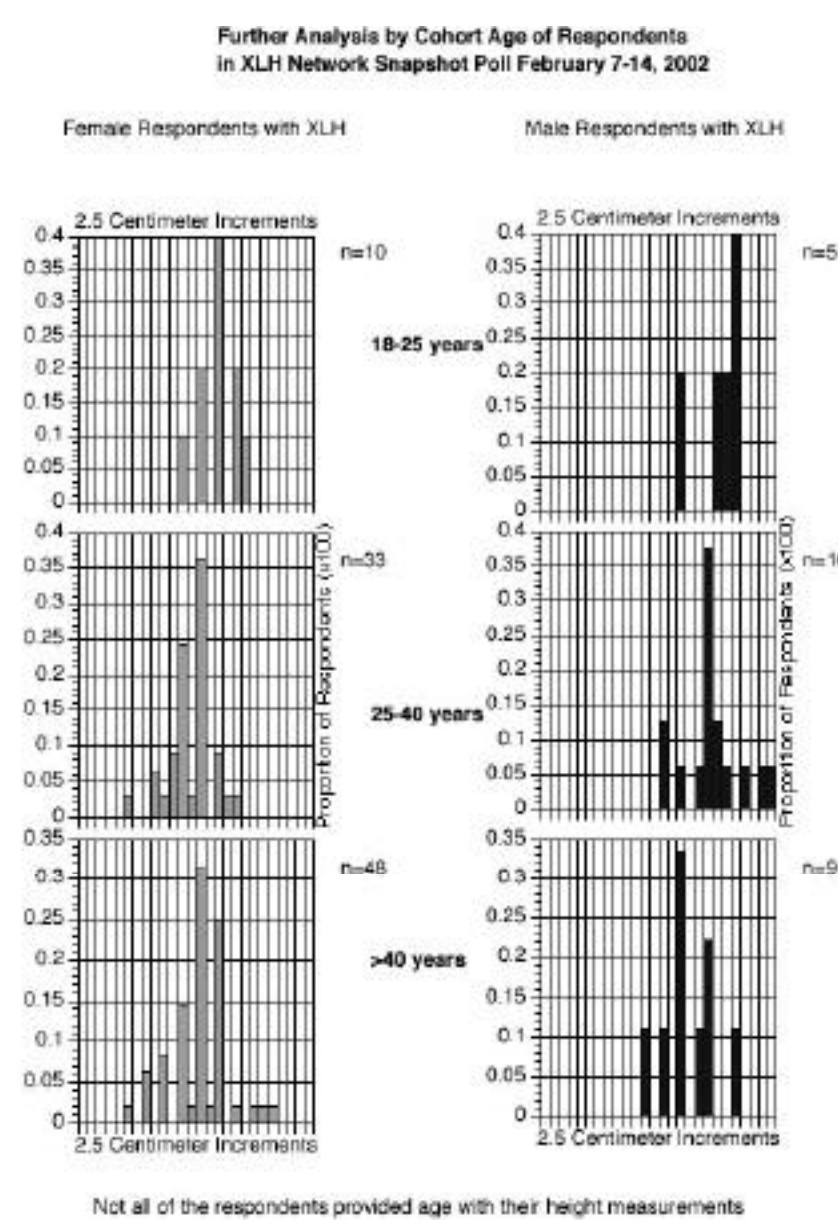
Results

With regard to the question of height, 121 responses

were elicited from members reporting for XLH+ adults (>18), of which 31 were male and 90 were female. The distribution of height, in both cm and inches, is recorded in the histograms below, which are also differentiated into age cohorts in the third figure. Members of the XLH Network are in the region of 4 to 6 inches shorter than 'average.' This rapid poll also revealed that respondents aged between 18 and 25 (the period since the early 1980s during which contemporary medication (calcitriol and phosphate supplements) has been administered, do not exhibit the profoundly short stature of their elders who had not been managed in this way. A subsequent poll (shown in the table) revealed that approximately half of adult respondents were not taking any medication for XLH, while 100% of respondents for juveniles with XLH indicated that they were under careful medical/medication management.



