

# Personalized Medicine

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## A New Treatment Paradigm

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# What is Personalized Medicine?

**Personalized medicine**, sometimes referred to as *precision* or *individualized* medicine, is an emerging field of medicine that uses diagnostic tools to identify specific biological markers, often genetic, to help assess which medical treatments and procedures will be best for each patient.

# Why is Personalized Medicine so Important?

Personalized medicine incorporates risk assessment, prevention, detection, diagnosis, treatment and management in its approach to patient care.



## **RISK ASSESSMENT:**

Genetic testing to reveal predisposition to disease



## **DIAGNOSIS:**

Accurate disease diagnosis enabling individualized treatment strategy



## **PREVENTION:**

Behavior/Lifestyle/  
Treatment intervention to prevent disease



## **TREATMENT:**

Improved outcomes through targeted treatments and reduced side effects



## **DETECTION:**

Early detection of disease at the molecular level



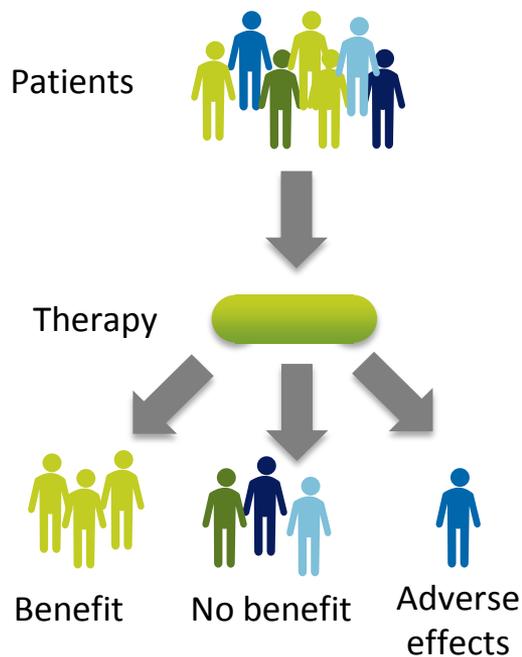
## **MANAGEMENT:**

Active monitoring of treatment response and disease progression

# A New Treatment Paradigm

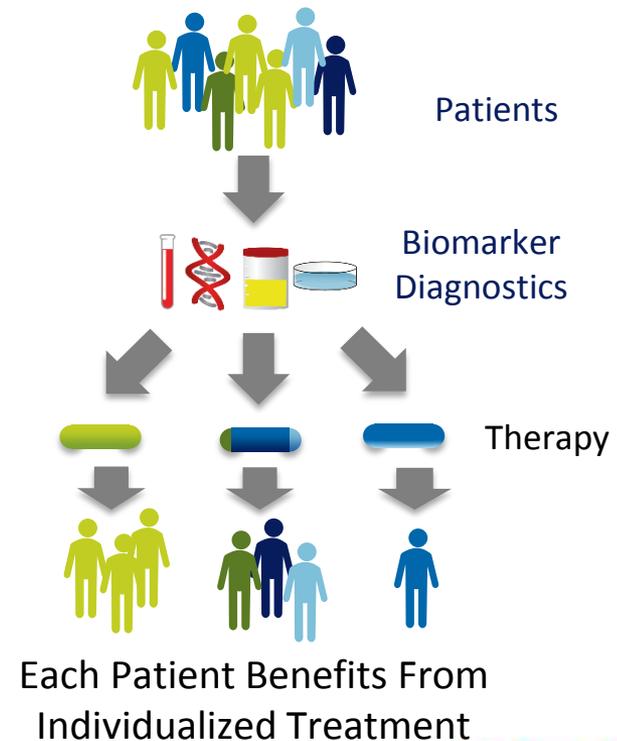
## Without Personalized Medicine:

