

INTRODUCTION

Dante Labs Pharmacogenetics Report is for informational purposes only.

This report is NOT intended for US persons. This report was not submitted for approval to the US Food and Drug Administration (FDA).

The Pharmacogenetics Report analyzes a large amount of genomic data, associating genetic variants found in the genomic files with variants known from the scientific literature. While this Report does not require EMA approval, we do want to point out that it has not been reviewed or approved by the EMA for such use.

The genetic analysis and reporting are based on information from one or more published third party scientific and medical studies. We do not independently judge the validity or accuracy of such published scientific information.

Because scientific and medical information changes over time, your risk assessment and genetically tailored prevention for one or more of the medications contained within this report may also change over time.

Therefore, this report may not be 100% accurate (e.g., new research could mean different results) and may not predict actual results or outcomes. A person's risk of any particular phenotype, condition or trait is also based on other factors not yet analyzed in this report (e.g., diet, lifestyle, etc.).

This report may be updated from time to time so that the analysis and reporting incorporates new or changed research or scientific results. Because of this, the reports produced may change over time.

This genetic report should not be used in place of a visit with or advice from your doctor or other qualified healthcare professional. You should always get the advice of your doctor or other appropriate health care professional if you have any question about diagnosis, treatment, prevention, mitigation, or cure of any medical condition, phenotype, condition, impairment, or the status of your health. Do not stop any medications you have been prescribed, start any new medications, or modify any medical treatments ordered by your healthcare provider without first talking with your healthcare provider. If you have any healthcare related questions, please promptly consult your physician or other qualified healthcare provider.

QUICK SUMMARY

MEDICATIONS		
CONDITION NAME	RESULTS	MAIN MESSAGE
Acenocoumarol	✓	No variants detected
Adalimumab	✓	No variants detected
Allopurinol	✓	No variants detected
Amisulpride	✗	We found a variant related to your reaction to Amisulpride
Amitriptyline	✓	No variants detected
Anastrozole	⚠	We found a variant related to your reaction to Anastrozole
Aripiprazole	✗	We found a variant related to your reaction to Aripiprazole
Asparaginase	✓	No variants detected

MEDICATIONS		
CONDITION NAME	RESULTS	MAIN MESSAGE
Aspirin	⚠️	We found a variant related to your reaction to Aspirin
Ataluren	✅	No variants detected
Atazanavir	✅	No variants detected
Atorvastatin	⚠️	We found a variant related to your reaction to Atorvastatin
Azathioprine	❌	We found a variant related to your reaction to Azathioprine
Boceprevir	⚠️	We found a variant related to your reaction to Boceprevir
Budesonide	✅	No variants detected
Buprenorphine	⚠️	We found a variant related to your reaction to Buprenorphine
Caffeine	✅	No variants detected
Carbamazepine	✅	No variants detected
Carboplatin	⚠️	We found a variant related to your reaction to Carboplatin
Celecoxib	✅	No variants detected
Cerivastatin	✅	No variants detected
Cetuximab	⚠️	We found a variant related to your reaction to Cetuximab
Chlorproguanil	✅	No variants detected
Cisplatin	⚠️	We found a variant related to your reaction to Cisplatin
Citalopram	⚠️	We found a variant related to your reaction to Citalopram
Clomipramine	✅	No variants detected
Clozapine	❌	We found a variant related to your reaction to Clozapine
Cocaine	✅	No variants detected
Cyclosporine	⚠️	We found a variant related to your reaction to Cyclosporine
Dapsone	✅	No variants detected
Daunorubicin	✅	No variants detected
Desipramine	✅	No variants detected
Diclofenac	✅	No variants detected
Digoxin	⚠️	We found a variant related to your reaction to Digoxin
Docetaxel	⚠️	We found a variant related to your reaction to Docetaxel
Doxepin	✅	No variants detected
Doxorubicin	⚠️	We found a variant related to your reaction to Doxorubicin
Efavirenz	⚠️	We found a variant related to your reaction to Efavirenz
Epirubicin	⚠️	We found a variant related to your reaction to Epirubicin
Erlotinib	✅	No variants detected
Escitalopram	✅	No variants detected
Etanercept	✅	No variants detected