No profoundly short respondents 18-25



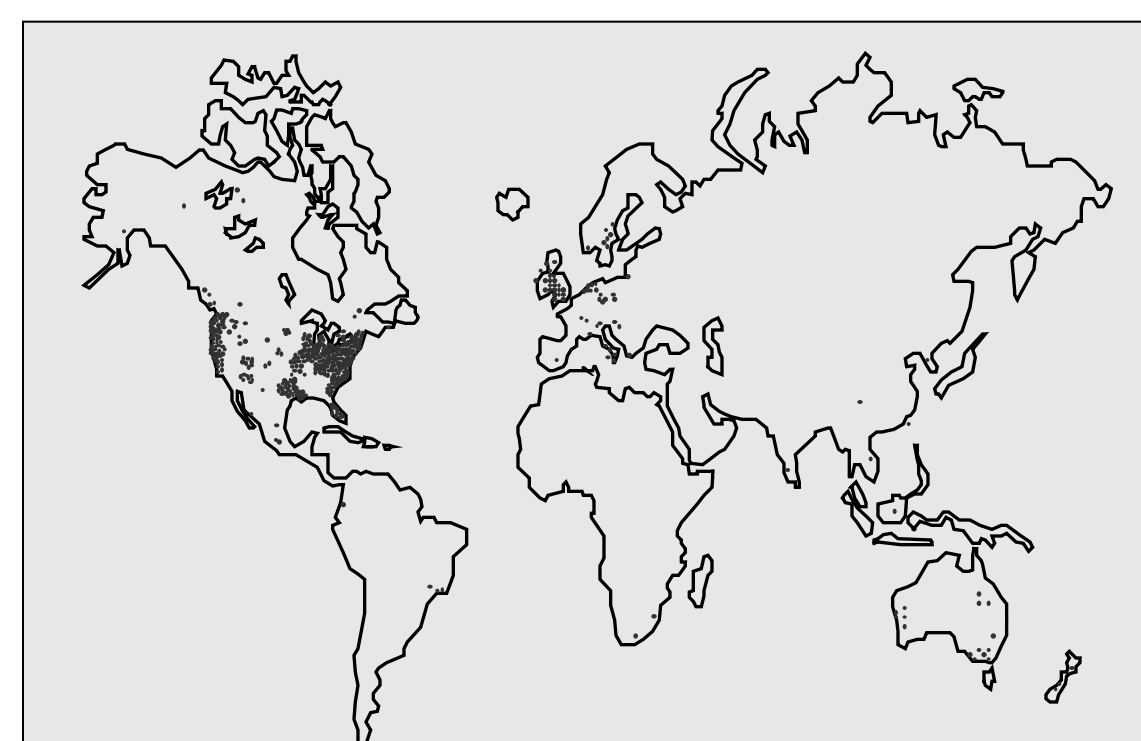
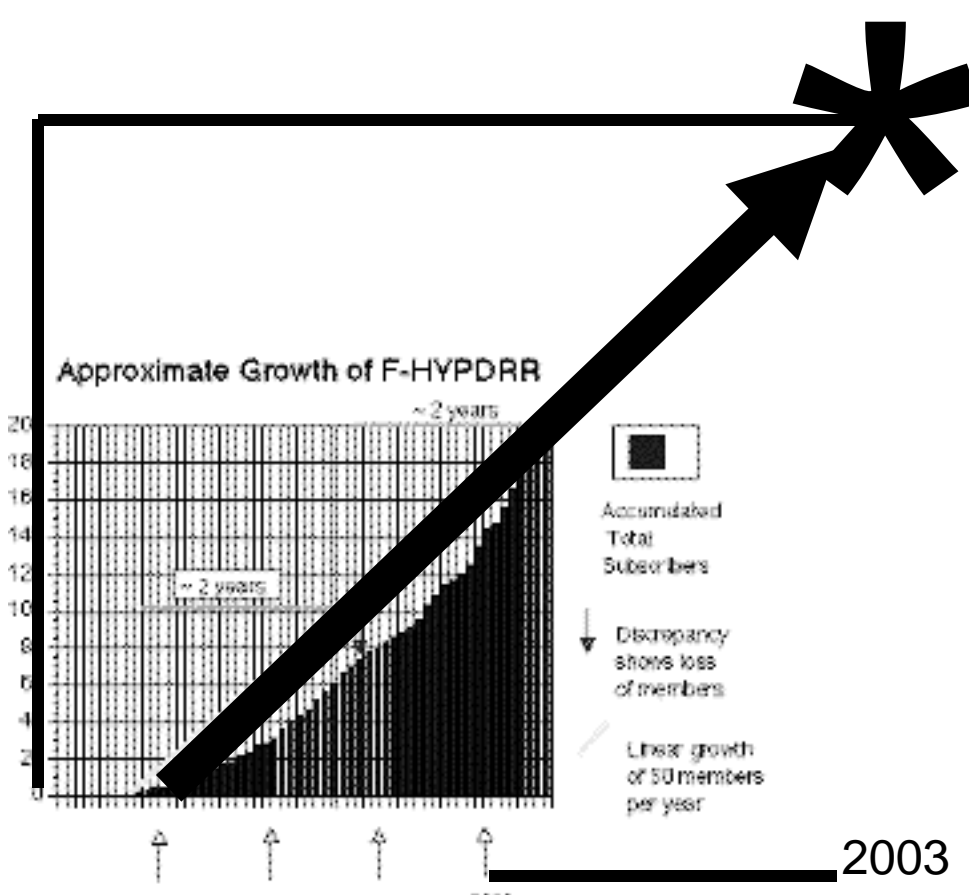
Introduction