Some Benefit, Some Do Not



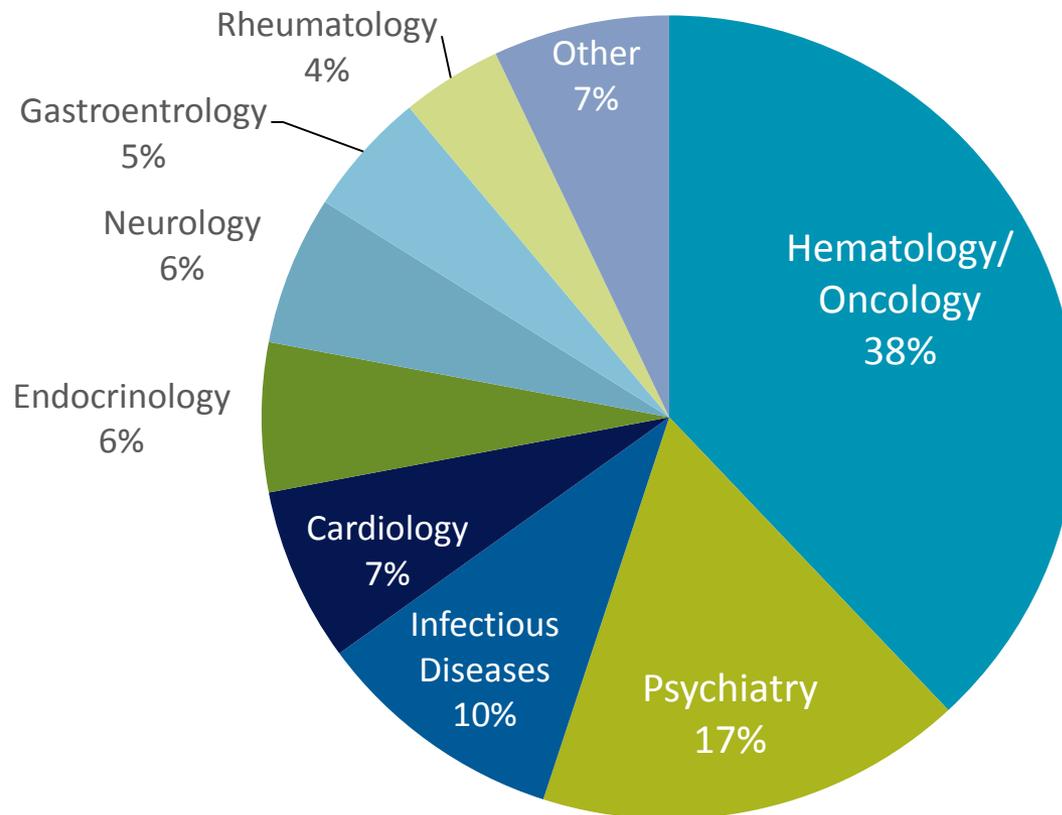
## With Personalized Medicine:

Each Patient Receives the Right Medicine For Them



# Personalized Medicines Are Benefiting Patients Across Many Different Diseases

*FDA Approvals with Biomarker information in the approved labeling*



# Pop Quiz!

Q: What was the first personalized medicine?



# Pop Quiz!

Q: What was the first personalized medicine?

A: Herceptin, first approved in 1998 to treat HER2-positive breast cancer.



*“Development and approval of Herceptin marked the dawn of a new era of cancer treatment by bringing an emerging understanding of cancer genetics out of the laboratory and to the patient’s bedside. The story of Herceptin also emphasized a profound lesson: not all cancers are the same. Breast cancer – as well as other cancers – cannot be viewed as a single disease, but rather as a group of several subtypes, each with its distinct molecular signature.”*