MEDICATIONS		
CONDITION NAME	RESULTS	MAIN MESSAGE
Ethambutol	✓	No variants detected
Ethanol	✓	No variants detected
Etoposide	✓	No variants detected
Exemestane	⚠	We found a variant related to your reaction to Exemestane
Fentanyl	⚠	We found a variant related to your reaction to Fentanyl
Fluorouracil	⚠	We found a variant related to your reaction to Fluorouracil
Fluoxetine	⚠	We found a variant related to your reaction to Fluoxetine
Fluticasone propionate	✓	No variants detected
Fluticasone-salmeterol	✓	No variants detected
Furosemide	⚠	We found a variant related to your reaction to Furosemide
Gefitinib	✓	No variants detected
Glibenclamide	⚠	We found a variant related to your reaction to Glibenclamide
Gliclazide	⚠	We found a variant related to your reaction to Gliclazide
Glimepiride	⚠	We found a variant related to your reaction to Glimepiride
Glipizide	⚠	We found a variant related to your reaction to Glipizide
Haloperidol	✗	We found a variant related to your reaction to Haloperidol
Heroin	✓	No variants detected
Hydrochlorothiazide	✓	No variants detected
Imipramine	✓	No variants detected
Infliximab	✓	No variants detected
Interferon alfa-2b	✓	No variants detected
Isoniazid	✓	No variants detected
Ivacaftor	✓	No variants detected
Lamotrigine	✓	No variants detected
Latanoprost	⚠	We found a variant related to your reaction to Latanoprost
Leucovorin	✓	No variants detected
Lorazepam	⚠	We found a variant related to your reaction to Lorazepam
Lovastatin	✓	No variants detected
Mercaptopurine	✓	No variants detected
Metformin	✓	No variants detected
Methadone	⚠	We found a variant related to your reaction to Methadone
Methotrexate	✓	No variants detected
Mirtazapine	⚠	We found a variant related to your reaction to Mirtazapine
Morphine	⚠	We found a variant related to your reaction to Morphine

MEDICATIONS		
CONDITION NAME	RESULTS	MAIN MESSAGE
Naloxone	✓	No variants detected
Nevirapine	✗	We found a variant related to your reaction to Nevirapine
Nicotine	✓	No variants detected
Nortriptyline	✓	No variants detected
Olanzapine	✗	We found a variant related to your reaction to Olanzapine
Omeprazole	✗	We found a variant related to your reaction to Omeprazole
Ondansetron	⚠	We found a variant related to your reaction to Ondansetron
Oxaliplatin	⚠	We found a variant related to your reaction to Oxaliplatin
Oxazepam	⚠	We found a variant related to your reaction to Oxazepam
Oxycodone	⚠	We found a variant related to your reaction to Oxycodone
Paclitaxel	⚠	We found a variant related to your reaction to Paclitaxel
Paliperidone	✗	We found a variant related to your reaction to Paliperidone
Panitumumab	✓	No variants detected
Paroxetine	⚠	We found a variant related to your reaction to Paroxetine
Peginterferon alfa-2a	⚠	We found a variant related to your reaction to Peginterferon alfa-2a
Peginterferon alfa-2b	⚠	We found a variant related to your reaction to Peginterferon alfa-2b
Phenprocoumon	✓	No variants detected
Phenytoin	✓	No variants detected
Platinum based therapies	⚠	We found a variant related to your reaction to Platinum based therapies
Pravastatin	⚠	We found a variant related to your reaction to Pravastatin
Prednisolone	✓	No variants detected
Pyrazinamide	✓	No variants detected
Quetiapine	✗	We found a variant related to your reaction to Quetiapine
Radiotherapy	✗	We found a variant related to your reaction to Radiotherapy
Ribavirin	⚠	We found a variant related to your reaction to Ribavirin
Rifampin	✓	No variants detected
Risperidone	✗	We found a variant related to your reaction to Risperidone
Rituximab	⚠	We found a variant related to your reaction to Rituximab
Rosiglitazone	✓	No variants detected
Rosuvastatin	✓	No variants detected
Salbutamol	⚠	We found a variant related to your reaction to Salbutamol
Salmeterol	⚠	We found a variant related to your reaction to Salmeterol
Sildenafil	⚠	We found a variant related to your reaction to Sildenafil
Simvastatin	⚠	We found a variant related to your reaction to Simvastatin

