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Abraham Lincoln and the 'Lincoln sign'

Here is a gentleman of a medical type, but with the air of a military man. Clearly an army doctor, then. He has just come from the tropics, for his face is dark, and that is not the natural tint of his skin, for his wrists are fair. He has undergone hardship and sickness, as his haggard face says clearly. His left arm has been injured. He holds it in a stiff and unnatural manner. Where in the tropics could an English army doctor have seen much hardship and got his arm wounded? Clearly in Afghanistan.

— Sir Arthur Conan Doyle, *A Study in Scarlet*

Even today, Marfan syndrome can be very difficult to diagnose

IT IS NOT COMMONLY KNOWN that the character Sherlock Holmes was in fact based on a physician. Sir Arthur Conan Doyle met Dr. Joseph Bell while training in medicine at the University of Edinburgh. Besides having a charismatic bedside manner, Bell was considered a master of observation, logic, deduction, and diagnosis. His students would delight in his ability to deduce an illness from the doorway without speaking to or examining the patient. It is precisely this quality, inherited from Dr. Bell, that makes Holmes such an intriguing literary character.

Physical diagnosis still inspires the same feelings when finding pathognomonic evidence of disease or when elucidating eponymous signs and symptoms. Indeed, the historical background of eponymous signs can often be more interesting than the diseases they describe. The realm of cardiology is full of eponyms and acronyms,[1] and no disease more than aortic regurgitation. From Corrigan pulse to de Musset sign, the names of both physicians and patients can be found.[2] One very interesting story combines a logical deduction worthy of Sherlock Holmes with a relatively little-known sign named for Abraham Lincoln.

A BLURRY FOOT IN A PHOTOGRAPH

The story, as related in the book *Lincoln in Photographs*,[3] is a dialogue between President Lincoln and Noah Brooks, a newspaperman and confidant. While reviewing a recent photograph of himself, Lincoln didn't understand why his foot looked so blurry (**FIGURE 1**). Brooks suggested that throbbing of the arteries may have caused his foot to be moving. Any movement would have caused blurring due to the long wait during exposure that was necessary for photographs at the time. Interested to see if he could recreate this, the President crossed his legs and watched his foot, exclaiming "That's it! That's it!"

DID LINCOLN HAVE MARFAN SYNDROME?

The theory that Lincoln had Marfan syndrome was first proposed by Gordon[4] in 1962. Two years later, Schwartz[5] made the clinical diagnosis of Marfan syndrome in a 7-year-old boy who had a common relative in Lincoln's paternal great-grandfather.

Schwartz later suggested that Lincoln had aortic insufficiency due to Marfan syndrome and used the above story and photograph as evidence, stating "the clinician will recognize in the suspended throbbing and pulsating foot of the President, a phenomenon entirely compatible with the hemodynamic effects of aortic regurgitation."[6] He then went further to claim that the President was in declining health and would have been unlikely to survive a full second term.[7]

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The preponderance of evidence used to make the diagnosis of Marfan syndrome in President Lincoln comes from descriptions of him by contemporaries. His physical stature, 6 feet 4 inches tall, was extraordinary for the time. He was very thin, and his arms and legs were very long. Although his long-standing law partner and biographer William H. Herndon described him as short-breasted and with a sunken chest, at autopsy he was not said to have a chest deformity, and one physician remarked surprise at seeing such "powerful musculature built upon strong bones."[8] He was described by several as "loose-jointed," or having a loose build. His hands were also very large. However, his hands were not thin and spider-like, but rather massive and powerful. His joints were certainly not loose, as attested to by his almost mythic stature as a wrestler and his feats of strength that were exhibited almost up to the time of his death.

Other than the story concerning the photograph, there seems to be no evidence that Lincoln had aortic insufficiency. He suffered from several maladies during his lifetime, including recurrent malaria and varuloid, a mild form of smallpox. At no time, however, was he known to have a heart murmur or to have any physical findings of heart disease. Also, there was no mention of aortic abnormalities at his autopsy.

Does a blurry toe of the seated Lincoln provide proof of a "suspended throbbing and pulsating foot"? Although many parts of the celebrated picture are blurry owing to their distance from the focal point of the camera, we do have the reassurance of the President and Mr. Brooks that his leg was indeed moving when the pose was recreated. However, this effect is likely to be recreated in many who have neither aortic regurgitation nor elevated pulse pressure.

WHAT MODERN GENETICS CAN TELL US

Marfan syndrome, a common inherited disorder of connective tissue, can be very difficult to diagnose. The typical features of tall, thin body habitus and joint hypermobility are more often seen in those without the disease.

The genetic mutation for most phenotypes occurs in the fibrillin-1 gene, localized to chromosome 15q12.1. This gene is very large, and mutations, which are very heterogeneous, cause disruption of microfibril assembly in the extracellular matrix.[9]

Does this man have aortic insufficiency?

FIGURE 1. Some believe that the blurriness in Lincoln's left foot in this photograph is due to excessive pulsation in the popliteal artery due to aortic regurgitation secondary to Marfan syndrome—the eponymous Lincoln sign.

PHOTOGRAPH BY ALEXANDER GARDNER, NOVEMBER 8, 1863.

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Although it is variably expressed, Marfan syndrome has full penetrance. Lincoln had only one son who lived to adulthood, Robert. Neither he nor any of his children were known to possess the Marfan phenotype. Celebrated geneticist Victor McKusick[10] placed Lincoln's chances at 50:50.

Although this controversy may seem to be just an interesting historical footnote at first, the issue is far from trivial for patients with genetic diseases, who saw in Lincoln a way not only to deflect a prevalent negative attitude, but also to increase awareness. A panel of experts was convened by Congress to decide if a request for a sample of Lincoln's DNA was ethical and met the criteria for scientific scholarship. Dr. McKusick and the panel did encourage the study to proceed; however, no DNA has been released as yet.[11,12]

Whether or not Lincoln did have Marfan syndrome, the possibility of diagnosing this based on descriptions of him and a blurry photograph of his foot is just the sort of Holmesian pleasure that can be found in the lore of medical history.

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