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An argument of logic for the diagnosis of familial hypomobility

To the Editor:

Flexibility is a continuous variable influenced by many factors. Obviously, environmental factors such as fitness, mobility, and activity are very important. However, the range of innate flexibility of which an individual is capable is determined by their genetic blueprint. The "stretchiness" of connective tissue is determined by the tensile properties of the component proteins, all of which are derived from connective tissue genes. Nature has great symmetry, and mutations are not unidirectional in function. Therefore, the same pathogenetic mechanisms that lead to increased flexibility should also allow for decreased flexibility.

If population flexibility is plotted on a basic Gaussian distribution (Fig. 1), approximately 95% of individuals will fall within 2 standard deviations. Approximately 0.15% on either end will constitute the truly pathologic syndromes of marked increased and decreased flexibility. The former includes Marfan Syndrome and the Ehlers-Danlos Syndromes, the latter, Beals Syndrome. Another 2% of the population will bridge the gaps between the truly pathologic and the average. On one end, this 2% is a grab bag of increased flexibility that is not truly syndromic by formal criteria but often familial in nature. We have termed this familial hypermobility. Increased flexibility can be advantageous if one pursues excellence in dance or gymnastics or employment with the Circ du Soleil. Alternatively, such flexibility can lead to premature degenerative arthritis secondary to increased joint movement.

But what of the remaining 2% between syndromic hypoflexibility and average? This category must exist. By analogy, it would be a grab bag of decreased flexibility that is not truly syndromic but, often, familial in nature.

What symptoms and signs would one expect in heritable hypomobility?

These patients will complain of tightness in ligaments and tendons. That is, they have less flexibility than their peers. And they suffer more ligamentous injury (e.g., the Achilles tear experienced when simply walking). Should they exercise, they find they must significantly warm up before they stretch to prevent injury. Subsequently, they rarely exercise in the early morning but rather wait until daily activities have loosened them up. They might complain that they walk "like an old person" immediately on arising. Often there are similarly affected individuals in the family. On examination, they demonstrate decreased flexibility, both active and passive, compared with individuals of similar age and fitness. On the positive side, they may experience later skin wrinkling than average and, should they avoid ligamentous damage, they may develop later or less osteoarthritis.

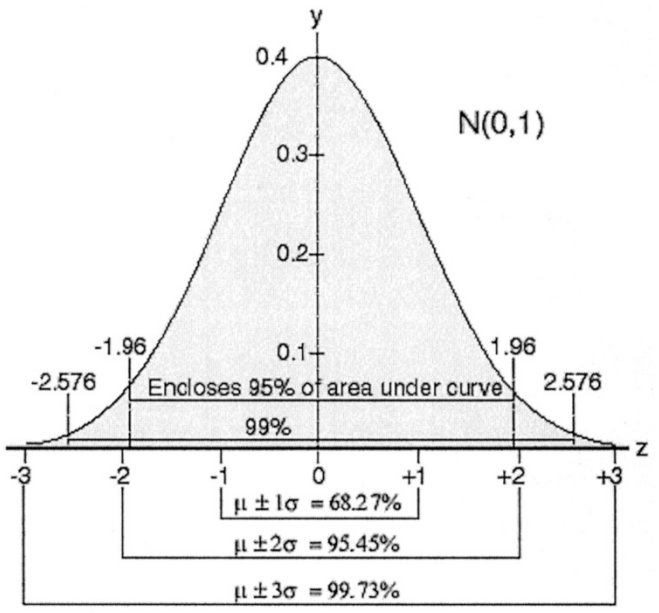

Fig. 1 Basic Gaussian distribution of population flexibility.

Why have we not recognized this condition?

It is difficult to diagnose individuals marginally outside of standard deviation for any continuous trait. Flexibility is a subjective, and there is significant environmental influence. Therefore, the tendency would be to dismiss any complaints of "tight" connective tissue as a lack of fitness.

What advantage is there to recognizing this condition?

From the scientific point of view, characterization of mutations that produce "tight" connective tissue proteins will allow us to appreciate the full spectrum of genetic variability. Clinically, this diagnosis may explain to us and our patients why some individuals seem to be particularly inflexible to the point of suffering injury. Better advice regarding the maintenance of flexibility may be offered and, in particular, these patients should be strongly advised to warm up generally before attempting stretching exercises.

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SUMMARY

Based on basic principles of Gaussian distribution and genetic variability, there must be a significant population of individuals with genetically determined hypomobility.

References

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