MEDICATIONS		
CONDITION NAME	RESULTS	MAIN MESSAGE
Sirolimus	⚠	We found a variant related to your reaction to Sirolimus
Spironolactone		We found a variant related to your reaction to Spironolactone
Streptomycin	✓	No variants detected
Sunitinib	✓	No variants detected
Tacrolimus	⚠	We found a variant related to your reaction to Tacrolimus
Tamoxifen	⚠	We found a variant related to your reaction to Tamoxifen
Telaprevir	⚠	We found a variant related to your reaction to Telaprevir
Tramadol	⚠	We found a variant related to your reaction to Tramadol
Trastuzumab	⚠	We found a variant related to your reaction to Trastuzumab
Triamcinolone	✓	No variants detected
Trimipramine	✓	No variants detected
Valproic acid	✓	No variants detected
Venlafaxine	⚠	We found a variant related to your reaction to Venlafaxine
Vincristine	✓	No variants detected
Warfarin	⚠	We found a variant related to your reaction to Warfarin
Ziprasidone	✗	We found a variant related to your reaction to Ziprasidone
Remifentanyl	✓	No variants detected
Cyclophosphamide	✓	No variants detected
Irinotecan	✓	No variants detected
Abciximab	✓	No variants detected
Enalapril	✗	We found a variant related to your reaction to Enalapril

KEY SUMMARY

The above Summary provides an overview of the predicted risks for the patient. This information is based solely on genotype information and does not replace a doctor visit or a complete patient profile. Healthcare providers should consider also family history, presenting symptoms, current prescriptions, and other factors before making any clinical or therapeutic decisions.



No negative assertion based on your genotype.



We have found one or more variants associated with alterations in your reaction to the medication.



We have found one or more variants potentially associated with an increased risk for side effects.

DETAILED INFORMATION

AMISULPRIDE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Amisulpride, sold under the brand name Solian among others, is an antipsychotic medication used to treat schizophrenia. It is also used to treat dysthymia. It is usually classed with the atypical antipsychotics. Chemically it is a benzamide and like other benzamide antipsychotics, such as sulpiride, it is associated with a high risk of elevating blood levels of the lactation hormone, prolactin (thereby potentially causing the absence of the menstrual cycle, breast enlargement, even in males, breast milk secretion not related to breastfeeding, impaired fertility, impotence, breast pain, etc.), and a low risk, relative to the typical antipsychotics, of causing movement disorders. It has also been found to be modestly more effective in treating schizophrenia than the typical antipsychotics. Amisulpride is believed to work by reducing signalling via the dopamine D2 receptor. In amisulpride's case this is by blocking, or antagonizing, the receptor. Amisulpride's effectiveness in treating dysthymia and the negative symptoms of schizophrenia is believed to stem from its blockade of the presynaptic dopamine D2 receptors. These presynaptic receptors regulate the release of dopamine into the synapse, so by blocking them amisulpride increases dopamine concentrations in the synapse. This increased dopamine concentration is theorized to act on dopamine D1 receptors to relieve depressive symptoms (in dysthymia) and the negative symptoms of schizophrenia.

ANASTROZOLE

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Anastrozole, sold under the brand name Arimidex among others, is a medication used in addition to other treatments for breast cancer. Specifically it is used for hormone receptor-positive breast cancer. It has also been used to prevent breast cancer in those at high risk. It is taken by mouth. Common side effects include hot flushes, altered mood, joint pain, and nausea. Severe side effects include an increased risk of heart disease and osteoporosis. Use during pregnancy is known to harm the baby. Anastrozole is in the aromatase-inhibiting family of medications. It works by blocking the creation of estrogen.

