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Is personalized care really here?

Why advancements in precision medicine are making personalized care a reality even sooner than expected

A Definitive Healthcare report

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Introduction

The relationship between consumer interest and technological development has always been a question of chickens and eggs. As new technologies are developed, the surrounding media cycle generates consumer interest. And yet, simultaneously, organizations spend massive sums tracking that interest to focus their development efforts on the most marketable tech.

Precision medicine, a healthcare model centered on highly personalized care (that is, personalized down to the patient's genetic makeup), seems to sit perfectly at that uncomfortable intersection. On one hand, most people probably wouldn't even know to ask their doctor for a genomic sequencing test to identify a potential tumor. On the other hand, millions of consumers are eagerly mailing their saliva to Silicon Valley in hopes of learning about their health and ancestry from companies using the same technology.

Trends within — and adjacent to — precision medicine suggest that consumer preferences for more effective, convenient, and affordable care are driving rising rates of precision medicine procedures. Furthermore, the data reveal the ways that consumers are bucking norms and claiming greater agency over their lives: they're having children later, skipping both necessary and elective treatments when they deem costs too high, and are generally placing less trust in their providers, often turning to the internet for crowd sourced clinical guidance.

You can see consumers' willingness to pay for control in their preferences for devices and wearables that offer at-home insights into key health metrics. Or in their gravitation toward on-demand telecare appointments that fit seamlessly into their schedules. Or even in the rise of retail clinics that make clinical tests, checkups or vaccinations just another item on the shopping list.

In this report, we'll use healthcare commercial intelligence from Definitive Healthcare to tease out some emerging trends in precision medicine procedures that should be of special interest to life science developers and the providers who use their treatments:

- **Multi-gene sequencing procedures** offer improved accuracy and value, potentially replacing single-target molecular pathology procedures for disease diagnosis and treatment.
- **Parents-to-be are increasingly getting screenings** for severe birth defects, even as birth rates continue to fall.
- **Exciting new methods for identifying cancers** have hit the market within the last year, and life-changing genetic tests for deadly diseases and transplant matching are getting cheaper.

Through these trends, we can understand the current state of precision medicine and its role in preventive care, predict where the field might be heading, and consider how consumers' desire for greater control and custom-tailored care impacts the field. In turn, we'll pick up insights into the real-world data produced by these care events, offering utility both to providers targeting specific patient cohorts and life science companies navigating the development and commercialization process.

What is precision medicine and why does it matter now?

Precision medicine is the practice of customizing disease prevention and treatment to a patient's unique genes, environments, and ways of living. Sometimes known as "personalized medicine," precision medicine offers patients more effective care with fewer adverse effects and better health outcomes.

You might be wondering: Isn't this just the story of modern medicine? Clinicians have always aimed to tailor their recommendations and treatments to the unique needs of their patients.

So why has the concept of precision medicine only emerged in the past couple of decades?
The answer is specificity.

Clinicians' diagnostic capabilities skyrocketed in 2003 when scientists from around the world mapped the entirety of the human genome. This discovery spurred massive advancements in pharmacogenetics, the science of developing pharmaceuticals around specific genetic markers, and enabled a previously unimaginable level of precision in genetic diagnostics.

Modern precision medicine also benefits from the unprecedented availability of real-world biomedical and social data, acquired from electronic health records, claims activities, disease registries, wearables, at-home sensors, and mobile devices.

Improving treatment outcomes

Today, practitioners of precision medicine combine artificial intelligence, machine learning, genomic and molecular testing, and real-world data to create highly personalized plans of care for patients who might otherwise be treated according to condition or disease alone. This personalized approach to care can improve patient outcomes and increase operational efficiency.

Once considered a treatment tool for only the sickest patients—usually those receiving cancer treatment—precision medicine is becoming increasingly available to patients for a variety of preventive and curative purposes.

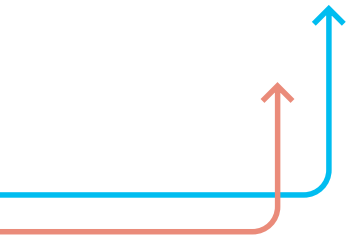
Genetic tests capable of identifying risk factors or the presence of disease can be ordered for hundreds of dollars instead of thousands, and the Food and Drug Administration (FDA) now recognizes nearly 120 pharmacogenetic associations. Even everyday consumers are gaining access to low-cost genetic testing through DNA counseling and ancestry services like 23andMe.

Driving the transition to value-based care

Some healthcare professionals see precision medicine as a stepping stone in the transition from fee-for-service to value-based care. Precision medicine's ability to proactively identify potential health risks and guide treatment development around an individual's unique genetic and social factors provides cost-savings, improved outcomes for patients, and healthier populations overall—three goals for value-based care cited by the Centers for Medicare and Medicaid Services (CMS).

Proponents say that value-based care—and a greater focus on preventive care more broadly—could help to combat two of the top concerns among American physicians and patients alike: the high cost of our healthcare and its relatively lackluster outcomes (compared to other industrialized nations, who spend about half as much as the U.S. does per patient).

A 2022 Gallup poll found that 64% of Americans are either "somewhat dissatisfied" or "very dissatisfied" with the availability of affordable healthcare. The same poll found that about 53% are at least somewhat dissatisfied with the quality of U.S. medical care overall.



Two of the top concerns among American physicians and patients alike: the high cost of our healthcare and its relatively lackluster outcomes.

A value-based model would address these concerns by rewarding health outcomes rather than procedure volumes. This would incentivize preventive care, which has long been associated with reduced costs for patients, payors, and providers. Carrier and genetic testing used in precision medicine offers particular value in preventive care plans, as they provide advanced information about health risks that patients can use to make smarter lifestyle choices and gain control over their health.

