Amazing how parts of a chromosome and chromosomes inside the nucleus of a cell. National Institutes of Health what you learn in medical school changes years later. Some examples come to mind. Intravenous aminophylline drips and inhaled isoetharine are no longer first-line therapies for status asthmaticus (Little did we know we were giving significant adverse effects). Behavioral therapies and medications have replaced psychoanalysis. (Recently I uncovered a paper I wrote as a fourth-year medical student entitled, "Childhood Enuresis: An Interplay Between ID and Superego."

I dread to read past the title page. Who knows what embarrassing sentences lurk within?) Back then, the practice of genetics consisted of comparing physical findings of children with chromosomal patterns seen on karyotypes and telling parents the typical prognosis associated with those karyotypes. (I remember as a medical student thinking how frustrating it was to have to cut out those individual chromosomes from a photograph and match them to a chart to make a diagnosis and how boring it was not to be able to do much once a diagnosis was made). At least years ago, "The Human Genome Project" held the promise of curing genetic diseases.

Siddhartha Mukherjee, author of "The Gene: An Intimate History." Medical beliefs and practices since then have indeed changed, particularly in the field of genetics. We now have genomes, proteomics, metabolomics, and epigenetics. FISH, SNP chips, and whole genome sequencing have surpassed karyotyping. Deciphering the human genome was the beginning not the end of understanding and treating genetic diseases. We discovered that a genotype is affected by many factors, including the environment
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from a generation before. We encountered perplexing permutations of mutations. A genetic laboratory finding may not lead to a predicted phenotype after all. Genetics was no longer boring but instead more challenging to master since those days of cutting out chromosomes from a photograph.

In "Beyond the Genetic Diagnosis: Providing Parents What They Want to Know," Drs. Robert A. Saul and Stephanie Hall Meredith use Down syndrome as a model to discuss one very important issue in genetics: Making a genotype diagnosis does not necessarily predict the outcome of the phenotype. That simple statement means that we as pediatricians must work with parents of children with genetic syndromes to discover each child’s individuality and unique potential.

James Glieck, in his May 15, 2016, New York Times Book Review of "The Gene: An Intimate History," by Siddhartha Mukherjee, could not have said it better: "The gene is, and is not, the determiner of our identity ... As we learn how our genome defines us, we also learn how we transcend our genome." Fascinating.

Further Reading

- Peds in Review on Facebook
- AAP Journals on Twitter