ARIPRAZOLE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Aripiprazole, sold under the brand name Abilify among others, is an atypical antipsychotic. It is primarily used in the treatment of schizophrenia and bipolar disorder. Other uses include as an add-on treatment in major depressive disorder, tic disorders, and irritability associated with autism. A Cochrane review found evidence in schizophrenia not sufficient to determine effects on general functioning. Additionally, as many people dropped out of the studies before they were completed, the strength of the conclusions was low. It is taken by mouth or injection into a muscle. Common side effects include vomiting, constipation, sleepiness, dizziness, weight gain, and movement disorders. Serious side effects may include neuroleptic malignant syndrome, tardive dyskinesia, and anaphylaxis. In the older people there is an increased risk of death and it is not recommended for psychosis due to dementia. In pregnancy there is possible evidence of harm to the baby. It is not recommended in women who are breastfeeding. It has not been very well studied in people less than 18 years old. How it works is not entirely clear, but may involve effects on dopamine and serotonin.

ASPIRIN

Variant found:

- Gene: GPIBA
- Marker: rs6065
- Position: chr17:4836381
- Genotype: CT

Aspirin, also known as acetylsalicylic acid (ASA), is a medication used to treat pain, fever, or inflammation. Specific inflammatory conditions which aspirin is used to treat include Kawasaki disease, pericarditis, and rheumatic fever. Aspirin given shortly after a heart attack decreases the risk of death. Aspirin is also used long-term to help prevent further heart attacks, ischaemic strokes, and blood clots in people at high risk. It may also decrease the risk of certain types of cancer, particularly colorectal cancer. For pain or fever, effects typically begin within 30 minutes. Aspirin is a nonsteroidal anti-inflammatory drug (NSAID) and works similarly to other NSAIDs but also suppresses the normal functioning of platelets. One common adverse effect is an upset stomach. More significant side effects include stomach ulcers, stomach bleeding, and worsening asthma. Bleeding risk is greater among those who are older, drink alcohol, take other NSAIDs, or are on other blood thinners. Aspirin is not recommended in the last part of pregnancy. It is not generally recommended in children with infections because of the risk of Reye syndrome. High doses may result in ringing in the ears.

ATORVASTATIN

Variant found:

- Gene: KIF6
- Marker: rs20455
- Position: chr6:39325078
- Genotype: GG

Atorvastatin, sold under the trade name Lipitor among others, is a statin medication used to prevent cardiovascular disease in those at high risk and treat abnormal lipid levels. For the prevention of cardiovascular disease, statins are a first-line treatment. It is taken by mouth. Common side effects include joint pain, diarrhea, heart burn, nausea, and muscle pains. Serious side effects may include rhabdomyolysis, liver problems, and diabetes. Use during pregnancy may harm the baby. Like all statins, atorvastatin works by inhibiting HMG-CoA reductase, an enzyme found in the liver that plays a role in producing cholesterol.

AZATHIOPRINE

Variant found:

- Gene: ITPA
- Marker: rs7270101
- Position: chr20:3193893
- Genotype: AC

Azathioprine (AZA), sold under the brand name Imuran among others, is an immunosuppressive medication. It is used in rheumatoid arthritis, granulomatosis with polyangiitis, Crohn's disease, ulcerative colitis, and in kidney transplants to prevent rejection. It is taken by mouth or injected into a vein. Common side effects include bone marrow suppression and vomiting. Bone marrow suppression is especially common in people with a genetic deficiency of the enzyme thiopurine S-methyltransferase. Other serious risk factors include an increased risk of lymphoma. Use during pregnancy may result in harm to the baby. Azathioprine is in the purine analogue and antimetabolite family of medication. It works via 6-thioguanine to disrupt the making of RNA and DNA by cells.

BOCEPREVIR

Variant found:

- Gene: IFNL3, IFNL4
- Marker: rs12979860
- Position: chr19:39738787
- Genotype: CT

Boceprevir (INN, trade name Victrelis) is a protease inhibitor used to treat hepatitis caused by hepatitis C virus (HCV) genotype 1. It binds to the HCV nonstructural protein 3 active site. It was initially developed by Schering-Plough, then by Merck after it acquired Schering in 2009. It was approved by the FDA in May 2011. In January 2015, Merck announced that they would be voluntarily withdrawing Victrelis from the market due to the overwhelming superiority of newer direct-acting antiviral agents, such as ledipasvir/sofosbuvir.

