AS I SEE IT

Genome discoveries could help children, families

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Now that the public and private drafts of the human genome sequence have been announced, hundreds of thousands of previously unknown genetic differences can now be used to distinguish otherwise identical human chromosomes from one another.

What do these developments mean for you, now and in the future?

At Children's Mercy, we are building a world-class genetics program to address these issues and to take advantage of the latest developments to provide better care to children and their families. Children's Mercy geneticists embrace the opportunity to discuss these issues openly and advance the cause of medical science through greater understanding.

Perhaps the most immediate effect will come from increased numbers of tests to detect defects in previously known disease genes. Tests for many genetic mutations known to cause diseases have not been available because the DNA sequences have been incomplete. The genome sequence will now simplify the development of these tests. Many of the new tests will be considered experimental at first because results for different patients will have to be verified in multiple laboratories.

The complete sequence will accelerate the discovery of genes that cause or predispose to many different diseases. Geneticists narrow down a region on the chromosome containing a disease gene by searching for genetic differences found only in family members who are diagnosed with a particular disorder. With the genome sequence, it will take months (rather than years) to thoroughly sift through all of the candidate genes in a particular chromosomal region.

Once genetic differences are found in individuals affected with genetic disorders, it may be possible to develop drugs that potentially correct genetic defects or prevent symptoms from developing. Therapies targeted to the cause of the disease may be more effective and in some cases may be the only effective treatments for some diseases. Gene-based products to replace deficient proteins are already available for some rare disorders. Unfortunately, drug development and FDA approval can be lengthy and expensive, so don't expect to see a large influx of new drugs resulting from the genome sequence on the market for several years.

The future will bring tests to estimate how likely we are to develop certain diseases long before the symptoms become apparent. We will all have to decide whether to take preventative measures, for example, to make lifestyle changes or to begin therapies that could forestall these conditions. Knowing the odds of developing a genetic disorder may help us decide whether to act, but each of us has an individual threshold for tolerating the uncertainty of these predictions. Interpretation of genetic findings will be even more complex for conditions where more than one gene has an impact on disease severity.

We need to ensure that the risks for developing genetic diseases are correctly estimated. Patients and their health-care providers must be properly counseled about these risks and should understand the meaning of this information. Genetic privacy should be protected by federal statute because of the possibility, however remote, that this information might be misinterpreted.

These are all serious issues, but we believe they can be adequately addressed and that all of us will benefit from these scientific breakthroughs.

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