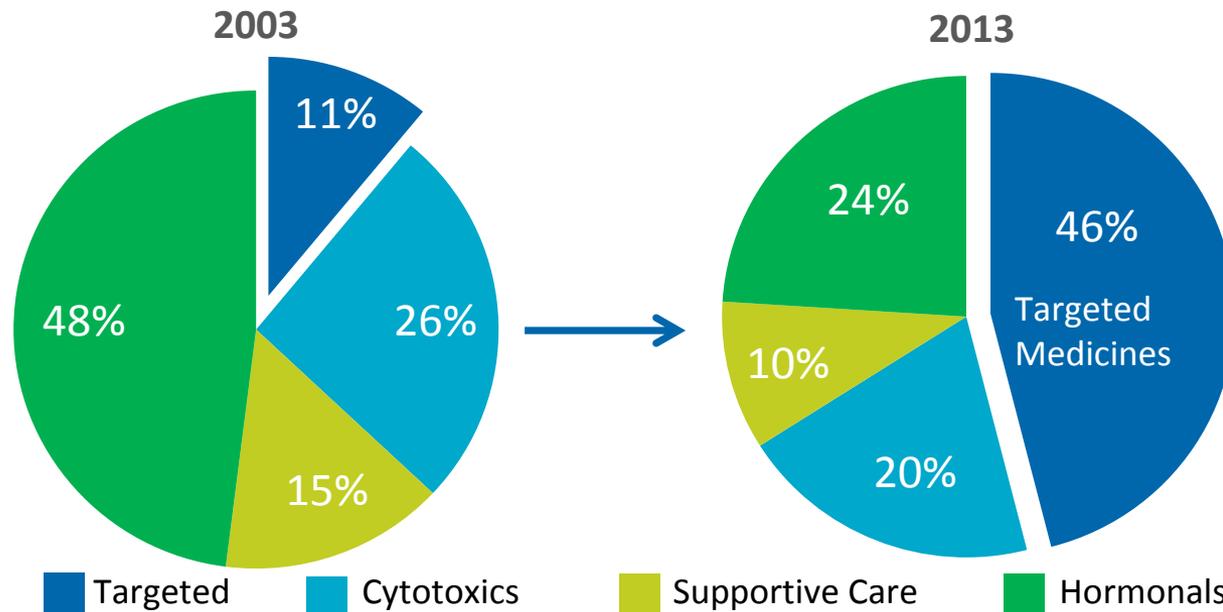


# Molecular Sub-typing in Breast Cancer

Subtype	Prevalence	Disease Characteristics	Prognosis
<b>Luminal A (HR+/HER2-)</b> <i>Hormone receptor-positive (either estrogen or progesterone), HER2-negative</i>	Majority of breast cancers (30-70%)	Slow growing, less aggressive	Most favorable prognosis, likely to respond to hormonal therapy
<b>Triple-negative (HR-/HER2-)</b> <i>Hormone receptor-negative (neither estrogen nor progesterone), HER2-negative</i>	15-20% of breast cancers (Nearly 2X more common in African American women than Caucasian women; more common in women with a BRCA1 gene mutation)	Fast growing and aggressive	Currently a poor prognosis due to lack of response to hormonal or HER2 targeted therapies
<b>Luminal B (HR+/HER2+)</b> <i>Hormone receptor-positive (either estrogen or progesterone), HER2-positive</i>	10-20% of breast cancers	Tend to be more aggressive than luminal A breast cancers due to high levels of Ki67 protein, which triggers excess cell growth	Fairly high survival rates due to high treatment response
<b>HER2-enriched (HR-/HER2+)</b> <i>Hormone receptor-negative (neither estrogen nor progesterone), HER2-positive</i>	5-15% of breast cancers	Fast growing and spreads aggressively	Recent advances in HER2-targeted therapies improve prognosis
<p><i>Abbreviations: HR (hormone receptor; either estrogen or progesterone); HER (human epidermal growth factor receptor). A positive (+) sign denotes the presence of a receptor, while a negative sign (-) denotes the absence.</i></p>			