BUPRENORPHINE

Variant found:

- Gene: Intergenic_non-coding
- Marker: rs2952768
- Position: chr2:208494234
- Genotype: CC

Buprenorphine, sold under the brand name Subutex, among others, is an opioid used to treat opioid addiction, acute pain, and chronic pain. It can be used under the tongue, by injection, as a skin patch, or as an implant. For opioid addiction it is typically only started when withdrawal symptoms have begun and for the first two days of treatment under direct observation of a health care provider. For longer term treatment of addiction a combination formulation of buprenorphine/naloxone is recommended to prevent misuse by injection. Maximum pain relief is generally within an hour with effects up to 24 hours. Side effects may include respiratory depression (decreased breathing), sleepiness, adrenal insufficiency, QT prolongation, low blood pressure, allergic reactions, and opioid addiction. Among those with a history of seizures, there is a risk of further seizures. Opioid withdrawal following stopping buprenorphine is generally less severe than with other opioids. It is unclear if use during pregnancy is safe and use while breastfeeding is not recommended. Buprenorphine affects different types of opioid receptors in different ways. Depending on the type of receptor it may be an agonist, partial agonist, or antagonist.

CARBOPLATIN

Variant found:

- Gene: XRCC1
- Marker: rs25487
- Position: chr19:44055726
- Genotype: CC

Carboplatin, sold under the trade name Paraplatin among others, is a chemotherapy medication used to treat a number of forms of cancer. This includes ovarian cancer, lung cancer, head and neck cancer, brain cancer, and neuroblastoma. It is used by injection into a vein. Side effects generally occur. Common side effects include low blood cell levels, nausea, and electrolyte problems. Other serious side effects include allergic reactions and increased future risk of another cancer. Use during pregnancy may result in harm to the baby. Carboplatin is in the platinum-based antineoplastic family of medications and works by interfering with duplication of DNA.

CETUXIMAB

Variant found:

- Gene: EGF
- Marker: rs4444903
- Position: chr4:110834110
- Genotype: GG

Cetuximab is an epidermal growth factor receptor (EGFR) inhibitor used for the treatment of metastatic colorectal cancer, metastatic non-small cell lung cancer and head and neck cancer. Cetuximab is a chimeric (mouse/human) monoclonal antibody given by intravenous infusion that is distributed under the trade name Erbitux in the U.S. and Canada by the drug company Bristol-Myers Squibb and outside those countries by the drug company Merck KGaA. In Japan, Merck KGaA, Bristol-Myers Squibb and Eli Lilly have a co-distribution. In July 2009, the FDA approved cetuximab (Erbitux) for treatment of colon cancer with wild-type KRAS, since it had little or no effect in colorectal tumors harboring a KRAS mutation (this also applied to the EGFR antibody panitumumab). This was the first genetic test to guide treatment of cancer. In July 2012, the FDA approved a real time PCR companion diagnostic test for KRAS, the therascreen KRAS test.

CISPLATIN

Variant found:

- Gene: XRCC1
- Marker: rs25487
- Position: chr19:44055726
- Genotype: CC

Cisplatin is a chemotherapy medication used to treat a number of cancers. These include testicular cancer, ovarian cancer, cervical cancer, breast cancer, bladder cancer, head and neck cancer, esophageal cancer, lung cancer, mesothelioma, brain tumors and neuroblastoma. It is given by injection into a vein. Common side effects include bone marrow suppression, hearing problems, kidney problems, and vomiting. Other serious side effects include numbness, trouble walking, allergic reactions, electrolyte problems, and heart disease. Use during pregnancy is known to harm the baby. Cisplatin is in the platinum-based antineoplastic family of medications. It works in part by binding to DNA and inhibiting its replication.