Increasing efficiencies in drug and device development

Widespread access to the real-world data associated with precision medicine could also increase efficiency in the life science development process.

Every care event, from consultation to diagnosis to procedure to discharge, produces real-world data about a patient's unique health characteristics, the provider delivering the care, and the facility or other setting where care took place. Because precision medicine deals with highly individualized molecular or genomic markers, these care events usually produce more granular data than those that rely on more traditional identifiers, like microbial cultures or blood counts.

Therapies targeting rare diseases are often especially dependent on highly specific gene interactions. The increased availability of genomic data from precision medicine care events in turn makes it easier for developers to test therapeutic molecules against a wider variety of genetic makeups—leading to more efficient and affordable procedures that produce even more data, driving the cycle further.

Gene sequencing procedure growth shows value of doing more with less

As mentioned, precision medicine offers value for both the diagnosis of existing disease and determining one's risk of future disease. Procedures capable of analyzing multiple biomarkers at once are especially valuable, allowing doctors to draw conclusions from one test where multiple single-marker tests would otherwise be necessary.

Most precision medicine procedures are designated into one of three claims categories, each with its own applications: molecular pathology, genomic sequencing, and multianalyte assays. The first major trend we'll explore in this report is the exciting growth within genomic sequencing and multianalyte assays, designations of procedures capable of multi-gene analysis.

Let's look at each of these methods in turn, see how they stack up in popularity, and learn why single-marker procedures are still leading in volume despite showing minimal growth.

Molecular pathology targets individual genes to predict risk

Molecular pathology is one of the most prominent disciplines related to precision medicine. Centered on the study and diagnosis of disease using individual molecules or genes found within the human body, molecular pathology procedures are usually performed to evaluate a patient's genetic risk factor for a specific disease. They can also be used to diagnose certain cancers and identify the mutation profile of tumors, allowing for patient-specific treatments.

Claims data show that 7,290,653 molecular pathology procedures were performed in 2019. This figure dropped to 5,172,194 procedures in 2020 (a 29.0% decrease), as COVID-19-related lockdowns and restrictions on elective procedures led to reductions in procedures across most disciplines. But molecular pathology saw a rise in 2021, with 5,500,066 procedures performed (a 6.3% increase from 2020 levels).

With a total of 17,962,913 procedures performed during the three-year period (representing over 73% of all precision medicine procedures in that period), molecular pathology is the biggest precision medicine subcategory by volume. This isn't surprising, considering the sheer variety of conditions and diseases that these procedures are capable of diagnosing.

Across all three years, six molecular pathology procedures consistently ranked the highest by volume of procedures performed. These procedures address some of the most common heritable conditions, as well as some cancers with hard-to-spot symptoms:

- → Gene analysis for cystic fibrosis [CPT code 81220]
- → Gene analysis for spinal muscular atrophy [CPT code 81329]
- → Gene analysis for hereditary hypercoagulability (5, 10-methylenetetraydrofolate reductase) [CPT code 81329]
- → Level 2 molecular pathology procedure (2-10 SNPs) [CPT code 81401]
- → Level 7 molecular pathology procedure (11-25 exons) [CPT code 81406]
- → Level 6 molecular pathology procedure (6-10 exons) [CPT code 81405]

As the chart shows, these popular procedures experienced a dip in volume from 2019 to 2020, likely due to COVID-19. Likewise, all but one procedure (81406) rose again in 2021, but only the gene analysis for spinal muscular atrophy has yet managed to meet or exceed its 2019 volume.

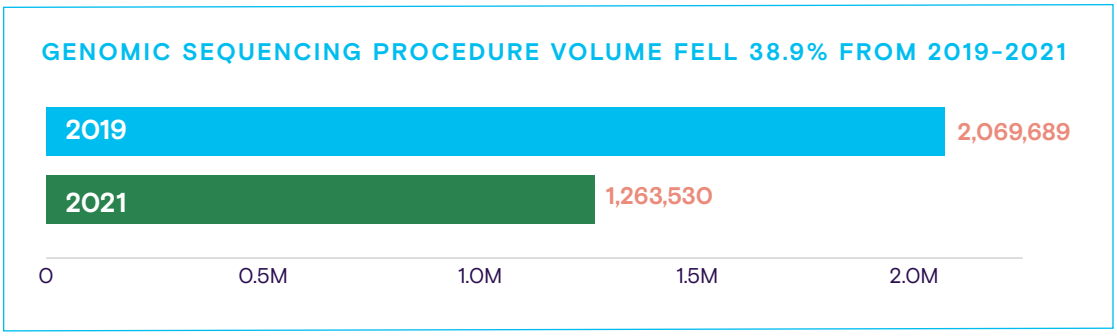
MOLECULAR PATHOLOGY PROCEDURE CODE VOLUMES ACROSS THREE YEARS

Patients who undergo these procedures may have a family history of disease, be demonstrating early symptoms of one of the targeted conditions, or may simply be curious about their risks. Each of these procedures can help a physician determine a patient's risk for a debilitating disease, diagnose the presence of the disease, or identify biomarkers that can predict treatment response or how the disease might progress.

While molecular pathology is by far the dominant procedure subcategory within precision medicine, other methods are gaining popularity thanks to their improved accuracy and value.

Genomic sequencing helps diagnose and manage inherited disease

Where molecular pathology procedures tend to focus on the presence of specific molecules or mutations within individual genes, genomic sequencing procedures analyze multiple genes or genetic regions simultaneously.



