Simple gene test can fully prevent devastating skin-peeling condition

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It starts with a fever, a cough, red eyes, the kind of symptoms that might accompany a bad cold. Then, everything gets much worse. A rash appears on the skin, with target-like dots that blister and burst. Lips crust and bleed. Eyes become inflamed. The throat develops such painful ulcers that swallowing becomes impossible. Skin starts peeling off in sheets. The damage is so extensive that patients typically end up in burn wards, in excruciating pain, unable to eat, urinate, or open their eyes. Many beg to die; some do.

In its milder forms, this condition is called Stevens-Johnson syndrome (SJS). “It’s the worst thing I’ve seen in 30 years in clinical medicine,” says Teri Manolio at the National Human Genome Research Institute.

SJS/TEN is a disease of devastating irony. Most cases happen when people take drugs that are meant to improve their health and their bodies revolt in catastrophic fashion. These hypersensitivity reactions are rare. They only affect people with specific genetic variants in a cluster of immunity genes.

Which means that SJS/TEN should be almost entirely preventable. As Wasun Chantratita from Mahidol University puts it, it’s “the low-hanging fruit of genomic medicine.” To deal with it, you don’t need to edit genes, or turn to stem cells, or prescribe drugs that target mutations in a patient’s DNA. You just need to screen people for the risky variants and withhold the triggering drugs.

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