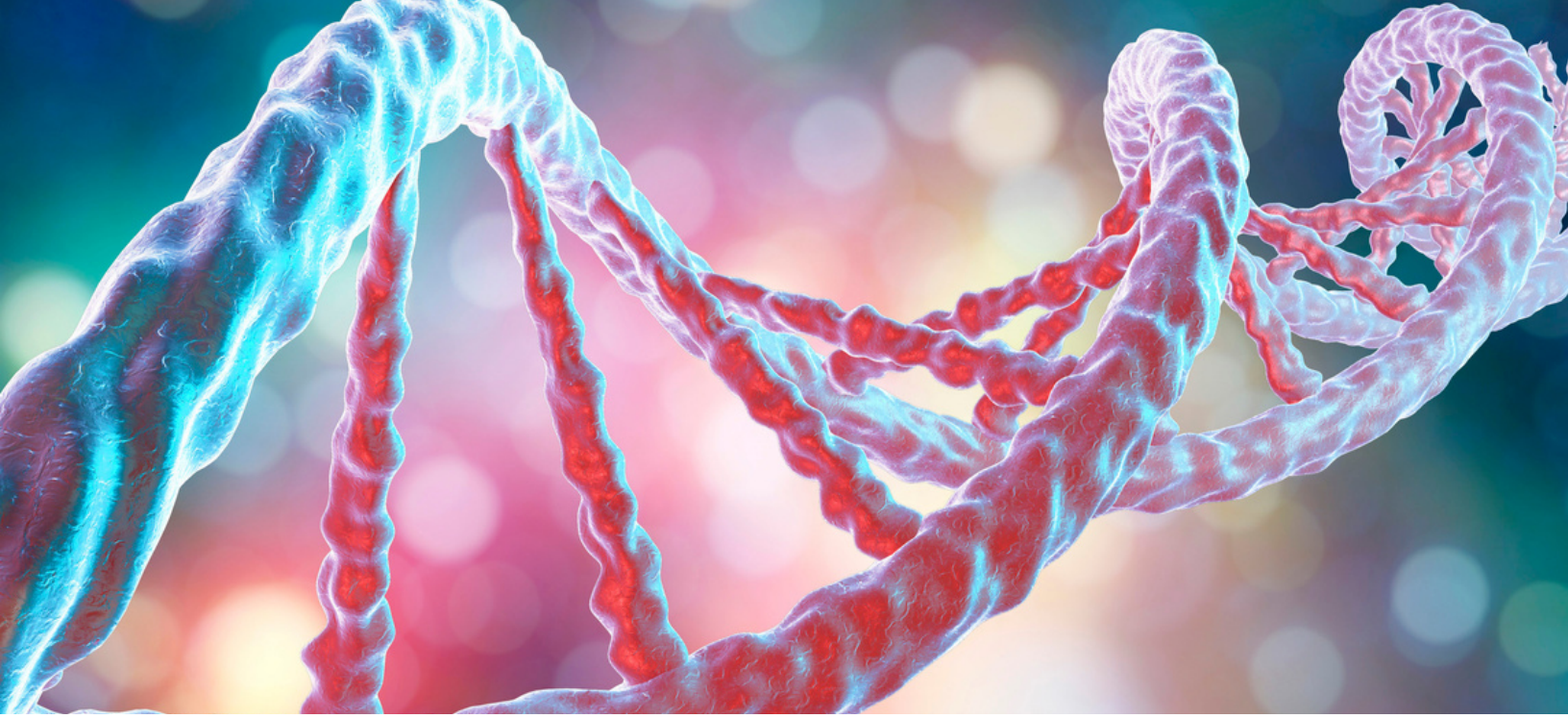
Parent's Guide to Newborn and Infant Genomics

By: Fore Genomics, Inc.

Outline:



Introduction:

Imagine holding a storybook that contains the unique tale of your child's life. Each page is filled with chapters yet to unfold, characters waiting to emerge, and plots that are beautifully complex. This isn't a fantasy; it's the reality of what lies within our DNA. Welcome to the world of genetic testing where we can translate these genetic tales into comprehensible insights. Join us on this enthralling journey as we explore what genes, chromosomes, and proteins tell us about your child's health and the captivating story that is their genetic legacy.

But first, just what exactly is a genome? A genome consists of all of the genes in your child. Your child has over 20,000 genes that are defined by the 4-letter language of DNA. Your child’s genome is 3.2 billion letters long! That’s amazing! It would take a person who types 60 words per minute, 8 hours per day, over 60 years to type your child’s genome. That computational challenge used to prevent the science and medical community from using the information in the genome to directly diagnose patients. In fact, the first fully tested human genome cost $100,000,000 using the technology available 20 years ago. Today, modern computation and innovation has changed the world of medicine forever by bringing genetic testing to everyone. It has taken development of tools and testing approaches that can handle this large task. Let’s step through this evolution of genomic capability, starting with genetic testing.

At its core, genetic testing focuses on the following:

1. Genes: Mapping the Blueprint of Life

Genetic testing doesn't just look at individual elements; it dives into our genetic blueprints, identifying variations in our genes that might lead to or increase the risk of specific conditions. Whether zooming in on a single DNA building block or scanning the entirety of a person's DNA (their entire genome), genetic testing opens a window to understanding what makes us unique.

2. Chromosomes: Finding the Big Picture

Chromosomal genetic tests go beyond individual genes, analyzing whole chromosomes or extended lengths of DNA. These investigations reveal significant genetic changes, like an extra copy of a chromosome, which could lead to specific genetic conditions.

