



WHY GENOMICS TEST IS DONE

Hospital - NICU

Emma is born and suffers severe seizures.

Primary Care

Emma is now in elementary school and is suspected to have ADHD.

OB-GYN

Emma's genomics tests had revealed she was a higher risk for breast cancer. Her physician instituted early screening protocol and was able to detect the disease sooner, when Emma was 42.

Patient Home

At 69, Emma is being treated for multiple conditions including high blood pressure, high cholesterol, GERD and depression. Her conditions are well controlled, and she leads a full and active life.

WHAT HAPPENS

Genomics test (germline and pharmacogenomics/PGx) is performed, uncovering the specific genetic disease causing the disorder. The clinician is able to arrive at the correct diagnosis earlier and prescribes the appropriate medication to control Emma's seizures. She is discharged to home.

Physician refers to the genomics test performed at birth to verify suspicions and decides to prescribe Adderall, which Emma metabolizes normally (discovered through the PGx test at birth). Emma responds and behavior returns to normal.

Disease is diagnosed early and somatic tests (tumor profile) are ordered. Following lumpectomy and using information about the mutation of the tumor (instead of relying only on the site of the cancer), physician prescribes Tamoxifen to achieve better results and avoid drug-drug interactions with other medications Emma takes.

Genomics testing not only revealed variants that helped Emma's internist diagnose her conditions in a timely manner, it allowed the doctor to prescribe medications that she would respond to and from which she would suffer fewer side effects. In addition, the doctor was able to evaluate possible drug-drug-gene interactions, which meant no adverse reactions between the medications.

WHAT IS AVOIDED

Without genomic testing, it would be more difficult and time consuming for the physician to arrive at a diagnosis. This could result in lengthy hospitalization, readmission to the hospital, multiple office visits. Because of genomic testing, what would have been the the seventh-line medication is now the first, avoiding months of healthcare problems for Emma.

Uncertainty about what is contributing to Emma's behaviors and what medication will be most effective with fewest side effects.

Emma responds well to prescribed chemotherapy and recovers quickly. No additional treatment (e.g., radiation) is required and cancer is in remission.

Emma has not been hospitalized or in the ED since her breast surgery decades earlier. PGx testing has helped her ensure her conditions are controlled and adverse drug reactions don't cause problems like dizziness or falls, which could land her in the hospital or skilled nursing facility.