Towards more logical classification of osteogenesis imperfecta

In order to create a logical classification system for osteogenesis imperfecta we must be brave enough to admit the label is at the present moment so broad as to say nothing of value to patients. Were a doctor to diagnose osteogenesis imperfecta than drop dead a patient of even the highest IQ and medical training could not imagine what their prognosis may be.

The label “osteogenesis imperfecta” applies to cases as different as a patient with type I who was an inactive child, suffered few fractures, and discovered her OI (Sillence type I) due to an at-home genetic test, and OI so severe it was discovered *in utero* and led to the death of the fetus hours post-birth (Sillence type II)—and everything in between (Sillence types III–IV, genetic types V+).

To say these are one disorder is absurd. Patients want to know what they’re likely to expect, parents of patients want the same. Other diseases are not so broadly labeled; this labeling system leads to numerous negative effects downstream: for one thing, OI life expectancy studies are so muddled as to be useless, combining types I, III, and IV (because the contributing physicians think they look to quantify the statistics of one disease and not two); OI drug trials are the same, attempting to apply treatments to both those with a *null* COL1A1 allele (most cases of Sillence type I OI), structural abnormalities in either COL1A1 or COL1A2 (most cases of Sillence types III and IV), and genetic abnormalities having nothing to do with either. Why should one expect diseases of such vastly different pathologies and effects to be treated the same?

I propose therefore three new major diseases:

* Osteogenesis imperfecta — to cover the existing Sillence types III and IV.
* Fragilitas ossium — to cover the existing Sillence type I.
* Osteogenesis fatalis — to cover the existing Sillence type II.

This is the most logical way of doing it. It reserves the name *osteogenesis imperfecta* for the disease its name, meaning “imperfect bone formation”, most applies to. It brings back a name from the past, *fragilitas ossium*, which is extant in the literature.[1] The only new name is *osteogenesis fatalis*, which is necessary as we have many patients who in confusion self-describe as having type II because a doctor thought their OI was not survivable at birth then turned out to be, even though the definition of type II explicitly disallows including such cases. (Some self-describe as “type II/III”.)

The merger of III and IV is incredibly logical as no one can agree which is which, leading to absurdities like “type III/IV” which means either “something between type III and IV” and also “either type III or IV, I don’t know” (no one knows).

To differentiate these diseases, the genetic cause can be used. For example, my OI would have as its full name “Osteogenesis imperfecta, COL1A2, c.974G>A”.

The current system harms patients and the general public and is intolerable.

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1*Roger S, Francis MJ, Houghton GR (1983). The brittle bone syndrome: osteogenesis imperfecta. London: Butterworth-Heinemann. p. 4. ISBN . OCLC 9784850.*