3. Proteins: The Building Blocks of Life

But genetics isn't just about DNA; it's also about the proteins and enzymes that our genes create. By studying the amount or activity level of these vital substances, genetic tests can uncover changes that may result in genetic disorders.

Your Personal Journey Through Genetic Exploration

The decision to explore genetic testing for your child is an incredibly personal and multifaceted one. While the benefits of understanding genetic traits and potential risks are invaluable, there can be limitations and emotional aspects to consider.

A Tool for Today's Parents

Historically, genetic testing was primarily used to diagnose or rule out suspected conditions when symptoms were present. Today, it has evolved into a proactive tool for parents like you, who want to understand and safeguard your child's future health.

With FORESITE 360, you're not just peeking into your child's genetic code; you're embracing an opportunity to protect, nurture, and celebrate what makes them uniquely themselves. Welcome to a world of possibilities and empowerment. Your journey begins here.

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History of Genetics:

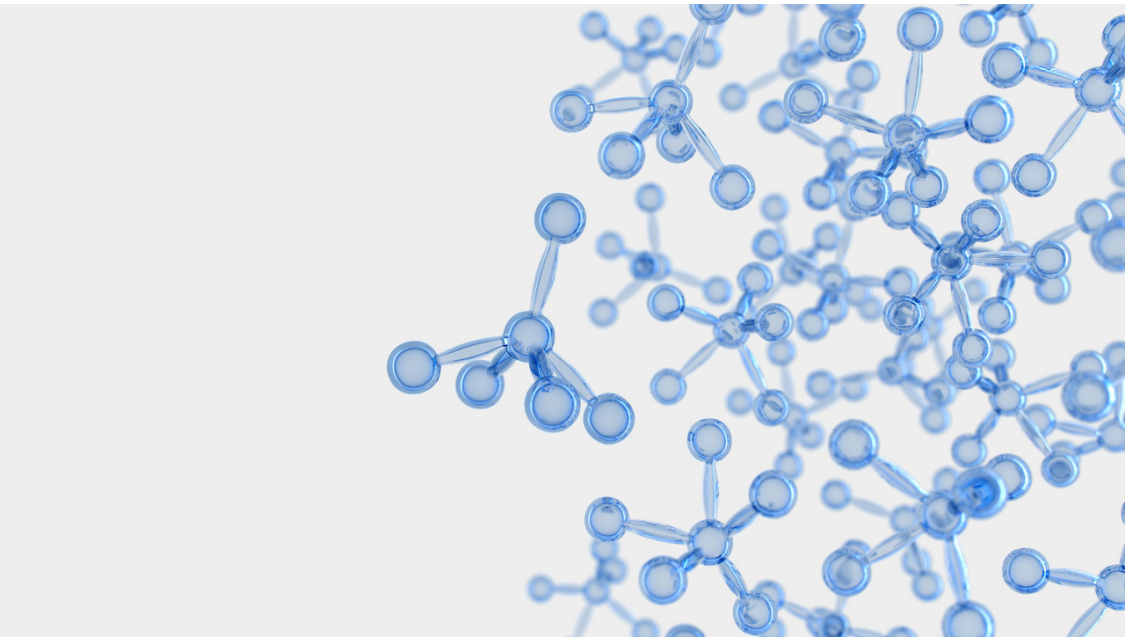
Genetics may seem like a futuristic concept, but it’s actually been studied for many years. Genetics, in essence, is the exploration of heredity—the genes and the intriguing factors that surround them. Let's embark on the history of genetics to appreciate how far we've come.

The Pioneer:

The genetics story truly took flight in the mid-19th century with Gregor Mendel, often dubbed the father of modern genetics. Before Mendel's groundbreaking pea plant experiments, genetics was mostly a game of theory. Mendel changed that, turning genetics into a vibrant field brimming with experiments and discoveries.

A Century of Revolution:

Fast forward to the 20th century, an era that witnessed remarkable strides in genetics. The first half was foundational, setting the stage for the molecular revelations of the latter half. Our understanding of the intricate dance between genes, proteins, and cells deepened, moving from mere observation to understanding the molecular mechanics at play.



Into the Modern Era:

The closing decades of the 20th century were monumental. The Human Genome Project, mapping the vast universe of our DNA, was a monumental feat, exponentially expanding our understanding of genetic factors and their roles in health, disease, and the human experience.

And the results? Astounding! Today, we've identified over 1800 genes where a mutation has a well known clinical impact, have access to more than 2000 genetic tests, and have witnessed the launch of over 350 biotech products that harness this knowledge for the betterment of human health. What's more, the global research community is identifying about 100-200 gene-disease validations every year.

The Dawn of a New Era:

And now, we're amidst a technological renaissance. With next-generation sequencing, we're decoding DNA at a pace once deemed unthinkable, redefining boundaries and setting new standards for biological research. What used to cost $100,000,000 to sequence one genome about 20 years ago, and later became available only to the extra fortunate "1%", is now accessible to everyone. Many believe whole genome sequencing to be the future of precision medicine since it's finally affordable for most families.

So, as you consider the genetic mysteries genetic testing can unveil for your child, remember: you're not just accessing a test, but tapping into centuries of discovery, innovation, and hope.

The Potential of Genetic Testing

By learning about our DNA, we can identify subtle (and sometimes not-so-subtle) changes, known scientifically as mutations or variants.

