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What Does It Mean for a Sign to Be Pathognomonic?

Treat pathognomonic findings with caution.

Practice Perfect is a continuing every-issue column in which Dr. Shapiro offers his unique personal perspective on the ins and outs of running a podiatric practice

Everyone in the medical profession has heard the term "pathognomonic." Without anyone saying so, it may be everyone's favorite term because it simplifies making a diagnosis. Unfortunately, disease in humans is most often a complex affair, and it is rare that any one clinical symptom or sign will give us the diagnosis.

The rash (Figure 1) erythema chronicum migrans, for example, is pathognomonic of Lyme disease. From a definition of pathognomonic standpoint, this means seeing this rash means one has the disease without doubt, that there is a consensus that the rash is not due to something else, and that this finding is highly important to make the diagnosis. However, the absence of a pathognomonic sign like erythema chronicum migrans does not rule out the disease. If you see it, the patient has the disease (i.e., it is "diagnostic" of the disease). If you don't see it, you still can't be sure they don't have it.

In the podiatric community, one may at first blush argue that the complaint of post-static dyskinesia of the heel with pain to palpation at the plantar medial calcaneal tubercle is pathognomonic of plantar fasciitis. But when one looks a little more deeply, this is not truly the case. In fact, the one and only Edward Janeway, MD (of Janeway lesion eponymous fame) warned caution to physicians in 1884 not to overuse the concept of a pathognomonic finding.[1] Dr Janeway said, "Those diagnoses which are rightly denominated snap, may at times be brilliant, but he who indulges in them frequently, no matter how well informed, will be sure to make mistakes."

Figure 1

Who can argue with such wisdom? Considering Dr. Janeway's warning, as one might expect, a number of conditions may lead to pain in the medial heel, although it is fair to say that the association of these signs and symptoms with plantar fasciitis are high.

A Quick Review: Sensitivity and Specificity

When discussing the details of a pathognomonic sign or symptom, it's important to understand what we're actually talking about. This requires a diversion into statistics.

Oh no! Statistics! Dum Dum Dum, Dummmmm!

Yes, statistics, my friends. This is very important, especially in the medical literature when we're comparing various tests for the diagnosis of disorders and understanding what they are capable of doing. For example, how well is palpation of pedal pulses able to determine the presence of peripheral arterial disease? Boyko found that in patients with ABI < 0.5, palpating pedal pulses (non-palpable or diminished dorsalis pedis and posterior tibial pulses) had a sensitivity of 65.2 and a specificity of 78.3.[2]

Table 1 shows the conceptual 2x2 table of disease diagnosis. In research, this table is used to calcu-

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TABLE 1

Disease Diagnosis

	DISEASE PRESENT	DISEASE ABSENT
Test Positive	True Positives (TP)	False Positives (FP)
Test Negative	False Positives (FP)	True Negatives (TN)

Pathognomonic *(from page 28)*

late sensitivity, specificity, and positive and negative predictive values. A quick definition of these terms, as they arise from the table, is listed below.

2x2 table for determining probability of a diagnosis based on a certain test. TP = true positives; FN = false negatives

- *Sensitivity (the true positive rate)*—the proportion of patients with the disease that test positive.
  - Not affected by disease prevalence.
  - TP / (TP + FN)

- *Specificity (the true negative rate)*—the proportion of patients without the disease who test negative
  - TN / (TN + PF)
  - Not affected by disease prevalence.

- *Positive Predictive Value*—proportion of patients with a positive test who have the disease.
  - TP / (TP + FP)
  - Affected by disease prevalence.

A pathognomonic sign, then, has a very high sensitivity (since having the sign means having the disease) and positive predictive value (for the same reason) but does not require a high specificity (since not having the sign does not have to mean an absence of the disease). This of course begs the question what level of sensitivity/specificity/positive predictive value is the cut off for a sign, symptom or test to be considered pathognomonic?

Unfortunately, after a somewhat long literature search, there wasn't an answer. Indeed, if one were to look at the literature discussing certain signs considered pathognomonic, there's a paucity of reporting on the statistical data. Many of these appear to be considered pathognomonic as shared wisdom handed down from others. When you look at many of these findings, it's common to see that the sign in question diagnoses a disorder with high fidelity, but it usually also includes other diseases as well.

The longer you are in practice, the more you see the large number of ways many disorders present. With that in mind, it is recommended to treat pathognomonic findings with caution and to fall back on the regular model of abductive reasoning that most doctors use when diagnosing and treating disease: obtain a detailed history and physical, consider the differentials, order confirmatory testing when needed and re-evaluate all diagnoses at subsequent visits. With these skills firmly in place most disorders will fall to your rational diagnosis and treatments. PM

What level of sensitivity/specificity/positive predictive value is the cut off for a sign, symptom or test to be considered pathognomonic?

References

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[2] Boyko EJ, Ahroni JH, Davignon D, Stensel V, Prigeon RL, Smith DG. Diagnostic utility of the history and physical examination for peripheral vascular disease among patients with diabetes mellitus. J Clin Epidemiol. 1997 Jun;50(6):659-668.

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