These procedures are ideal for diagnosing and managing inherited diseases that may be associated with any one of a multitude of genes, or for multiple conditions with overlapping symptoms. The broader view of these tests enables rapid analysis of multiple genomic regions with a single sample, making them considerably faster and more efficient than many single-marker tests. Doctors and patients seem to be responding to that efficiency (and subsequent cost-savings) as many of these procedures are seeing increased usage over time.

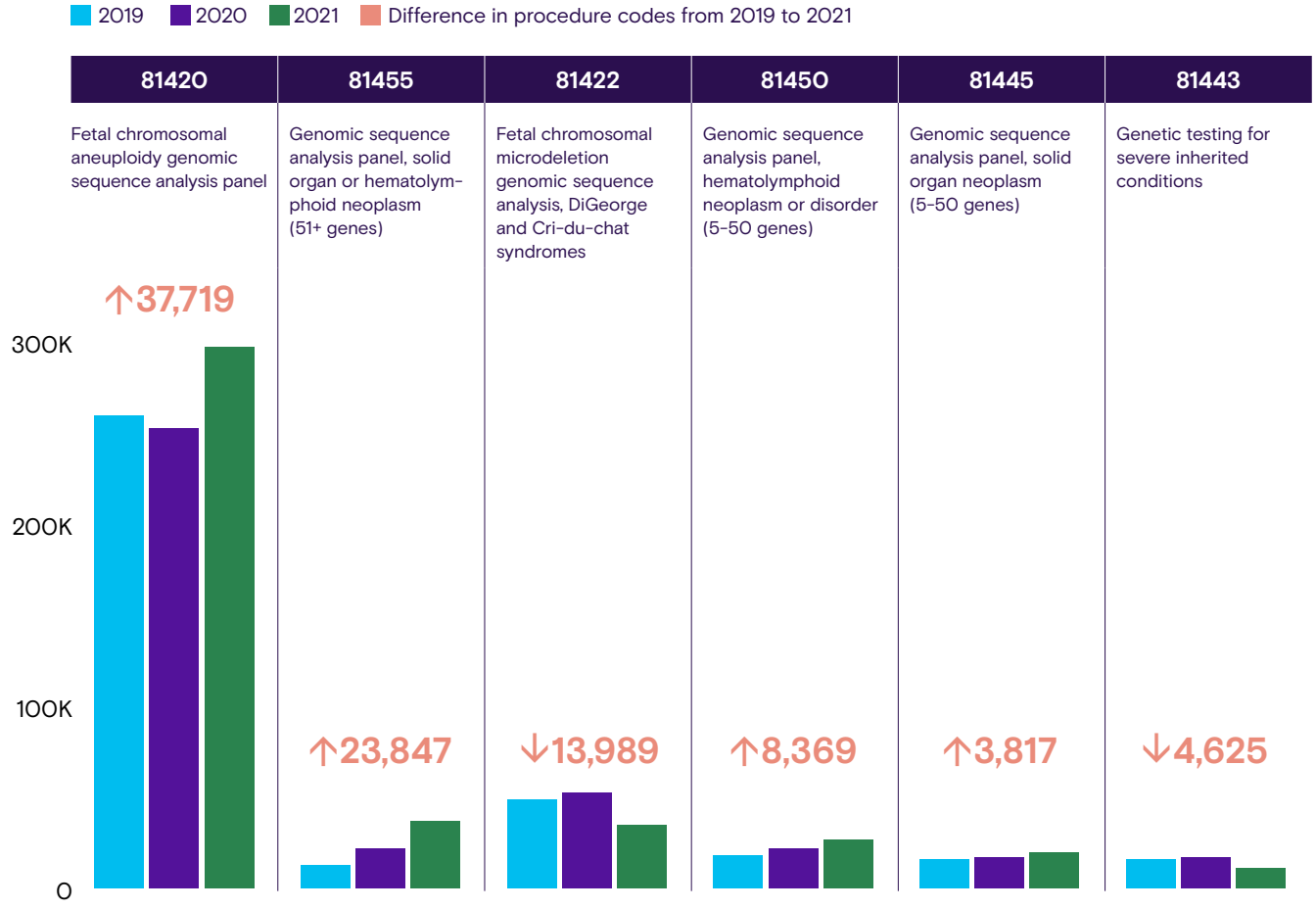
Claims data show that 2,069,689 genomic sequencing procedures were performed in 2019. This figure dropped precipitously in 2020 to 1,258,893 (a 39.1% decrease). As with the previous procedure subcategory, genomic sequencing saw a modest recovery in 2021 with 1,263,530 procedures performed—but still far below 2019 volumes.

For the past three years, the top codes in this category include procedures designed to identify common cancer tumors, as well as a maternal blood test for chromosomal disorders that has demonstrated considerable growth in volume over this period:

- → Fetal chromosomal aneuploidy genomic sequence analysis panel [CPT code 81420]
- → Genomic sequence analysis panel, solid organ or hematolymphoid neoplasm (51+ genes) [CPT code 81455]
- → Fetal chromosomal microdeletion genomic sequence analysis, DiGeorge and Cri-du-chat syndromes [CPT code 81422]
- → Genomic sequence analysis panel, hematolymphoid neoplasm or disorder (5-50 genes) [CPT code 81450]
- → Genomic sequence analysis panel, solid organ neoplasm (5-50 genes) [CPT code 81445]
- → Genetic testing for severe inherited conditions [CPT code 81443]

Two of the latter three procedures on this list look for the presence of genetic mutations associated with cancerous tumor growth, a fairly typical target within precision medicine.

GEONOMIC SEQUENCING PROCEDURE CODE VOLUMES ACROSS THREE YEARS



The hematolymphoid and solid organ neoplasm variants of this test demonstrated similar trends in volume during the examined period, rising consistently year over year from 2019 to 2021.