What are genetic tests looking for?

When a genome is sequenced, all 3.2 billion letters of a person's DNA expressing over 20,000 genes are examined. Anywhere an incomplete or incorrect expression of a gene is detected, that will be flagged as a "variant" in their genome. Variants can be both significant or insignificant. Some variants are not indicative of health issues. However, the variants that indicate a disposition to disease will be stored and communicated. Genomic technology surfaces findings every year that can be brought to any patient with a fully-sequenced genome.

So, what can genetic testing show us?

- **Pinpointing Conditions:** Genetic testing can give a name to a mystery. When you suspect a condition based on symptoms, genetic testing can confirm that crucial confirmation. Conversely, failing to perform the right tests can lead to years of suffering.
- **Peeking into the Future:** Some of us have silent, invisible markers that could lead to health challenges in the future, such as cancer. Genetic tests can unveil these markers, offering us the chance to prepare, prevent, or even alter that potential future.
- **Simplicity in Process:** You might think that accessing this wealth of knowledge requires a complicated procedure. In reality, it's as simple as a cheek swab sample. And within a few weeks, a world of information is at your family and physician's fingertips.
- **Guidance on the Journey:** While the idea of genetic testing is fascinating, it's essential to navigate the process wisely. This is where genetic counseling shines. A counselor ensures that you can comprehend the results.

What is DNA and Whole Genome Sequencing:

Think of DNA as the intricate blueprint of life. It's the inherited script present in humans and nearly every living creature. Picture it this way: almost every single cell in our bodies carries the same set of instructions, encoded in DNA. While the majority of our DNA resides in the cell's control center, the nucleus, some can also be found in the cell's energy factories, the mitochondria.

Now imagine these instructions as a coded language composed of four letters: A (adenine), G (guanine), C (cytosine), and T (thymine). These letters string together in a sequence that spans approximately 3 billion characters.

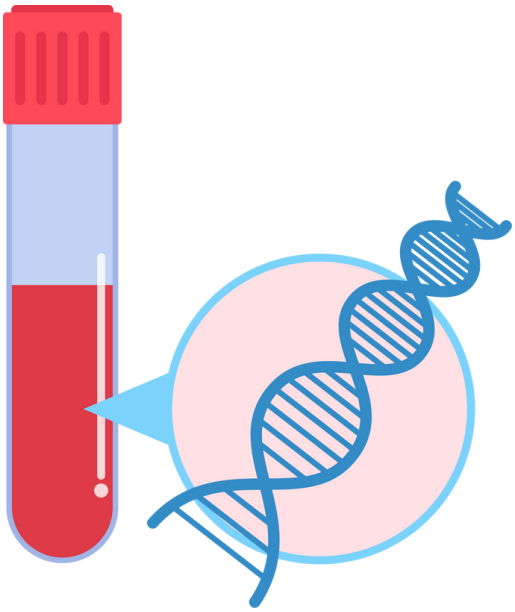
It's impressive to consider that over 99% of this sequence is identical in all humans. The specific order of these letters, much like words in a book, tells the unique story of every organism, guiding its growth and functioning. These letters also come together in pairs - A with T and C with G. They, along with sugar and phosphate molecules, form the building blocks, or nucleotides, of DNA.

One of DNA's miraculous features is its ability to self-copy. It's like having a built-in photocopier that ensures that whenever a cell divides, its successor receives the exact same instruction manual.

Now, enter the realm of whole genome sequencing (WGS). Imagine having the capability to read through this entire 3 billion-letter-long book of life. WGS does precisely that, offering a panoramic view of the entire genome. This powerful technique has revolutionized our understanding of inherited diseases, and it is now available to anyone.

Benefits of Whole Genome Sequencing:

- Offers a detailed and complete, letter-by-letter reading of the genome.
- Captures both big-picture changes and subtle nuances, which targeted methods might overlook.
- Pinpoints potential key changes for deeper exploration into how genes function and regulate.
- Produces vast amounts of data swiftly, aiding in piecing together new genomic puzzles.



What types of Genetic Testing are available?

When we talk about genetic testing, we want to distinguish between clinical genetic tests and direct-to-consumer (DTC) tests. That's because we've all seen consumer tests that help understand ancestry or provide limited, non-clinical insights into DNA.

Both clinical genetic tests and DTC tests analyze DNA, but their purposes, accuracy, and regulatory compliance differ significantly.
Here's a comparison:

	Clinical Genetic Tests	DTC Tests
Purpose:	These are ordered by healthcare providers to screen, diagnose, or evaluate the risk of medical conditions. They are typically used in a medical context to guide healthcare decisions.	Often marketed as "at-home" genetic tests, these are primarily designed for personal interest, such as tracing ancestry or understanding non-medical traits like earwax type or sensitivity to cilantro. Some DTC tests do offer health insights, but they tend to provide a broader, less medically actionable view.
Accuracy and Depth:	These are designed to be highly accurate, providing detailed information about specific genes or genetic mutations. They utilize advanced techniques, ensuring results are reliable enough to guide medical decisions.	While many are reasonably accurate for their intended purpose, they typically use simpler techniques that may not capture the depth or detail of clinical tests. They might not detect all genetic variants related to a particular condition.