# Cancer is on the Leading Edge of Personalized Medicine

*Breakdown of Oncology Treatment Modalities,  
Global Market share 2003-2013\**

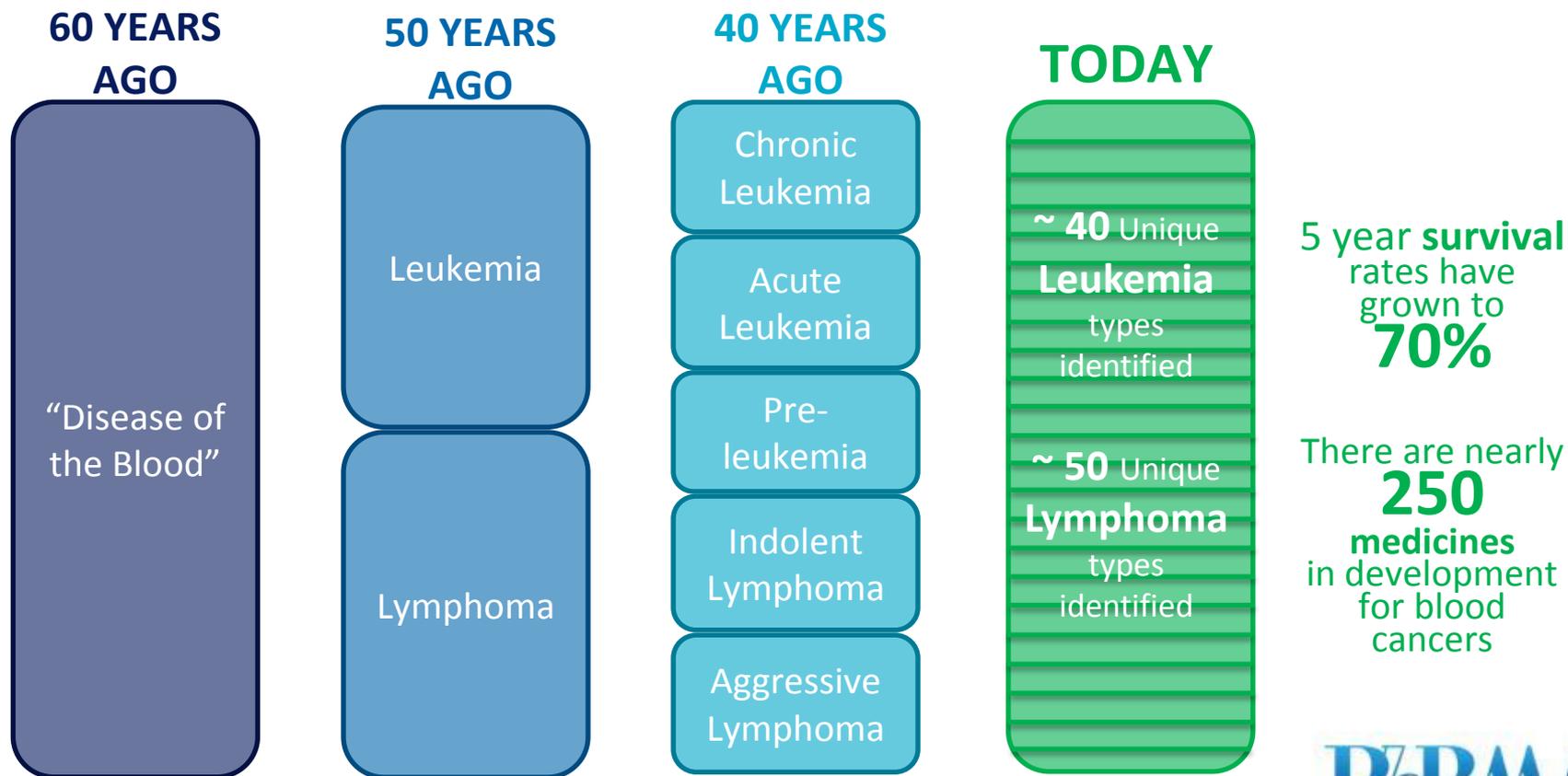


\*Definitions: Targeted therapies - identify and attack specific types of cancer cells with less harm to normal cells; Cytotoxics – agents that kill rapidly developing cells (as in chemotherapy); Supportive care - care given to improve quality of life by preventing or treating the symptoms of a disease or the effects of its treatment; Hormonals - treatments that add, block, or remove hormones to slow or stop the growth of certain cancers.



# Advances in Personalized Medicine Improve Outlook for Patients with Blood Cancers

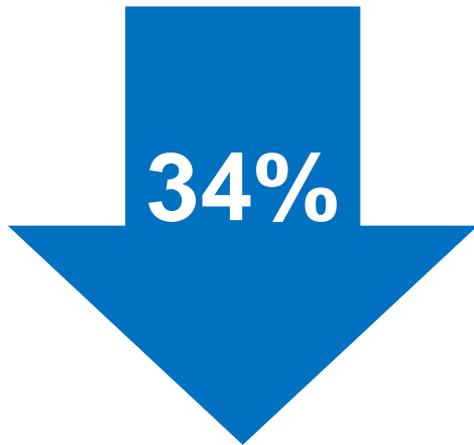
A greater understanding of the molecular basis of disease has transformed what was once known collectively as “disease of the blood,” into multiple subtypes of leukemias and lymphomas.