Interestingly, genomic sequencing procedures capable of targeting solid organ or hematolymphoid neoplasms more than doubled in volume from 2019 to 2021, indicating that providers may be finding greater utility in these tests compared to similar, but less-efficient single-biomarker assays.

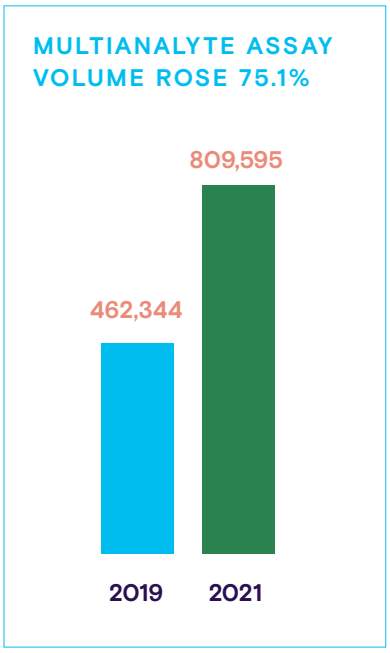
Also of note is the impressive growth of fetal chromosomal aneuploidy genomic sequencing, which aligns with another interesting trend that we'll dig into a little later.

Some genomic sequencing procedures saw impressive increases in adoption from 2019 to 2021, but the greatest growth in that period belongs to a decades-old methodology with some powerful new applications.

Super-sensitive multianalyte assays didn't slow for COVID-19

The third and final subcategory of precision medicine procedures is also showing some of the most exciting growth in the industry due to rapidly rising procedure volumes and cutting-edge technological development. Multianalyte assays with algorithmic analyses (MAAAs) combine results from two or more biochemical or molecular tests, then factor in a patient's demographic and clinical information to produce highly detailed, complex datasets. This data is then fed into an algorithm that can generate diagnostic, prognostic, or predictive information about a disease or condition specific to a single patient.

MAAAs offer far greater clinical sensitivity and specificity than single-biomarker tests, especially for the detection and treatment of ovarian and prostate cancers. In recent years, the capabilities of these tests have expanded to cover a wide variety of organ conditions, cancers, sepsis, and more.



Claims data reflect the massive expansion in capabilities that MAAAs have undergone in recent years. With 462,344 procedures recorded in 2019, MAAAs experienced an atypical rise in 2020 volume: a 25.3% increase to 579,424, bucking the trend of COVID-related dips we've seen in other categories. The next year delivered even greater growth: 809,595 MAAA procedures were performed in 2021, demonstrating the greatest volume increase during the examined period across all subcategories (a 75.1% increase from 2019 to 2021).

MAAAs have been a standard part of gynecological and obstetric care for over 30 years, and many of the highest-volume procedures within this category reflect that specialization.

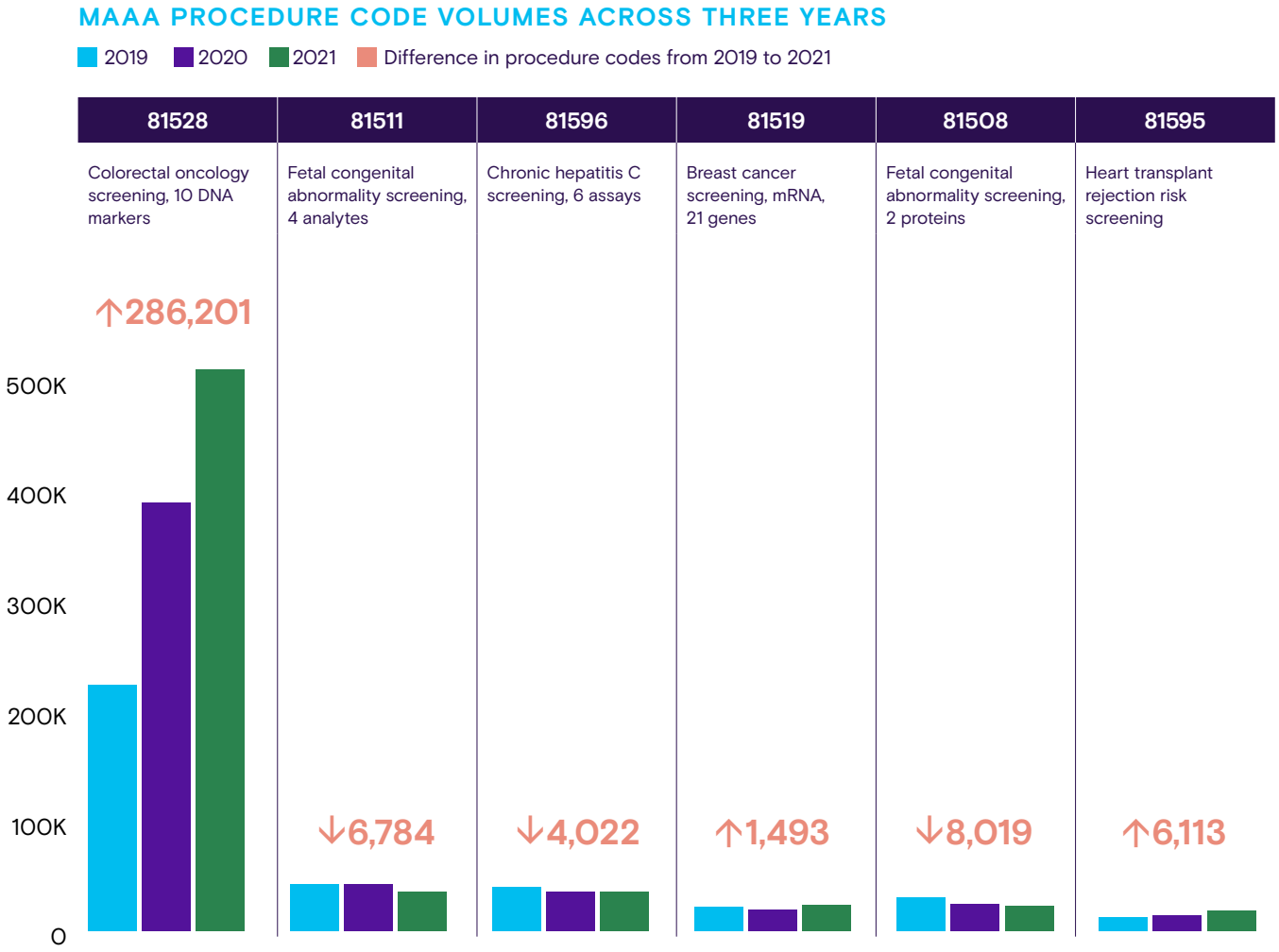
Interestingly, MAAAs related to fetal health seem to have consistently dropped in volume from 2019 to 2021. As we'll touch on in the next section, this doesn't mean that women are seeing doctors for prenatal counseling less often—instead, it likely means that molecular pathology and genomic sequencing procedures are simply more cost-effective at addressing the same concerns.