	Clinical Genetic Tests	DTC Tests
Regulatory Compliance:	They are strictly regulated by health authorities and must adhere to high standards for accuracy, reliability, and validity. They are performed in specialized labs that meet clinical standards.	The regulatory landscape for DTC tests is less stringent. While some tests have undergone review by health authorities, many haven't. This difference in oversight means the onus often falls on consumers to evaluate a test's credibility.
Interpretation:	Results are typically interpreted by medical professionals, including genetic counselors, ensuring that patients receive accurate, relevant information and understand the implications for their health.	Results are often presented in user-friendly formats for consumers to interpret themselves.
Intended Use:	Given the serious implications of many genetic conditions, clinical tests are intended to be utilized in conjunction with medical advice.	While entertaining and enlightening about ancestry or specific traits, they generally should not be used as a substitute for clinical advice or diagnosis.

In summary, while both clinical genetic tests and DTC tests provide insights from our DNA, it's crucial to understand their differences and use them appropriately. Clinical tests are rigorous, medically-focused, and adhere to strict standards, while DTC tests cater more to curiosity and personal exploration. Put differently – your family physician will not find a DTC test useful. A Clinical Genetic Test, however, can drive treatment with precision and certainty.

Additionally, there are different scopes of genetic testing:

Single Gene Testing: This test targets a singular gene, particularly when symptoms suggest a specific condition. If your family history points towards a known genetic mutation, this test might be your go-to. Think conditions like Duchene muscular dystrophy or sickle cell disease. One challenge with this type of testing is that it is typically done when your child is already suffering or has suffered for years.

Panel Testing: Rather than focusing on one gene, panel testing is similar to reviewing several sentences in different chapters at once. It groups genes by medical themes. If you've ever wondered about underlying reasons for certain characteristics like low muscle tone or short stature, this test provides insights. And for those concerned about cancer risks, certain panels target genes associated with specific cancers, such as breast or colorectal cancer.

Large-scale Genetic or Genomic Testing: This is the deep dive into your genetic story. There are two main types:

- **Exome Sequencing:** This zooms into all the exons or protein coding regions of genes in your DNA. The exome is ~2% of the genomes.
- **Whole Genome Sequencing:** Think of this as reading the entire book, cover to cover. It evaluates all of a person's DNA, leaving no stone unturned. The 98% not included in exome sequencing was considered "junk DNA" or noncoding DNA for years but as sequencing technologies have improved and researchers have studied more genomes it is clear there is more function in noncoding DNA than previously thought. The old adage that one person's trash is another person's treasure is proving true with whole genome sequencing. What's really unique about Whole Genome Sequencing is that you can do the test once, and unlock a lifetime of insights. As new discoveries are made, your child's whole genome can be re-analyzed without the need for testing again. This way, you'll know as much as you can, as soon as you can, about treatable conditions.

How are Genetic Screening Tests Different from Diagnostic Genetic Tests:

Within genetics, there are two primary pathways: screening tests and diagnostic tests. Both serve crucial roles, but each offers different insights and serves unique purposes.

Genetic Screening Tests

Imagine screening tests as horizon-scanning binoculars. They give a broader view of what is ahead. Used primarily in those without noticeable symptoms, these tests gauge an individual's likelihood of developing a particular genetic condition compared to the general population. It becomes a compass, guiding individuals in making informed decisions about their health and future.

Positive Result: Indicates a higher-than-average risk. For monogenic conditions, this means the person is likely to have the disease. The question is when the symptoms will appear.

Negative Result: Suggests a lower-than-average risk.

Genetic Diagnostic Tests

As the name suggests, these tests provide specific details to confirm a diagnosis. Used for individuals already showing signs and symptoms of a genetic condition, these tests can either confirm suspicions or lay them to rest. By the time most individuals have a genetic diagnostic test, they have been through months or years of other diagnostic tests with no answers. Not to mention in the case of a child and family – each has likely undergone years of unnecessary suffering.

What is Newborn Screening vs. Whole Genome Screening:

When your baby is born, a world of possibilities opens up, each brimming with hope and promise. But before setting out on this new adventure, there's a checkpoint every newborn must pass: newborn screening. This standard procedure is just the beginning of learning everything you can about your new little one. Newborn screening is considered widely to be among the most successful public health initiatives in history. Many believe it will be replaced with WGS.

Newborn Screening

Newborn screening is a proactive public health initiative that checks newborns for very limited specific health conditions. Catching these potential problems early means treatments can commence immediately, preventing harmful effects from setting in. While the intent is consistent, the range of conditions tested varies across states, with some testing for as few as 10 conditions, and others casting a wider net with up to 60 conditions.

VS

Whole Genome Screening

Whole Genome Screening harnesses the power of Whole Genome Sequencing (WGS) to meticulously review the entire genome. The aim? To gauge an individual's vulnerability to certain health conditions. The most valuable time to conduct WGS is after birth or within the first 5-years. This is because the younger the child, the more likely to identify treatable childhood conditions.

As our understanding of genetics grows and new health-related discoveries come to light, the screening parameters can evolve and expand, ensuring that the test remains relevant and current. In general, up to 100 to 200 new genetic disease markers are discovered every year. This is why reanalyzing a genome on a consistent basis is pivotal in continuing to learn about your health as science progresses. When new discoveries are achieved, your child's genome does not need to be tested again… It is simply reanalyzed.

How is Genomic Data Reported?

Venturing into the world of genetics is similar to opening a book written in a complex language. It's fascinating, essential, yet often challenging to understand. A consumer product, put simply, is simply incapable of delivering reliable information to you, your family, or your physician.

