Finding more disease genes, faster

When you read stories about scientists identifying a new link between Gene X and Disease Y, underlying studies vary in quality. At one extreme, you get papers which show a variant of Gene X is common in a small group of people with Disease Y and not in healthy controls... and that's it. You don't know if X is really responsible for Y, or even if the result is genuine and not a false alarm produced by small numbers.

At the other extreme, you have this—a smorgasbord of experiments that identify 18 new genes behind hereditary spastic paraplegias (HSPs). This diverse group of genetic disorders all involve damage to the long neurons running between the brain and spinal cord, leading to stiffness and involuntary contractions in leg muscles.

Read the full, original story: Now This Is How You Find Disease Genes

Additional Resources:

- 11 new gene variants linked to Alzheimer's disease, CBS News
- Big data firm to help NIH's Undiagnosed Disease Program commercialize its genetic analysis techniques, MedCity News
- Study Expands the Cancer Genomics Universe, Bioscience Technology