In 2019 and 2020, two procedures led among MAAAs:

- → Quantitative real-time amplification of DNA markers for colorectal cancer [CPT code 81528]
- → Four-analyte biochemical assays for fetal congenital abnormalities with risk-score algorithm [CPT code 81511]

But in 2021, two new MAAA procedure codes were introduced, generating much of the growth we see in this subcategory:

- → Quantitative real-time amplification of RNA markers for bacterial vaginosis diagnosis [CPT code 81513]
- → Quantitative real-time amplification of DNA markers for bacterial vaginosis and vaginitis [CPT code 81514]



Precision health offers reassurance for parents-to-be, especially those who are older

Our second major trend involves growth around procedures that touch on two of the most personal aspects of personalized medicine: pregnancy and parenthood.

Gene analysis is often provided to couples who are thinking about having children, or to people who are already pregnant. They can help parents-to-be determine the chances that their child will inherit certain diseases and disorders, like cystic fibrosis, venous thrombosis, or blood cancers.

Diseases caused by genetic mutations, like cystic fibrosis, may only develop if a child inherits two flawed copies of a particular gene. In these cases, a child may inherit only one copy and become a carrier. Carriers won't experience the disease themselves, but their children may still inherit the gene.

Today, many people are choosing to delay parenthood, if they have children at all. While waiting to become parents can improve a couple's financial stability, having children later in life is associated with heightened risks to both mother and child. Precision medicine procedures can help women who are considering becoming pregnant in their 30s or 40s to determine the risk of fetal complications or inherited disease.

While birth rates fell 3.7% on average in the first year of the pandemic, noninvasive prenatal testing volumes only dipped 2.6% in the same period (259,068 - 252,143 procedures).

Of course, claims data can't fully explain societal shifts, but when those shifts align with healthcare decisions, we can see them reflected in the data.

For instance, the most-performed genomic sequencing procedure from 2019 to 2021 was the fetal chromosomal aneuploidy genomic sequence analysis panel. Where many other procedures from this category dipped sharply in 2020, the fetal genomic sequence analysis—known also as noninvasive prenatal testing (NIPT)—only fell 2.6% from 2019-2020, and ultimately saw 14.5% growth, with 296,787 procedures recorded in 2021. While birthrates have been on a gradual decline in the U.S. since 2008, and births dropped by about 3.7% in the first year of the pandemic, parents and their providers are increasingly seeking preventive precision care.

Testing for trisomy is getting cheaper

Increased provider awareness and education surrounding precision medicine is likely behind some of the growth we've seen in these procedures. But lower charges may also be prompting more patients to opt for preventive measures.

NIPT procedures examine the cell-free DNA (cfDNA) present in a pregnant woman's blood, which contains a mix of her own cells and cells from her placenta. NIPT typically looks for extra or missing copies of the sex chromosomes (X and Y) and chromosomes 13, 18, and 21, mutations that are more likely to occur the later in life a woman becomes pregnant.

Most people have 46 chromosomes. The presence of an extra (47th) chromosome is known as trisomy, a condition that can result in severe developmental and intellectual disability. For instance, trisomy 13, or the presence of an extra chromosome 13, is also known as Patau syndrome, and is associated with severe abnormalities to the brain, spine, eyes, appendages, and overall musculature.

While only 5-10% of children with trisomy 13 or trisomy 18 (Edwards syndrome) live past their first year, children born with trisomy 21 (Down syndrome) have a 70-75% chance of surviving past infancy.

We noted earlier that MAAAs that test for these disorders are being ordered less frequently in recent years. It's possible that this is due to a cost reduction among similar genomic sequencing procedures.

While genomic sequencing procedures overall only saw a modest drop in average charge from 2019 to 2021 (about $60 per procedure on average), charges for fetal chromosomal aneuploidy genomic sequencing procedures dropped by about $300 within the same period.

Incidentally, genomic sequencing procedures capable of targeting solid organ or hematolymphoid neoplasms saw a similar reduction in average charges, perhaps reflecting the previously noted increase in procedure volume.

SHOWING AVERAGE GENOMIC SEQUENCING PROCEDURE COSTS ACROSS THREE YEARS

Would-be parents want to understand and reduce their risks

Genetic testing for these disorders is an increasingly routine part of procreative and prenatal care. In fact, the most common molecular pathology diagnosis code in 2021 was an "encounter of female for testing for genetic disease carrier status for procreative management," accounting for 1,228,134 patient visits. Dads are covering their bases, too: The second-most common diagnosis code was an "encounter of male for testing for genetic disease carrier status for procreative management," with 691,321 patient visits.