Interpreting the Results

When diving into genetic test results, one cannot take them at face value. The interpretations hinge upon an individual's medical and family history. It's not just about the DNA; it's about the entire story the DNA is a part of. This is where genetic counselors come into play- the translators for the information.

Positive Test Results

Assuming you and your family elected to perform WGS with clinical diagnostics, a positive result indicates that the lab pinpointed a change in the DNA or protein being studied.

This might:

- Validate a diagnosis.
- Show carrier status for a genetic variation.
- Highlight an elevated risk for diseases, like cancer.
- Recommend more comprehensive tests.

Negative Test Results

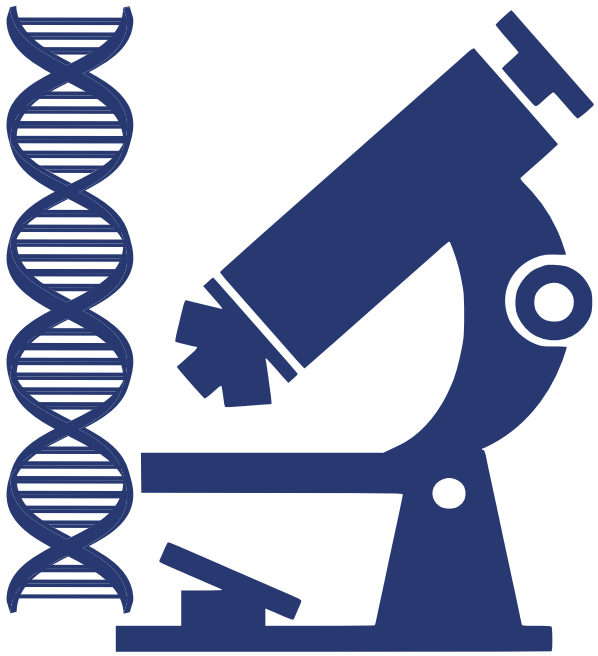
A negative result suggests no detectable change in the gene, chromosome, or protein in focus.

This could mean:

- The person isn't affected by the examined disorder.
- They aren't carriers of the analyzed genetic variation.
- They don't have an elevated risk for the particular disease.

The Gray Area

Sometimes, the test results might reside in a gray area, neither confirming nor denying any specific information. Such results are termed uninformative, inconclusive, or ambiguous. This might arise from common DNA variations (polymorphisms) that don't impact health. Another challenge arises from the variant of uncertain significance (VUS). Determining whether a VUS causes disease or is just a benign variant can be like navigating uncharted waters.



What is Genetic Counseling?

Genetic counseling is a specialized form of counseling that focuses on assisting individuals and families to understand and adapt to the implications of genetic test results.

This usually involves:

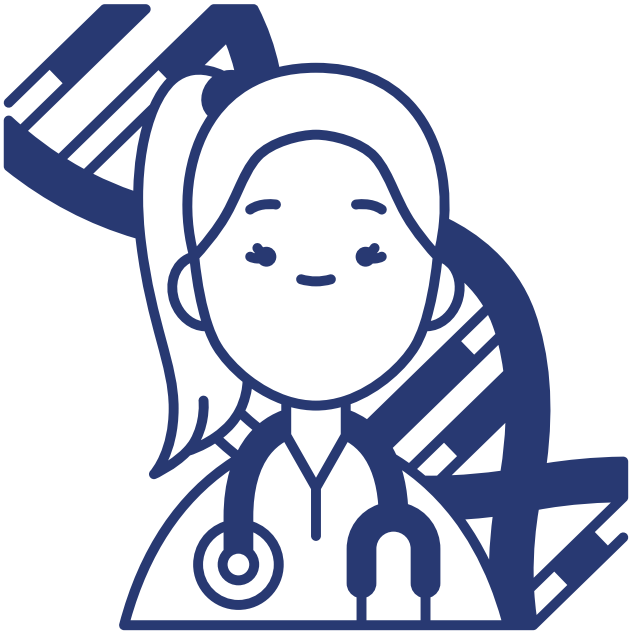
Interpretation of Medical Histories: A genetic counselor reviews your personal and familial health records to look at potential genetic risks.

Pre-Testing: The counselor helps a family understand what to expect with whole genome sequencing before the testing occurs.

Education: The counselor educates about inheritance, genetic testing, disease management, prevention, and available resources.

Risk Assessment: Based on your history, the counselor estimates the risk of occurrence or recurrence of a genetic condition.

Result Interpretation: Once tested, they help interpret the results and what it means for the individual and family.



Support: Genetics can bring forth emotional and complex family dynamics. Counselors provide supportive counseling and can point individuals and families to additional resources or specialists.

Empowerment: Armed with knowledge, individuals can make informed decisions about their health and lifestyles. For example, individuals at high risk for certain cancers might opt for more frequent screenings or preventive surgeries.

Referral: If necessary, genetic counselors can refer patients to other healthcare professionals or specialists for further advice or treatment.

Research Updates: With the rapid evolution of genetics, new information emerges regularly. Genetic counselors stay updated and can relay new findings or treatment options that may be relevant.

Genetic counseling is a bridge between complex genetic information and an individual's understanding of how this pertains to their health and life decisions. This service proves invaluable, especially when navigating the multifaceted world of genetics, ensuring that individuals and families make informed medical and personal choices. Genetic counselors stand as a beacon of guidance and support.

What is Pharmacogenetics?

