Sensitivity / Specificity / Prevalence

Positive predictive value

- 60% / 60% / 0.1% 0.15%
- 80% / 80% / 0.1% 0.4%
- 80% / 80% / 1.0% 3.9%
- 80% / 80% / 10% 30.8%

Table: Selected Personalized Medicine Drugs, Treatments and Diagnostics as of September 2011*

Indications in quotes and otherwise unattributed, are cited from the therapeutic or diagnostic product label.

Therapeutic product labels contain pharmacogenomic information as:

- *Information only*
- *Recommended*
- *Required*
- *Unhighlighted products have no pharmacogenomic information, recommendations or requirements in the label.*

THERAPY	BIOMARKER/TEST	INDICATION
Mivacron® (mivacurium)	Cholinesterase gene	**Anesthesia adjunct:** "Mivacron is metabolized by plasma cholinesterase and should be used with great caution, if at all, in patients known to be or suspected of being homozygous for the atypical plasma cholinesterase gene."
Ansaid® (flurbiprofen)	*CYP2C9*	**Arthritis:** "*In vitro* studies have demonstrated that cytochrome P450 2C9 plays an important role in the metabolism of flurbiprofen to its major metabolite, 4'-hydroxy-flurbiprofen."
Depakote® (divalproex)	UCD *(NAGS; CPS; ASS; OTC; ASL; ARG)*	**Bipolar disorder:** "Hyperammonemic encephalopathy, sometimes fatal, has been reported following initiation of valproate therapy in patients with urea cycle disorders [UCDs]...particularly ornithine transcarbamylase deficiency [OTC]."
Aromasin® (exemestane) **Arimidex® (anastrozole)** **Nolvaldex® (tamoxifen)**	Estrogen Receptor (ER)	**Breast cancer:** Exemestane is indicated for adjuvant treatment of post-menopausal women with ER-positive early breast cancer. Anastrozole is for treatment of breast cancer after surgery and for metastases in post-menopausal women. Tamoxifen is the standard therapy for estrogen receptor-positive early breast cancer in pre-menopausal women.
Chemotherapy	Mammostrat®	**Breast cancer:** Prognostic immunohistochemistry (IHC) test used for postmenopausal, node negative, estrogen receptor expressing breast cancer patients who will receive hormonal therapy and are considering adjuvant chemotherapy.
Chemotherapy	MammaPrint®	**Breast cancer:** Assesses risk of distant metastasis in a 70-gene expression profile.
Chemotherapy	Onco*type* DX® 16-gene signature	**Breast cancer:** A 16-gene signature (plus five reference genes) indicates whether a patient has a low, intermediate, or high risk of having a tumor return within 10 years. Low-risk patients may be treated successfully with hormone therapy alone. High-risk patients may require more aggressive treatment with chemotherapy.

December 28, 2007

THE DNA AGE

After DNA Diagnosis: ‘Hello, 16p11.2. Are You Just Like Me?’

By AMY HARMON

The girls had never met, but they looked like sisters.

There was no missing the similarities: the flat bridge of their noses, the thin lips, the fold near the corner of their eyes. And to the families of 14-year-old Samantha Napier and 4-year-old Taygen Lane there was something else, too. In the likeness was lurking an explanation for the learning difficulties, the digestion problems, the head-banging that had troubled each of them, for so long.

Several of the adults wiped tears from their eyes. “It’s like meeting family,” said Jessica Houk, Samantha’s older sister, who accompanied her and their mother to a Kentucky amusement park last July to greet Taygen.

But the two families are not related, and would never have met save for an unusual bond: a few months earlier, a newly available DNA test revealed that Samantha and Taygen share an identical nick in the short arm of their 16th chromosomes.

With technology that can now scan each of an individual’s 46 chromosomes for minute aberrations, doctors are providing thousands of children lumped together as “autistic” or “developmentally delayed” with distinct genetic diagnoses. The symptoms, they are finding, can be traced to one of dozens of deletions or duplications of DNA that were previously hard or impossible to detect.

Some mutations are so rare that they are known only by their chromosomal address: Samantha and Taygen are two of only six children with the diagnosis “16p11.2.”

23andMe Bows To FDA's Demands, Drops Health Claims

by SCOTT HENSLEY

December 06, 2013 1:49 PM ET

23andMe will still perform genetic tests, but it won't be making health-related interpretations of the results.

YouTube

The maker of a $99 personal genome test blinked.

Silicon Valley's 23andMe said late Thursday that it would comply with the Food and Drug Administration's demand that the company stop marketing health-related genetic tests.

People will still be able to pay 23andMe to have their DNA analyzed to learn about their ancestors. And customers will get a file of their raw genetic info.

But while 23andMe tries to work things out with the FDA, the company won't be telling people they have genetic profiles that predispose them to particular illnesses, or predict their responses to prescription drugs. In other words, no more health claims.

"We remain firmly committed to fulfilling our long-term mission to help people everywhere have access to their own genetic data and have the ability to use that information to improve their lives," Anne Wojcicki, co-founder and CEO of 23andMe, said in a statement. "Our goal is to work cooperatively with the FDA to provide that opportunity in a way that clearly demonstrates the benefit to people and the validity of the science that underlies the test."

The company has a pretty deep regulatory hole to crawl out of.