

Reviewer's report

Title: Osteogenesis Imperfecta: a nomenclature (re-named to Syndromes with congenital brittle bones)

Version: 3 **Date:** 2 August 2004

Reviewer: Jay Shapiro

Reviewer's report:

General The focus has been appropriately changed.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

page 3 line 3: insert it.. "and it is therefore"

page 3 line 11: classification is no longer tenable

page 3 line 12: rather than "oversimplification" please consider "failed to encompass the obvious clinical variability apparent in this disorder"

page 3 line 17: after classification insert "initially proposed by Silience et al in 19-"

page 3 line 18: insert "types" for forms

page 3 line 19: amd have been "added as numbers V to VII", while others have been designated

page 3 last line: insert others designated with clinical features,

page 4: line 3: characteristics of each type "overlap"

page 4: line 3: add Furthermore, there is no

page 4 line 8: remove the sentence "There are many overlaps....")

page 4 line 18: add the LRP 5 mutation with "causal mutation"

page 4 line 19: page 4 line 20: Following this "example, I propose to define osteogenesis imeprfecta as syndromesw resulting from mutations...."

page 4: last line (SROI.. "pending the identification of their causal mutation."

page 5: line 13: only a few type I OI subjects have "normal stature"

page 5: line 12: add secondary to the effect of a null allele mutation

page 5: line 17: type I patients have obvious triangular facies (mentioned under type III later)

page 6: line4: insert "with age" for appropriately.

page 6: line 9: place the reference to the mutation higher in this section.

page 7: top: the pulmonary disorder in type III should be mentioned here.

page 7: paragraph 2 line 6: severe cases can be dismembered during delivery

page 7 paragraph 2 line 8: severity, "a few " patients...

page 9: line 4 add due to dislocation of the radial head

page 10: line 1: why include the osteoporosis pseudoglioma syndrome ? It really doesn't resemble OI except it does habe brittle bones, this should be more specifically differentiated from OI but included in a discussion of brittle bones.

Summary: add "a definition for OI is proposed based on the presence of type I collagen mutations"

Inclusion of related syndromes lacking mutations should be considered as SROI until further defined....

Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

Statistical review: No

Declaration of competing interests:

None