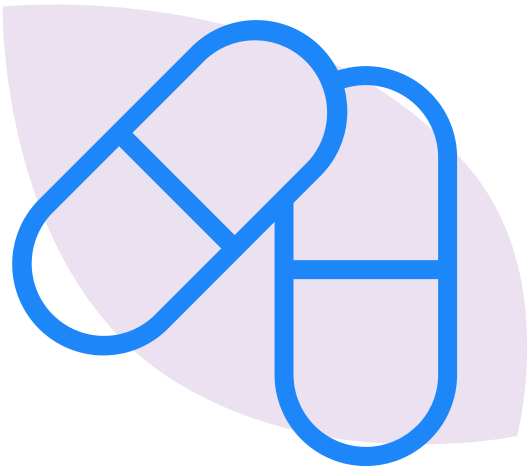
Pharmacogenetics, also known as personalized drug therapy, is a fascinating field that dives into the genetic determinants of drug response. **The central idea behind it is that our genes can influence how we react to medications.** Without this knowledge, your child might take medications that are unfit based purely on your child's genetic makeup.

At its core, pharmacogenetics seeks to move away from the "one size fits all" approach to medicine. It recognizes that individuals can metabolize or respond to drugs differently based on their genetic makeup.

Small differences in our DNA can significantly impact how a drug is processed or how it works in the body. Some genetic variants can speed up the metabolism of certain medications, rendering them less effective, while others might slow it down, leading to increased drug levels and potential toxicity.

Predicting Drug Response

By examining an individual's genetic makeup, healthcare providers might be better equipped to predict potential drug reactions, both beneficial and adverse. This information can be crucial when deciding drug types and dosages. No one wants to have to try multiple medications to find one that works, especially with children.



Examples of Pharmacogenetic Applications:

- **Codeine:** Some children possess variants of the CYP2D6 gene that causes them to metabolize codeine rapidly into its active metabolite, morphine. This can result in potentially toxic levels of morphine, causing respiratory depression. Knowing a child's CYP2D6 status can inform the decision of whether to prescribe codeine or opt for an alternative analgesic.
- **SSRIs:** Selective serotonin reuptake inhibitors (SSRIs) are sometimes prescribed to children and adolescents for depression and anxiety. Variations in genes like CYP2D6 and CYP2C19 can affect the metabolism of these drugs, which may necessitate dose adjustments to avoid side effects or to ensure therapeutic efficacy.
- **Atomoxetine:** Atomoxetine is used for attention-deficit/hyperactivity disorder (ADHD) treatment. Children who are poor metabolizers due to CYP2D6 variants might experience increased plasma concentrations, which can lead to more side effects. Genetic testing can assist in dose adjustments.

- **Clopidogrel:** Some individuals possess a variant of a gene that makes them poor metabolizers of this drug, which is used to prevent heart attacks. Such individuals might not receive the full protective benefits of the medication.
- **Warfarin:** This is an anticoagulant (blood thinner) whose effective dose can vary widely among individuals. Some people have genetic variations that make them more sensitive to the drug, requiring a lower dose, while others might be resistant and need a higher dose.
- **Malignant Hyperthermia:** People with certain genetic variants are at risk of developing this life-threatening reaction when exposed to specific anesthetic agents.
- **Stevens-Johnson Syndrome/Toxic Epidermal Necrolysis:** Some individuals are at an increased risk of developing these severe skin reactions when taking certain medications, and this risk can be influenced by genetics.

The eventual goal of pharmacogenetics is to enable truly personalized medicine, where treatments, including drug choices and dosages, are tailored to fit the individual's unique genetic makeup. This can lead to more effective, safer therapeutic interventions.

How is Whole Genome Sequencing data stored?

Genetic data presents unique challenges in the territory of storage, analysis, and ethics. Let's delve a bit deeper into some of the solutions and considerations being made to address these challenges:

Distributed Storage Systems: Given the volume of data, many genomic data repositories use distributed storage systems. These systems can scale to accommodate vast amounts of data and distribute the data across multiple servers or locations, which can also aid in redundancy and recovery in the event of data loss.

Tiered Storage: Not all genomic data needs to be accessed frequently. Tiered storage solutions allow for less frequently accessed data to be moved to cheaper, slower storage solutions, while frequently accessed data can be stored on faster, albeit more expensive, storage mediums.

Encryption and Differential Privacy: To protect the privacy of individuals whose genomic data is being stored, encryption is a must. Moreover, differential privacy techniques can be used to ensure that data used in research doesn't leak personal information.

Data Lakes: These are storage repositories that can store vast amounts of raw data in their native format until it's needed. Data lakes can handle the size and provide robust analytics capabilities when needed.

Collaborative Efforts: Global initiatives, such as the Global Alliance for Genomics and Health (GA4GH), aim to foster collaboration among scientists and create standardized protocols for data storage, sharing, and analysis.

Policy and Regulation: National and international bodies are continually working to establish and update policies that govern the collection, storage, and sharing of genomic data. These guidelines are crucial to address the ethical and privacy concerns associated with such data.

In the end, the storage of genomic data isn't just about having enough space; it's about ensuring that the data is secure, accessible, reliable, and ethically handled.

Leveraging a Global Research Community:

The global rise in genomic initiatives is a testament to the transformative power of genetics in understanding disease and improving human health. The rapid growth and proliferation of this technology offers a revolutionary opportunity with the ultimate goal of personalized, precision medicine.