In the summer of 1996, a website was created at

Newcastle upon Tyne, which sought to describe the current state of understanding of the disorder popularly known as 'Vitamin D Resistant Rickets.' This website came to the attention of a parent in British Columbia searching for information, and with email links between families in South Africa, Canada and the UK, served by a listserv at St. John's University, New York, the XLH Network was born. The network has grown steadily since that inception, recruiting professional members from the academic, biotechnology and clinical research and therapeutic management spheres, in addition to providing information, empathy and support for a small host of enquirers who have been diagnosed with X-Linked Hypophosphatemia (XLH), Autosomal Dominant Hypophosphatemic Rickets (ADHR), Oncogenic Hypophosphatemic Osteomalacia (OHO), or rare giant nevus rickets syndrome.

Growth of the XLH Network

355 members 1st September 2003
A remarkably consistent rate of 50 new members year on year for 7 years



Geographical Distribution of XLH Network

Methods

We wanted to determine whether the XLH Network, as a layperson support network (monitored by professional researchers and clinicians), could begin to characterize its membership, in effect describing the disorder as it is experienced by the network members. We reasoned that quick, simple polls would stand the best chance of eliciting responses, and so our first question asked, of respondents over the age of 18: 'How tall are you?' Similarly, a second question asked, of adults with XLH: 'Are you currently taking any medication for XLH?' These polls were distributed through the email listserv system, simultaneously to all members, and responses were collected within a two week period thereafter, and collated for presentation back to the network.

Medical Management of XLH in Adults

2002: Adult (>18) Reporting Members with XLH				
	Female	Male	Total Respondents	% of Total
Taking Medication	13	10	23	46
Not Taking Medication	18	8	26	54
Totals	31	18	49	100

Discussion

It is clear that the sort of rapid poll

'snapshot' of members in the XLH Network described here can elicit simple descriptive data which exemplifies some of the prevailing symptoms of XLH to those affected by this metabolic bone disorder. In addition to simple height data, of course, a constant stream of symptom descriptions is distributed among the network, with members carefully seeking to appreciate the nature of this condition as they experience it.

Obviously, these polls and anecdotal analyses are subject to a variety of caveats, not least of which is that specific diagnosis cannot easily be confirmed in an online community to which members are admitted on the basis of their diagnosis as they [claim to] have received from professional consultation.

But these sorts of polls and worldwide public analyses that are shared among hundreds of people affected by XLH can certainly be said to be delivering an innovative response to the identification, a decade ago, of lack of comprehension of this disorder among those who are affected by it.

Conclusions

- ❖ People affected by XLH can be engaged in direct consideration of their condition, its manifestations and sequelae, when empathetic coordination, in an online setting, empowers them to do so.
- ❖ People affected by XLH as empowered by their enhanced understanding of their condition acquired through the XLH Network, may seek to interact pro-actively with their physicians to ensure the best possible management and clinical practice in world terms relative to contemporary understanding of XLH treatment. This empowerment and pro-activity may have important consequences in terms of any individual physician-patient relationship. Best practice should be shared equivalently around the world when resources allow.

References

- Econs M et al (1994) *Bone Miner.* 24:17-24.
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