On Syndromes

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I assume that most readers have not heard of "notalgia paresthetica" (NP). It is a "real" syndrome. I had not heard of it until I received an unsolicited email about it from God knows where. I was struck by its unusual characteristics (see below) and asked my friend, a retired pediatrician, if he had heard of it. He hadn't, so, after looking it up, I explained to him what it is. NP is a "not uncommon" syndrome of episodic unilateral itching in the upper back, T2–T6. My friend then asked if I recalled him asking about his recurrent problem of an itchy right foot and my response of, "don't know, never heard of it. Some people get weird stuff. Not to worry." He opined, probably correctly, that he clearly had a variant of this new-to-us entity, notalgia paresthetica, so that I should take it more seriously. He mildly chastised me for "blowing him off." He was pleased that he now had a bona fide, "real" disorder, "notalgia paresthetica *variant*." Itching, of course, is a common symptom of many different disorders, and many people itch for unidentifiable reasons, but only some have episodes of itching affecting half their back, and no one knows how many have an episodically itchy single foot.

The first citation for NP in PubMed was published in *Neurology* in 1978, but the authors gave credit to Astwazaturow as the first to report this entity in 1934. Given the name of the discoverer, it is easy to understand why this syndrome has not been named after him, at least in the Western world.

Learning of this disorder got me to thinking about what a syndrome is and what it means to have one. A syndrome is, of course, a collection of signs, symptoms, or laboratory findings that are linked together in a statistically and clinically significant manner to form a discrete disorder. Some are considered diseases whereas others simply stay a "syndrome." Sometimes the term is used as an umbrella descriptor to encompass several discrete disorders, such as psychotic syndromes, failed back syndrome, Parkinsonian syndrome, etc. Some of these have ICD 10 billing codes. NP, for example, has an ICD billable code, G54.8, which appears to be a non-specific code, meaning it applies to several different disorders, but it gives a degree of robustness or life to this unusual collection of symptoms. In medical-legal practice, "if you didn't chart it, it didn't happen" translates, in daily medical practice to, "if it doesn't have an ICD 10 code, it's not billable and if it's not billable it doesn't exist." So, "painful legs and moving toes," my most favored name for a neurological syndrome (not a disease, since there is no known pathology), does not have an ICD 10 code. It is therefore not billable, and therefore doesn't exist, at least for a medical visit. I can bill for the painful legs with any number of diagnoses, but not the moving toes. There are 104 citations in PubMed for painful legs and moving toes, starting in 1971, and 146 for NP, starting in 1978, which is not a large difference, making me wonder why one is billable and the other not. The number of publications about a syndrome apparently doesn't seem to influence its ability to obtain an ICD code, and therefore medical importance. Yet, much rarer syndromes, like being sucked into a jet engine, has its own billing code (V 99.73), although it boasts few publications, and who would bill for it remains a puzzle.

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Defining syndromes

There is value in defining syndromes. If we identify three problems in a syndrome that includes four or more, we are compelled to look for the missing problems, which is helpful for the patient. Identifying a syndrome may allow us to review the spectrum of discrete diseases that are included under the inclusive umbrella term of the syndrome. Thus, syndromic names have diagnostic implications. In addition, syndromic names may provide the relief we need to feel in "knowing" what is wrong. Giving "Long Covid" a name is reassuring to both patient and caregiver. The problem has a name.

It "exists." There is a tangible disorder. Others have it. It is not so much that, "misery loves company," as it is that no one wants to have a disorder that can't be "googled," or discussed in a meaningful fashion with others without going into personal details.

While syndromic names often precede identification of specific disease states, it sometimes works the other way, in which presumed specific diseases are found to be collections of possibly unrelated pathologies. In very recent times, in my field of movement disorders, the disorder cortico-basal degeneration, a disorder with a characteristic pathology, was found to be clinically inseparable, in occasional cases, from at least two other distinct pathological entities, all of which, so far, share some pathological features, and the clinical signs are now combined as corticobasal syndrome. But these disorders, although sometimes looking like the others, usually look quite different, so that the individual pathologies have been lumped together into the "tau-opathies," as they share abnormalities of the tau protein, although each has a different one. Occasional patients have clinical features of more than one of these disorders. I have diagnosed a patient as having a "tau-opathy" as it did not fit a single disease set of clinical criteria, but had features of a few. This turned out at autopsy to have been correct. It shared pathological features of more than one distinct tau-disorder. Using a syndrome's name can allow a degree of flexibility in diagnosis, and this can be very helpful both for guiding patient care, as well as providing an explanation to the patient of the illness, so long as we understand that attaching a syndromic diagnosis is a label and not a pathology.

We should not be fooled into thinking we know more than we do by simply attaching a name. ❖

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