Here are some benefits of this growth:

Personalized Treatment: As more genomic data becomes available, healthcare providers will be better equipped to offer personalized treatments. By understanding the genetic makeup of an individual, doctors can prescribe treatments and drugs that are more likely to be effective for that individual, reducing the trial-and-error approach that is sometimes taken with medications.

Early Intervention: With insights into genetic variants that increase the risk for diseases, preventive measures can be taken early on. This could drastically reduce the burden of diseases that are better managed or prevented altogether with early intervention. ‘The earlier the better’ comes into play with children for this exact reason.

Research and Development: A richer genomic database will expedite drug discovery and development. Understanding genetic markers and their association with diseases can streamline the clinical trial process, leading to faster approvals and more targeted drugs.

Are children eligible for Whole Genome Screening?

Yes! Whole genome sequencing (WGS) for newborns, infants, and children is a promising stride forward in personalized medicine. By unveiling the unique genetic blueprint of a child, we have the opportunity to prevent, manage, or even treat potential health issues long before they manifest. Most notably, WGS is finally affordable for most families as a self-pay/elective procedure. WGS was once reserved for the proverbial “1%” of the population, but is finally accessible to parents who want to know more about their child’s health risks, know earlier to avoid unnecessary suffering and costs, and know what to do about it.

Here are some of the transformative benefits of this cutting-edge technology:

Early Detection for Proactive Care: Recognizing genetic predispositions early allows for timely interventions, reducing the risk of complications or progression of conditions. The preventive approach could be a lifesaver for many children.

Avoiding Painful Diagnoses: One of the major advantages of early genetic screening is eliminating the need for children to undergo numerous, often painful, diagnostic procedures later in life. Not only can this spare children unnecessary discomfort, but it can also save families from the emotional and financial burdens of undiagnosed conditions.

Tailored Health Plans: Understanding a child's genetic makeup can guide pediatricians in developing a health roadmap that's uniquely tailored for each child, ensuring they receive the best possible care tailored to their needs. Notably, these tailored health plans are generally not available if your family selects a “consumer” version of genetic screening. It is only available with WGS and associated clinical reporting.

Cost Savings: While there's an upfront cost to WGS, the potential savings are significant. By identifying risks early on, families can avoid the high costs associated with prolonged diagnostic processes and potential hospital stays later in life. This cost savings is distinct from the potential suffering resulting from delayed diagnosis or identification of treatable conditions.

Empowering Families with Knowledge: Knowledge is power. Being aware of potential health risks enables families to make informed decisions and take proactive steps for their child's health. Some experts believe that WGS will soon become a central tool in creating personalized, predictive, data-rich medicine tailored to an individual’s needs.

While these benefits are transformative, it's also vital to ensure that all procedures uphold the highest ethical standards and that families are informed about the potential findings and their implications. As we continue to deepen our understanding of the human genome, refining our approach to genetic testing will be paramount, ensuring we maximize the benefits while always prioritizing the well-being and rights of the child.

Example Genetic Conditions Identified by Whole Genome Screening:

There's a distinct difference between genetic predisposition and genetic diseases. The former is a mere indicator, suggesting that under certain circumstances, one is at an elevated risk of a disease.

Genetic tests can highlight predispositions for certain genetic diseases. Recognizing these can motivate individuals to adopt preventive strategies to mitigate disease onset.

Many diseases, including prevalent ones in the U.S., have recognized or speculated genetic predispositions, such as:

- Diabetes
- Heart Disease
- Cancers
- Asthma
- Obesity
- Addiction tendencies
- Autism
- Strokes
- Various mental health conditions, like schizophrenia and bipolar disorder
- Celiac disease
- Fibromyalgia
- Myalgic encephalomyelitis or chronic fatigue syndrome
- Irritable bowel syndrome
- Autoimmune disorders, like lupus, rheumatoid arthritis, and multiple sclerosis

In contrast, possessing a genetic disease's gene(s) confirms the disease's presence or eventual onset. Some genetic diseases necessitate genes from a single parent, while others demand contributions from both.

Examples of genetic diseases comprise:

- Cystic fibrosis
- Fragile X syndrome
- Hemochromatosis
- Huntington’s ailment
- Marfan syndrome
- Phenylketonuria
- Polycystic kidney ailment
- Tay-Sachs disease

Given the hereditary nature of these diseases, potential parents with family histories of these conditions increasingly seek genetic testing to gauge the risk of transmission to offspring.

Considering Genetic Testing for Your Child?

Fore Genomics, a pioneer in clinical genetics, introduces FORESITE 360. Tailored for healthy newborns, infants, and children, this state-of-the-art service requires just a cheek swab to equip both doctors and parents with key genetic data. What's more, it only reports on treatable, childhood conditions. At this time, over 300 treatable conditions, and over 40 medication interactions, are tested by FORESITE 360. In contrast to newborn screening, informed parents who are tech-forward have vastly more information and are equipped to leverage the global research community.

By assessing a child's DNA, it offers foresight into possible future health concerns. Relying on the proven expertise of CLIA-certified and CAP-sanctioned laboratories in the U.S., FORESITE 360 promises a comprehensive genetic overview, illuminating lifelong health trajectories.

An added benefit? The service also ensures yearly DNA reanalysis, empowering healthcare professionals to proactively address any identified health issues. As more information is unlocked from the global research community, those details are only unlocked for those families who have elected to perform WGS with re-analysis capabilities.

