Newborn Genetic Screening: Changing the Future of Pediatrics

Simply defined, genetic tests are “the analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes or karyotypes for clinical purposes” (Genome.gov). Such tests are useful in predicting an individual’s risk of contracting a disease and identifying carriers. More recently, a new wave of molecular genetic testing has surged. These new tests have the capabilities of scanning 2 million markers along a human’s entire genome in order to locate the ones correlated with specific conditions (Navigenics.com). With an increase in use of these technologies, it is likely that parents will screen their children in order to obtain valuable genetic information that bears the potential to help in their child’s development. Though genetic screening could provide several life-long benefits for infants, several societal inadequacies appear when considering an increase in newborn genetic screening. The individual child may benefit, but society could suffer from its inability to provide adequate healthcare in response to the information generated by genetic tests and health disparities based on socioeconomic status are expected to increase. Thus, access to knowledge regarding the current state of genetic testing and the change in pediatrics that results from newborn genetic testing becomes of supreme importance for the general population in order to ensure informed debate.

The principle behind genetic screening lies in the marvel of examining single nucleotide polymorphisms. These single nucleotide polymorphisms, commonly known as SNPS, “are
inherited variations [of the human genome] involving a single nucleotide base” (Sadava et. al 356). These variations can be detected through sequence comparison, and through their analysis can indicate predispositions for genetic diseases (23andMe.com). Sites such as 23andme, deCODE Genetics, and Navigenics capitalize on these scientific advances by facilitating at-home genetic screening. For example 23andMe advertises that their company “can help you interpret your genetic information and understand how your DNA makes you uniquely you” (23andMe.com). These tests are available in several different forms depending on the company in question. One way of completing the genetic test is to observe chromosome number and shape for obvious abnormalities; others examine small unusual portions of individual proteins or sections of DNA (FTC.gov). Typically, all that is required to conduct these tests is a blood sample or a swab from inside the cheek. Through these sites, the process of genetic screening becomes accessible for all. Consequently, parents are now able to conduct molecular genetic screening of their children before the advent of these technologies in hospitals and clinics. Now that genetic information is literally available at one’s fingertips, genetic testing on behalf of children by curious parents can be expected to increase.

According to the Federal Trade Commission, companies offering this form of testing assert, “genetic tests can measure the risk of developing a particular disease, like heart disease, diabetes, cancer, or Alzheimer’s” (FTC.gov). For the curious parent who chooses to genetically screen their newborn or toddler, this action is the key to unlocking the secrets of the genetic makeup of his/her child, and bears the potential to aid the parent in making informed decisions on fostering the best environment possible to host his/her child’s genes. From the screening of a child, parents can identify the risk their child bears for developing many diseases and can therefore take direct action to mediate against the precursors that lead to future onset of the
disease. Using obesity as an example, if a parent finds that their infant is more at risk for developing the disease as an adult than an average individual in the population, it is very likely this parent will take the appropriate steps in order to prevent obesity and its co-morbidities. This informed parent would be more likely to promote healthy eating patterns and encourage the self-regulation of their child. Additionally, this parent may have greater motivation than the average parent to encourage their child’s physical activity and discourage television time (Pediatrics.about.com). Outside of the consideration of future health outcomes, parents can even fashion a child’s environment to provide them greater comfort with the knowledge available through genetic testing. For example, if through genetic testing they find their child has the gene that makes many vegetables taste bitter (Harmon), parents will probably wish to consider alternative ways of introducing the same vitamins and minerals vegetables donate rather than force-feeding their children Brussels sprouts.