Together, these patients account for 46.9% of the top 25 molecular pathology diagnoses by volume.

Of special note here is the "Supervision of elderly multigravida, first trimester" diagnosis code volume. This refers to a woman having a child—but not her first—after 35.

Other high-volume pregnancy-related diagnoses from this period include "encounter for other screening for genetic and chromosomal anomalies" and "encounter for supervision of other normal pregnancy, first trimester."

Looking at diagnoses related to genomic sequencing procedures, we see many of the same trends emerging in 2021: Testing for genetic disease carrier status for men and women (both procreative and nonprocreative) and testing for genetic anomalies top the list.

10 DIAGNOSIS CODES ASSOCIATED WITH MOLECULAR PATHOLOGY PROCEDURES (2021)

Diagnosis code	Diagnosis description	Total claims
Z31430	Encounter of female for testing for genetic disease carrier status for procreative management	**1,228,134**
Z31440	Encounter of male for testing for genetic disease carrier status for procreative management	**691,321**
Z3490	Encounter for supervision of normal pregnancy, unspecified, unspecified trimester	**211,831**
Z3481	Encounter for supervision of other normal pregnancy, first trimester	**185,016**
C9210	Chronic myeloid leukemia, BCR/ABL-positive, not having achieved remission	**170,937**
Z3401	Encounter for supervision of normal first pregnancy, first trimester	**157,588**
Z1379	Encounter for other screening for genetic and chromosomal anomalies	**147,779**
Z803	Family history of malignant neoplasm of breast	**140,850**
Z1371	Encounter for nonprocreative screening for genetic disease carrier status	**124,222**
Z3480	Encounter for supervision of other normal pregnancy, unspecified trimester	**97,330**

Examining the relative volumes of certain procedure codes, we can see other procedures related to fetal health have gained sudden popularity in recent years, which further reflects the gradual increase in average age of first-time mothers.

Between 2019 and 2021, the following molecular pathology procedures saw the greatest increases in volume:

- → GJB2 gene analysis for nonsyndromic hearing loss increased by 31,356 procedures [CPT code 81252]
- → Level 8 molecular pathology procedure (26-50 exons), increased by 22,231 procedures [CPT code 81407]
- → SMN1 gene analysis for spinal muscular atrophy increased by 19,235 procedures [CPT code 81329]
- → CALR gene analysis for myeloproliferative disorders increased by 11,831 procedures [CPT code 81219]
- → BCR/ABL1 translocation analysis for chronic myelogenous leukemia increased by 11,071 procedures [CPT code 81206]

The procedure that saw the greatest increase in procedure volume from 2019-2021 was GJB2 gene analysis for nonsyndromic hearing loss. This procedure assesses the gene that provides the blueprints for the the connexin 26 protein, which permits the transport of nutrients, ions, and signaling molecules between cells in the inner ear and elsewhere throughout the body.

TOP 10 DIAGNOSIS CODES ASSOCIATED WITH GENOMIC SEQUENCING PROCEDURES (2021)

Diagnosis code	Diagnosis description	Total claims
Z3481	Encounter for supervision of other normal pregnancy, first trimester	**285,632**
O09521	Supervision of elderly multigravida, first trimester	**241,237**
D819	Combined immunodeficiency, unspecified	**197,600**
Z940	Kidney transplant status	**156,064**
Z1379	Encounter for other screening for genetic and chromosomal anomalies	**146,035**
Z3401	Encounter for supervision of normal first pregnancy, first trimester	**126,315**
Z31430	Encounter of female for testing for genetic disease carrier status for procreative management	**119,216**
Z3482	Encounter for supervision of other normal pregnancy, second trimester	**102,971**
O09511	Supervision of elderly primigravida, first trimester	**97,696**
O09522	Supervision of elderly multigravida, second trimester	**70,773**

Mutations to the GJB2 gene can cause hearing loss, as the connexin 26 protein is believed to be responsible for maintaining appropriate potassium ion levels within the inner ear and supporting normal maturation of the cochlea.