Understanding the Expense of Genetic Testing

Opting for clinical-grade whole genome sequencing is finally affordable for most families through FORESITE 360 as an elective, self-pay product. The company even offers financing.

Here's what this includes: a parent or guardian can effortlessly place an online order, which is subsequently confirmed by a medical professional who prescribes a screening test. This ensures the report's usability by your child’s primary health physician.

The process is child-friendly; using a simple, at-home saliva collection kit, ensuring no distress for young ones. Post-collection, samples undergo sequencing in trusted CLIA/CAP-accredited US labs.

Parents and healthcare providers receive a detailed clinical report and actionable insights, shedding light on over 300 health conditions. Pharmacogenetic data, crucial for early medical interventions, is also provided.

Furthermore, families can benefit from a personalized discussion with a domestic genetic counselor, who will interpret the results.

A notable feature? The package includes annual DNA reanalysis, underscoring the growing clinical significance of Whole Genome Sequencing.

Final Thoughts

In today's era, parents are increasingly looking to leverage cutting-edge technologies to ensure the best care for their children. With FORESITE 360's genetic sequencing, you're accessing the pinnacle of health science to understand your child's genetic makeup.

Recognizing specific genetic markers can pave the way for tailored healthcare plans throughout their life. FORESITE 360 stands as a testament to forward-focused, scientifically-backed healthcare solutions that every parent seeks. Discuss with your child's pediatrician to discover how this groundbreaking tool can sculpt a healthier future for your little one.

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FAQ:

How is the DNA collected?

FORESITE 360 uses a simple, non-invasive saliva swab to collect the sample of DNA used in the test.

How can this help babies and their parents?

FORESITE 360 will identify many genetically linked health risks even before a child is symptomatic and provide information on how the child will respond to common medications. This gives parents and healthcare providers the ability to take proactive steps to mitigate or possibly prevent conditions.

How is this different than DNA testing services like 23andMe?

Unlike other genomics products, FORESITE 360 is a comprehensive, clinical genetic screen that analyzes 100% of a child's DNA to identify genetic conditions and help create actionable treatment plans. FORESITE 360 also includes yearly re-analysis of their genomic profile using any new data on genetic conditions or potential new treatments. Access to a genetic counselor is also provided to translate the results.

Is this only for people who have a family history of disease?

No, anyone interested in learning how genetics may impact their child's health can use FORESITE 360, whether or not they have a genetic condition in their family.

Will parents be able to understand the analysis?

We make sure the results of a child's genomic analysis are communicated clearly to parents by setting up a one-on-one session with a specialized FORESITE 360 genetic counselor to discuss the results and any questions parents might have.

Why is this approach the best for this kind of analysis?

By sequencing all 22,000 genes and screening approximately 300 markers of genetically linked conditions and re-analyzing yearly, FORESITE 360 provides actionable insights about a child's genetics for physicians and parents to use when creating treatment plans. The yearly updates incorporate any new genomic insights and give parents the most information possible about their child's health.

What types of conditions and pharmaco-genetic interactions does this analysis screen for?

FORESITE 360 compares genomic profiles against genetically linked health risks such as childhood metabolic disorders, immunodeficiencies, and cancers, and it provides individualized information about a child's response to medications like antibiotics, immunosuppressants, and anticonvulsants.

How long does the analysis take?

Once the saliva sample is collected and sent for processing, it takes about 6 to 8 weeks for parents to receive their child's results and clinical action report.

What happens if parents have questions later?

When parents sign up for FORESITE 360, they have access to a personal genetic counselor, who is always available to answer any questions.

Who are these genetic counselors?

Genetic counselors are certified experts who specialize in assessing risk for genetic disease. They can guide parents through the FORESITE 360 process, explain the results of the initial report, as well as be available for any questions parents may have.

- [ ] **What happens if parents decide to wait a few years before using FORESITE 360?**

  Anyone at any age can take a FORESITE 360 test, though it is recommended to conduct genomic testing as a newborn, as genetic conditions are often easier to manage when preparation starts before symptoms appear. When used to screen newborns, FORESITE 360 can help you make proactive healthcare decisions, potentially increasing a baby's quality of life and minimizing symptoms.

- [ ] **Is it worth the money?**

  An investment in FORESITE 360 for a child today can unlock health insights for a lifetime and gives parents access to a genetic counselor who can answer any questions they might have throughout the testing and reporting process.

- [ ] **How do parents pay for this?**

  Payment plans can be customized to a family's budget, with plans ranging from a one-time payment to affordable, monthly payments.

  Will insurance cover FORESITE 360?

  Currently, FORESITE 360 is not covered by insurance.

- [ ] **Will a condition that might develop in the future impact a patient's insurability?**

  No, a federal law called the Genetic Information Nondisclosure Act (GINA) protects individuals against discrimination based on their genetic information in health coverage and in employment. Additionally, FORESITE 360 will never share any information with insurance companies, in accordance with HIPAA guidelines.

- How do parents let their physician see their baby's FORESITE 360 report?

  Parents can designate a physician of their choosing to receive their child's FORESITE 360 report. No FORESITE 360 information or information about a child will be shared with anyone other than parents or legal guardians, their designated physician, and the personal genetic counselor.

- Will this information be shared or sold?

  No. Fore Genomics will never sell a child's data, or store data outside of the United States.

- Does HIPAA apply to FORESITE 360?

  Yes, all information is 100% secure and private at every step of the process, and data storage is HIPAA-compliant.

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