

Genomics and Precision Medicine – Treating Patients at the Molecular Level

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Genomics is a science that analyzes a person’s genome, or DNA and genes. **Precision Medicine (PM)** is the new medical model that uses the genome to drive treatments for a specific patient. This is also known as Data Driven Medicine.

Data Driven Medicine uses all data collected to make medical decisions for the individual patient. The thought is that with using full patient data, with the help of the genome, doctors can derive the right drug to the right patient.

By sequencing the genome/DNA, technicians can take a patient’s gene and match it up against a base, or “Healthy” gene, to look at the variability of the specific gene that is causing the patients symptoms. Then, drug testing and screening can be done on the specific gene in order to determine which chemical would aid in correcting or healing the variance. We now have a process where “Science is Driving Medicine” and are able to find treatments for diseases at the genetic level that can manage patients’ conditions. In one specific example, a researcher was able to identify and treat a rare disorder using genetic profiling. He was able to isolate a damaged gene and to identify the specific enzyme that was not being produced in the patient’s cells. Physicians used pharmacological data to identify specific drugs that augment the enzyme within the body. In this case, an over the counter drug (Prilosec), which is a protein pump inhibitor used to treat heartburn, was able to treat the enzyme deficiency due to its side effects. The patient, a child, has subsequently been able to thrive in ways thought impossible just a few years earlier. Such is the power of Precision Medicine.

HL7 FHIR (Fast Health Interoperable Resources) will provide the means to access this data used by Analytical and Health tools in order to identify problems and patterns in peoples' health from their clinical data and make recommendations to clinicians for treatment. HL7 clinical data standards will play a key role in the technology involved in sending, retrieving, and storing this clinical, pharmaceutical, and genomic data.

As more genome sequencing and treatments are completed, there will be more data available for clinicians to use for matching genetic anomalies to effective treatments for the patients who desperately need them.