Who should make the recommendation of the appropriate dietary supplements in this case? Why, a pediatrician of course. Through this simple example, it becomes evident that the field of pediatric care will change greatly due to newborn/toddler molecular genetic screening. Normally, the field of public health has been the leader in encouraging preventative health measures through services such as entomology, pest control, and public health inspections (Wikipedia.org). With this eminent wave of newborn/toddler genetic screening, preventive medicine, medicine which functions at the level of the individual and engages with preventing diseases rather than curing them (Wikipedia.org), will largely be left in the hands of pediatricians. In addition to curative medicine, pediatricians will now be pressured to act as genetic counselors in hopes of early management of disease with the expansion of newborn screening (Wise).
Assuming pediatricians are able to help parents in preventing poor health outcomes, the advantages to genetically screening newborns are clear, particularly due to the enhanced access to clues for an infant’s best possible environment given their particular genes. Despite the advantages, newborn genetic screening poses several societal challenges that require acknowledgement and discussion. One challenge is in determining whether or not newborn genetic screening bears the potential to limit a child’s future development. The best environments for a child’s given genes are not always ideal for parents. In a New York Times report titled, “My Genome, Myself: Seeking Clues in DNA” author Amy Harmon comments on her decision against genetically testing her daughter’s DNA: “… I didn’t want to regard anything about her as predestined. If she wants to play the piano, who cares if she lacks perfect pitch? If she wants to run the 100-meter dash, who cares if she lacks the sprinting gene?” (Harmon). A challenge arises for the traditional parental voice of encouragement when genetic predispositions are known. “You can do anything you put your mind to” has the potential to become “You can do anything that your genes allow.” Policy and/or other interventions may be required to prevent this shift in parenting.

The widening of health disparities in developed nations due to newborn genetic screening poses another greater societal challenge that results from an increase in newborn genetic screening. A health divide would form between children whose parents have the monetary means for genetic testing and those who lack the access to genetic testing (Wise). Historically, scientific advancements create greater inequality, which is no surprise considering the price of genetic screening is currently one thousand dollars (Harmon). For some families this amount poses no challenge, a necessary expenditure for their child’s best health. Unfortunately, several families would be unable to provide their child the opportunity of preventing future health outcomes
through prior knowledge of their genetic predispositions. Policy and other governmental provisions, quite possibly monetary, would also be necessary to prevent this form of inequality.

Unmistakably, despite the aforementioned challenges, immense advantages can still be expected to accompany newborn molecular genetic testing. This is only the case, however, if the public is prepared for the information presented in these genetic reports. More importantly, benefits from newborn genetic screening only result if pediatric health professionals are prepared to provide the consequential preventative medicine required by a growing amount of newborn genetic testing. On May 28th, 2008, in a lecture titled “Children’s Health Policy” for Human Biology students at Stanford University, Dr. Paul Wise discussed the mismatch between the structure of pediatric care developed in the 1950’s and 1960’s that most pediatricians are currently versed in and the rapidly changing face of pediatrics. His argument: the new wave of genetic screening will forcibly shift pediatric medicine from a field of acute care to a field of preventative care (Wise).

Other sources echo Wise’s sentiments and offer a pessimistic outlook for how well physicians will adjust to this change in pediatrics. Researchers from RAND Health and the Department of Veterans Affairs recently synthesized all studies published from January 2000 to February 2008 concerning the delivery of genomic medicine for common chronic diseases. This study was conducted in order to develop a sense of the status of the use of genomic medicine in developed countries to diagnose, prevent and treat common chronic adult illnesses. The results were not as promising as had been hoped, and demonstrated primary care physicians felt “woefully underprepared” to deal with genetics in their practices. Physicians found themselves “having neither the time nor the skill necessary to obtain and interpret family histories that might detect disease patterns that merit a referral for genetic testing or specialty consultation” (RAND).
Now that genetic information of this sort is available for children, what can be done about it?

Unfortunately, very few health professionals are able to answer this question.

Infants could benefit greatly from newborn genetic screening of their SNPs. Current resources readily provide access to any given child’s susceptibility to certain diseases, which is information that potentially aids parents in enhancing a child’s environment and daily routine to promote the best development of their child’s genes. However, it is important to note that this foreseen benefit could prove to be a disappointment when considering the challenges society must face due to growing use of genetic testing for newborns and children. The general public and health professionals need to be fully informed regarding the implications of this movement toward genetic screening. Decisions and policies need to be established in order to prevent outcomes such as the limiting of children’s free will along with the furthering of health disparities due to socioeconomic status. More importantly, the pediatric healthcare system needs to become more innovative in order to incorporate genetic counseling and preventative medicine into current practice. The genetic revolution will affect newborns and children very soon.

Parents, citizens, consumers, and health professionals cannot be taken by surprise.
Works Cited


<https://www.23andme.com/gen101/phenotype/#learn.>

"At-Home Genetic Tests: A Healthy Dose of Skepticism May Be the Best Prescription."


"Health Professionals and the Public Unprepared to Make Use of Genomic Medicine for Adults."