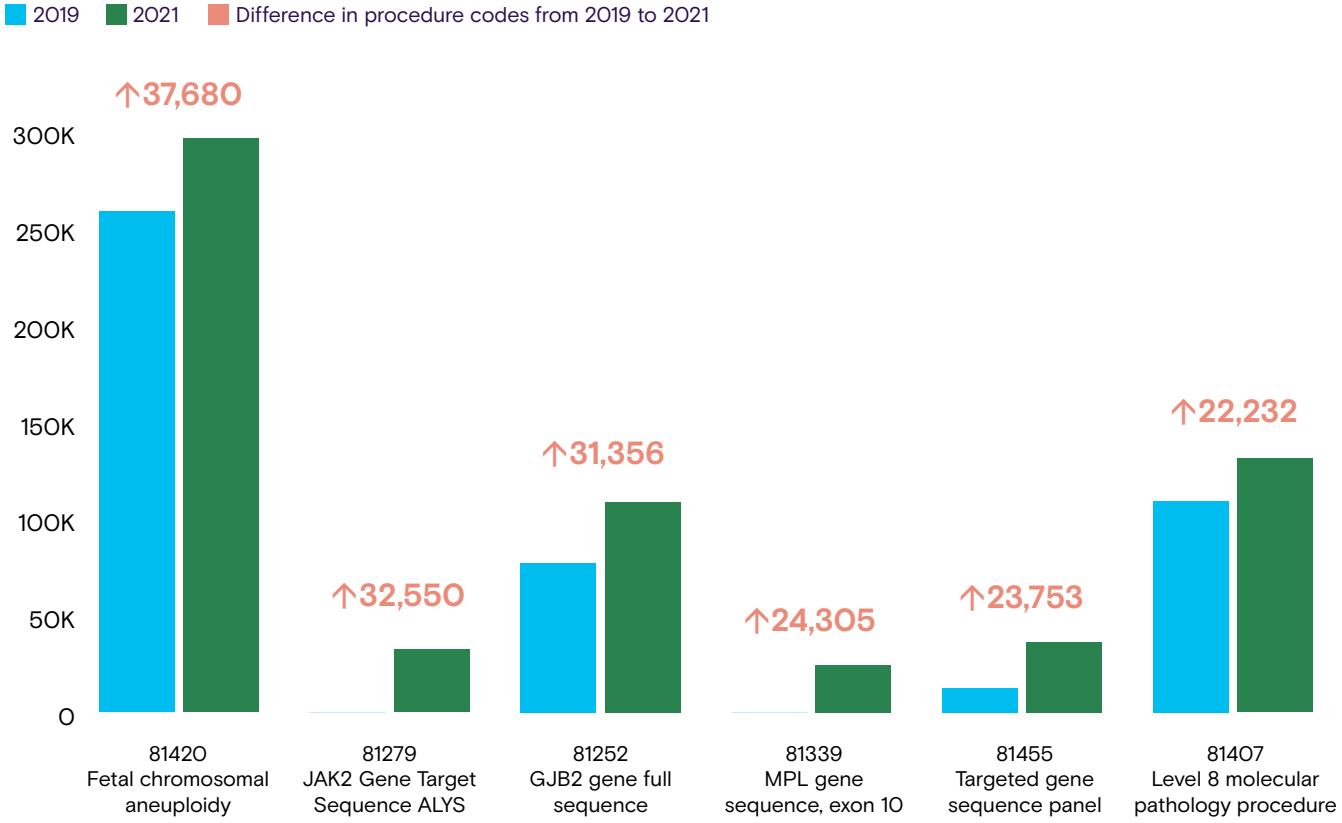
While the risk of passing inherited conditions like spinal muscular atrophy and cystic fibrosis doesn't necessarily grow with age, the risk of preterm birth and other complications do—factors that can make it more difficult to deliver children who have inherited these conditions.

Although a causal connection has not been found, research shows that pregnant women with bacterial vaginosis are also more likely to give birth too early. As previously mentioned, in 2021, two new MAAA claims codes relating to diagnosing this condition were added [CPT codes 81513 and 81514].

Precision medicine procedures focused on identifying these risks can help people make more informed decisions about parenthood, especially for mothers waiting to have children after the age of 35.

The growing use of precision medicine within fetal and pregnancy health represents an exciting expansion from its life-saving origins in oncology to broader applications for people at all stages of life. Still, new advancements in data and technology ensure that precision medicine remains at the cutting edge of care for cancer patients and their physicians.

MOLECULAR PATHOLOGY PROCEDURE CODE INCREASES FROM 2019 TO 2021



New oncology procedures reflect 2021 advancements and better data

Our final trend of interest centers on the development of several brand-new procedures for the detection and treatment of cancers.

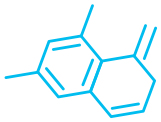
Claims data reveal the availability of new oncology-related molecular pathology procedures in 2021. These procedures were either entirely unavailable before 2021, or were performed fewer than 10 times per year before 2021:

- Janus kinase 2 (JAK2) gene sequencing for myeloproliferative disorders, often associated with blood cancers [CPT code 81279]
- Myeloproliferative leukemia (MPL) gene analysis, common variants [CPT code 81338]
- MPL gene analysis, sequence analysis [CPT code 81339]
- Tumor protein 53 (TP53) full-sequence gene analysis, associated with Li-Fraumeni syndrome and breast, bone and soft tissue cancers [CPT code 81351]

These procedures assess patients for genetic mutations that indicate a heightened risk for some especially rare and debilitating conditions, namely blood, bone, and soft tissue cancers. These procedures signal especially good news for the more than 200,000 patients in the US who currently live with myeloproliferative disorders (MPDs) that can lead to stroke, thrombocythemia, leukemia, and other diseases.

Prior to developments made within the past couple of decades, these patients had access to few or no targeted treatment options. Following the identification of the JAK2 and ABL genes, survival rates are up to nearly 85% for certain MPDs.

Precision medicine still faces economic hurdles in supporting patients with the rarest diseases. Life science developers are generally incentivized to develop treatments for larger markets—although the Orphan Drug Act of 1983 and Rare Disease Act of 2002 aim to fund manufacturers, reduce R&D costs, and ease testing protocols. But developers also face the unavoidable challenge of finding eligible clinical trial candidates among such small patient pools.



Four new molecular pathology procedures targeting rare bone and blood disorders became available in 2021, offering hope for 200,000+ patients in the U.S. who currently live with myeloproliferative disorders.

As precision medicine procedures proliferate, they produce valuable data about disease volumes in general and among particular populations, environmental factors associated with the disease, and the genetic factors that impact its severity. All of these data can help providers better understand who their patients are, which populations are more likely to become patients in the future, and how to treat those patients.

Likewise, these data can help life science organizations prioritize R&D, identify candidates and establish protocols for clinical trials, and market their treatments more effectively to patients and providers.

For cancer patients—or anyone—requiring an organ transplant for treatment, another development within precision medicine offers newfound hope.