Source: M Aspinal, former President Genzyme Genetics (cited at [http://www.comtecm.com/biomarker/2014/Uploads/Editor/PDF/ppt/Edward%20Abrahams\\_Key%20Note%20Lecture.pdf](http://www.comtecm.com/biomarker/2014/Uploads/Editor/PDF/ppt/Edward%20Abrahams_Key%20Note%20Lecture.pdf)); National Cancer Institute,; SEER Cancer Statistics Review, 1975-2011, [http://seer.cancer.gov/csr/1975\\_2011/](http://seer.cancer.gov/csr/1975_2011/), based on November 2013 SEER data submission, posted to the SEER web site, April 2014; PhRMA, “Medicines in Development for Leukemia & Lymphoma,” April 2015 (all cites accessed May 2015).

# Personalized Medicine Can Create Efficiencies in the Health Care System

## Breast Cancer



Reduction in chemotherapy use would occur

If women with breast cancer receive a genetic test of their tumor prior to treatment

## Metastatic Colorectal Cancer



In annual health care cost savings would be realized

If patients with metastatic colorectal cancer receive a genetic test for the KRAS gene prior to treatment

## Stroke



Strokes could be prevented each year  
If a genetic test is used to properly dose blood thinners

# Pop Quiz!

Q: How many personalized medicines are available today?



# Pop Quiz!

Q: How many personalized medicines are available today?

A: More than 140!



# 2015: Banner Year for Personalized Medicine



**28% of new medicines**  
approved by the FDA in 2015  
were personalized medicines

## 2015 Personalized Medicine Highlights:

- 35% of new **cancer** treatments were found to be personalized medicines, including:
  - Two new medicines for patients with different forms of non-small cell **lung cancer**
  - A new targeted therapy for **melanoma**
- A new combination therapy for patients with **cystic fibrosis**
- Two new medicines to help patients with a difficult-to-treat form of **high cholesterol**

# Where have we seen personalized medicine lately?



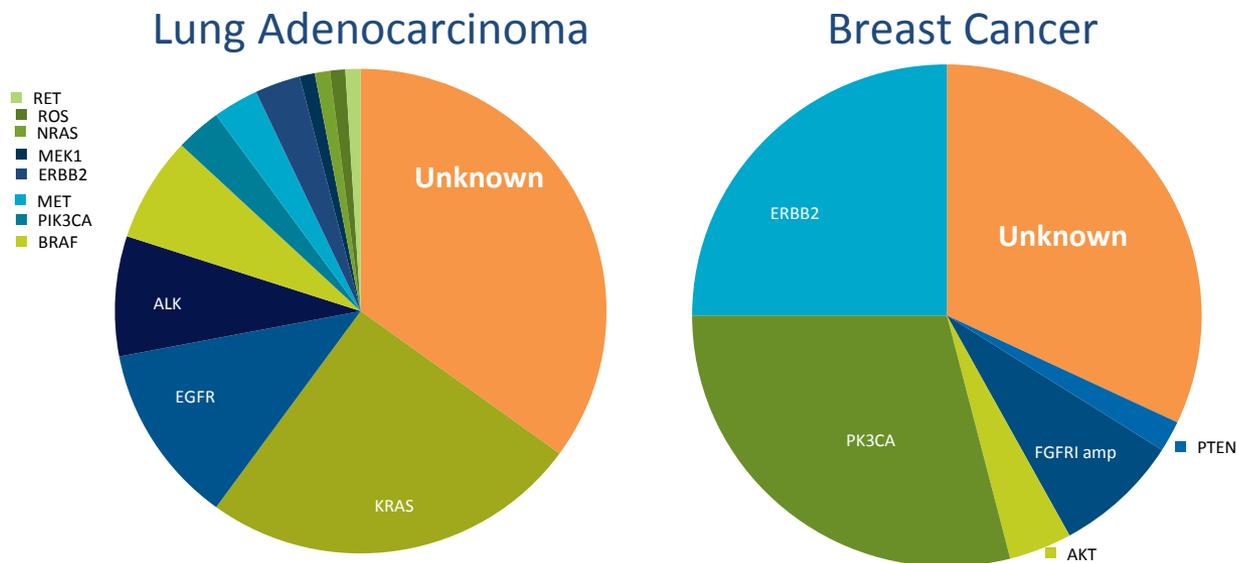
## THE PRECISION MEDICINE INITIATIVE



# Researchers Have Made Great Progress but Challenges Remain

We now know that cancer is not a single disease, but rather more than 200 unique diseases, many of which are caused by genetic mutations. Identifying these mutations has led to tremendous advances against many cancers, but the complexity of each disease presents great challenges for researchers, as they explore still yet unknown alterations.

