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The structure of DNA, the basic blueprint of cell function and reproduction of all organisms, was identified in 1953 by Watson and Crick. This was soon followed by the determination of the DNA code for gene structure and function. A less famous molecule, RNA, is necessary to carry out the instructions for making proteins from the genes encoded in DNA. In 2012, Doudna and Charpentier, identified a RNA structure, known as CRISPER-Cas9, which can insert, delete, or edit the genetic code of DNA in every species on earth.

To treat many diseases, CRISPER offers the potential to edit and repair mutated genes directly in human patients. Because CRISPER allows precise and relatively straightforward DNA editing, it has transformed every genetic disease into a potential target. Physicians have already begun treating some cancers with immune cells whose genomes have been edited. Gene therapy has resulted in life-changing treatments and, in some cases, lifesaving cures.

The Maine Newborn Screening Program was established in 1965, initially to identify infants with PKU, which causes severe mental retardation if not diagnosed and treated in the newborn period. In 1978, after completing a fellowship in genetics at Boston Children's Hospital, I came to Maine and established a clinic at MMC for newborns with rare inherited metabolic disorders. The newborns were identified clinically or through the state newborn screening program. By 1978, three more conditions had been added to the screening panel. Treatment consisted of dietary measures.

At present, over 30 conditions are now being screened through the Maine Newborn Bloodspot Screening Program. New conditions are being added due to breakthroughs in laboratory and treatment technologies, such as gene sequencing and gene therapy. Recent examples include drug therapy addressing the basic cellular defect in cystic fibrosis and gene editing to treat sickle cell disease.

There are profound implications of gene editing and therapy. It can be used not only to treat diseases in patients, but also to prevent diseases in future generations. The CRISPER-Cas9 technology is so simple and efficient that it is possible to edit the germline, allowing the transfer of genetic information from one generation to the next. It is possible to sequence the whole genome of a newborn from the screening filter paper blood spot and identify pre-symptomatic inherited conditions which express themselves in adulthood, such as inherited cancers and heart disease. Early identification would allow measures to prevent the onset of disease.

The technology has developed to the point that a Division of Genomic Medicine should be established within a healthcare delivery system in Maine. Such a division should collaborate with primary and specialty care, genetic counselors, clinical and research laboratories, CDC, and companies with interest and expertise in pharmacogenomics, artificial intelligence, bioinformatics, biobanks, gene sequencing, and somatic cell editing. The future is now.