Transplant matching is getting cheaper, too

Overall, average charges for molecular pathology procedures decreased slightly from $1,187 per procedure in 2019 to $1,087 per procedure in 2021 (an 8.4% decrease). Several individual procedures saw considerable decreases in average charge during the same period:

- Genetic analysis for hereditary retinal disorders [CPT code 81434] dropped $4,556 from an average of $8,713 per procedure in 2019 to $4,157 in 2021 **(a 52% decrease)**.
- Exome sequence analysis [CPT code 81415] dropped $3,426 from an average of $10,816 per procedure in 2019 to $7,390 in 2021 **(a 32% decrease)**.
- Mitochondrial gene sequencing [CPT code 81440] fell $3,354 from an average of $8,921 per procedure in 2019 to $5,567 in 2021 **(a 38% decrease)**.
- Genomic sequencing for hereditary peripheral neuropathies [CPT code 81448] fell $3,161 from an average of $5,843 per procedure in 2019 to $2,682 in 2021 **(a 54% decrease)**.

Stem cell and organ donation recipients are also seeing reductions in procedure costs.

These reductions could reflect the drop in demand that molecular pathology procedures saw throughout the pandemic. They might also be a product of doctors increasingly performing more complex, accurate, and less expensive MAAA procedures to address some of the same issues.

AVERAGE MOLECULAR PATHOLOGY PROCEDURE COST DECREASES ACROSS 3 YEARS

CPT Code	Short description	2019 Average charges	2021 Average charges	2019 to 2021 difference in procedure costs
81434	Hereditary retinal disorders analysis panel, 15 genes	$8,713	$4,157	**- $4,556**
81415	Exome sequence analysis	$10,816	$7,390	**- $3,426**
81440	Nuclear encoded mitochondrial analysis panel, ≥100 genes	$8,921	$5,567	**- $3,354**
81448	Hereditary peripheral neuropathies panel, ≥5 genes	$5,843	$2,682	**- $3,161**
81443	Genetic testing for severe inherited conditions, ≥15 genes	$9,636	$6,591	**- $3,044**
81266	Comparative Short Tandem Repeat (STR) marker analysis	$4,205	$1,258	**- $2,947**
81410	Aortic dysfunction or dilation analysis panel, ≥9 genes	$4,290	$1,402	**- $2,888**
81333	Transforming growth factor beta-induced gene analysis	$2,383	$150	**- $2,233**
81455	Solid organ or hematolymphoid neoplasm analysis, ≥51 genes	$9,550	$7,485	**- $2,065**
81439	Hereditary cardiomyopathy analysis panel, ≥5 genes	$3,208	$1,262	**- $1,946**

The first of these procedures is a 15-gene analysis of the genes associated with hereditary retinal disorders, including retinitis pigmentosa, Leber congenital amaurosis, and cone-rod dystrophy.

Exome sequence analysis can sequence large amounts of DNA rapidly by examining all of the pieces of DNA responsible for specific functions (known as exons) in a single genome. Exome sequencing is used to identify the genetic causes of disease in patients, and to map rare variants of complex disorders.

Mitochondrial sequencing, on the other hand, examines the DNA within human cells' mitochondria, enabling the detection of mutations that can cause diseases at any stage of life.

The cost reductions in advanced molecular pathology procedures could provide some hope for patients facing these rare diseases.

Patients, providers see benefits in preventive care—and precision medicine can help

It's been a tough few years for anyone trying to make sense of the healthcare market and its shifting trends. The ongoing pandemic has shaken up long-term patterns and added new layers of complexity to an already complicated ecosystem.

- One trend has carried through all this chaos, however: Patients want greater control over their care and their lives, and precision medicine is offering solutions.
- Improved technology makes it simpler and less expensive to screen for multiple disease markers at once, saving patients time, money, and worry over factors that may have otherwise been missed.
- Parents now have greater insight than ever into the health risks they may be passing on to their children, as well as into the health of their developing fetus in real time.
- Plus, some of humanity's most concerning diseases are a little less terrifying, as screening methods become cheaper and more accurate.

Every year, new developments in precision medicine give patients greater insight into life-changing health decisions:

- What genetic risks should I be prepared for as I age?
- Is a traditional pregnancy right for me?
- How can I mitigate the risks associated with my lifestyle?

In a consumer culture centered on personal control and custom-tailored solutions, these advances are helping patients find more value and greater choice in healthcare.

For providers, precision medicine is an avenue toward patient-tailored preventive care. The results of a single genetic sequencing panel can give physicians the information necessary to build a lasting, productive relationship with their patients around prevention and risk counseling, hopefully reducing the need for more intensive, expensive care options.

Life science developers should see opportunities in precision medicine, too. If not in the development of new tests and procedures, biopharma and medical device companies should at least understand the value of the data that precision medicine produces. Whether in finding ideal candidates for clinical trials, identifying high-value molecules for future treatments, or simply understanding the demographics of potential patient bases, a healthcare market with more precision medicine means the output of more data that can be leveraged throughout R&D and commercialization.

The rise of precision medicine is good news for everyone.

The providers and developers who are able to turn the data it provides into meaningful intelligence could find themselves ahead of the next market-defining trend. But they'll still need to keep a close eye on technological markets outside of healthcare, where consumers' purchasing trends continue to provide a barometer for their expectations toward care.

Methodology

The evaluation was based on a longitudinal analysis from our medical claims database that is comprised of data from the top clearinghouses in the U.S. Patient healthcare utilization covers all settings of care, including hospitals, surgery centers, long-term care, and office visits.

The analysis included patients of all ages and all payor categories (Medicare, Medicaid, Commercial, and VA) to ensure the results reflected a broad and clinically representative view of the U.S. population.

About Definitive Healthcare

At Definitive Healthcare, our passion is to transform data, analytics, and expertise into healthcare commercial intelligence. We help clients uncover the right markets, opportunities, and people, so they can shape tomorrow's healthcare industry. Our SaaS platform creates the path to commercial success in the healthcare market, so companies can identify where to go next.

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