## Selected Genomic Alterations Known to Drive Disease Progression in Common Cancers



Sources: LA Garraway, "Genomics-Driven Oncology: Framework for an Emerging Paradigm," *J Clin Onc.* 2013; 31(15):1806-1814; American Society of Clinical Oncology, "Studies Reveal Potential New Targeted Therapies for Common, Hard-to-Treat Cancers," <http://www.asco.org/press-center/studies-reveal-potential-new-targeted-therapies-common-hard-treat-cancers>, May 2014 (accessed May 2015).

# Pop Quiz!

Q: What portion of the pipeline has the potential to be personalized medicines?



# Pop Quiz!

Q: What portion of the pipeline has the potential to be personalized medicines?

A: 42%!

# The Biopharmaceutical Industry is Committed to Personalized Medicine



Source: PhRMA Innovation Hub, "Biopharma Companies Are Driving Personalized Medicine Forward," <http://innovation.org/gallery/biopharma-companies-are-driving-personalized-medicine-forward>.

# Ensuring Future Innovation in Personalized Medicine

At a time when the scientific promise is greater than ever, thoughtful policies are necessary to accelerate advances in targeted therapy for patients.



- Align regulatory policy with rapid pace of science
- Ensure that payment and delivery models recognize patients' differences, both scientifically and in terms of their own preferences

# Resources

- Innovation Hub

- [www.innovation.org](http://www.innovation.org)



- PhRMA Personalized Medicine Chartpack

- <http://chartpack.phrma.org/personal-medicines-in-development-chartpack>



- Personalized Medicine Coalition

- <http://personalizedmedicinecoalition.org/>



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# The Impact of Genetics: Beyond the Bench and Bedside



<https://youtu.be/6ymoPRWxZI8>

# Personalized Medicine in Practice

A mini case study.

# Personalized Medicine in Practice



## Scientific Name *Crustulumitis*

### Symptoms

Irritability, abdominal cramping, growling stomach

### Causes

May flare up between meals, and often following a salty or savory meal. Symptoms may also be more pronounced during times of increased stress or emotional instability.

### Risk Factors

Similar to irritable bowel syndrome (IBS), many people experience symptoms of mild *Crustulumitis* as a result of inconsistent daily dietary habits or as a response to stressful or emotional situations.

# Personalized Medicine in Practice



## Scientific Name *Crustulumitis*

### Diagnosis

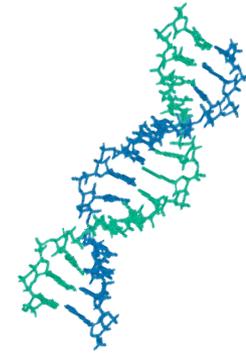
*Crustulumitis* can often be detected by conducting a thorough Q&A assessment with a knowledgeable professional. Verbal confirmation of symptoms often can indicate the level of severity of a case of *Crustulumitis*, though it can sometimes be confused with *Glacies crepitosis* and *Fructusitis*.

### Treatment

While mild *Crustulumitis* may be treated by preliminary administration of easily available oral glucose supplements, a simple lab test can be conducted on any available deoxyribnucleic acid sample, which will indicate the mechanism of treatment most likely to relieve symptoms for patients.

# Results

Lab result
<b>CHO</b> Regular, wild type (55% of patients)
<b>CHO+</b> Gene amplification (30% of patients)
<b>CHO-</b> Gene deletion (10% of patients)
<b>CHO~</b> Missense mutation (5% of patients)



# Results

Lab result	Treatment
<b>CHO</b> Regular, wild type (55% of patients)	Oatmeal chocolate chip cookie
<b>CHO+</b> Gene amplification (30% of patients)	Chocolate brownie cookie
<b>CHO-</b> Gene deletion (10% of patients)	Sugar cookie
<b>CHO~</b> Missense mutation (5% of patients)	Carrots

*\*1 to 3 doses, depending on the severity of the flare-up.*