CITALOPRAM

Variant found:

- Gene: FKBP5
- Marker: rs4713916
- Position: chr6:35669983
- Genotype: AG

Variant found:

- Gene: HTR2A

- Marker: rs7997012
- Position: chr13:47411985
- Genotype: GG

Citalopram, sold under the brand name Celexa among others, is an antidepressant of the selective serotonin reuptake inhibitor (SSRI) class. It is used to treat major depressive disorder, obsessive compulsive disorder, panic disorder, and social phobia. Benefits may take one to four weeks to occur. It is taken by mouth. Common side effects include nausea, trouble sleeping, sexual problems, shakiness, feeling tired, and sweating. Serious side effects include an increased risk of suicide in those under the age of 25, serotonin syndrome, glaucoma, and QT prolongation. It should not be used in someone on a MAO inhibitor. Antidepressant discontinuation syndrome may occur when stopped. There are concerns that use during pregnancy may harm the baby.

CLOZAPINE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Clozapine, sold under the brand name Clozaril among others, is an atypical antipsychotic medication. It is mainly used for schizophrenia that does not improve following the use of other antipsychotic medications. In those with schizophrenia and schizoaffective disorder it may decrease the rate of suicidal behavior. It is more effective than typical antipsychotics, particularly in those who are treatment-resistant. It is taken by mouth. Clozapine is associated with a relatively high risk of low white blood cells (agranulocytosis), a condition of suppressed immunity which may result in death. To decrease this risk, it is recommended that the white blood cell count be regularly monitored. Other serious risks include seizures, inflammation of the heart, high blood sugar levels, and, in older people with psychosis as a result of dementia, an increased risk of death. Common side effects include drowsiness, decreased or increased saliva production, low blood pressure, blurred vision, and dizziness. The potentially permanent movement disorder tardive dyskinesia occurs in about 5% of people. Its mechanism of action is not entirely clear.

CYCLOSPORINE

Variant found:

- Gene: CYP3A5
- Marker: rs776746
- Position: chr7:99270539
- Genotype: TT

Ciclosporin, also spelled cyclosporine and cyclosporin, is an immunosuppressant medication and natural product. It is taken by mouth or by injection into a vein for rheumatoid arthritis, psoriasis, Crohn's disease, nephrotic syndrome, and in organ transplants to prevent rejection. It is also used as eye drops for keratoconjunctivitis sicca (dry eyes). Common side effects include high blood pressure, headache, kidney problems, increased hair growth, and vomiting. Other severe side effects include an increased risk of infection, liver problems, and an increased risk of lymphoma. Blood levels of the medication should be checked to decrease the risk of side effects. Use during pregnancy may result in preterm birth; however, ciclosporin does not appear to cause birth defects.

Ciclosporin is believed to work by decreasing the function of lymphocytes. It does this by forming a complex with cyclophilin to block the phosphatase activity of calcineurin, which in turn decreases the production of inflammatory cytokines by T-lymphocytes.

DIGOXIN

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Digoxin, sold under the brand name Lanoxin among others, is a medication used to treat various heart conditions. Most frequently it is used for atrial fibrillation, atrial flutter, and heart failure. Digoxin is taken by mouth or by injection into a vein. Common side effects include breast enlargement with other side effects generally due to an excessive dose. These side effects may include loss of appetite, nausea, trouble seeing, confusion, and an irregular heartbeat. Greater care is required in older people and those with poor kidney function. It is unclear whether use during pregnancy is safe. Digoxin is in the cardiac glycoside family of medications.

DOCETAXEL

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Docetaxel (DTX or DXL), sold under the brand name Taxotere among others, is a chemotherapy medication used to treat a number of types of cancer. This includes breast cancer, head and neck cancer, stomach cancer, prostate cancer and non-small-cell lung cancer. It may be used by itself or along with other chemotherapy medication. It is given by slow injection into a vein. Common side effects include hair loss, low blood cell counts, numbness, shortness of breath, vomiting, and muscle pains. Other severe side effects include allergic reactions and future cancers. Side effects are more common in people with liver problems. Use during pregnancy may harm the baby. Docetaxel is in the taxane family of medications. It works by disrupting the normal function of microtubules and thereby stopping cell division.

DOXORUBICIN

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Doxorubicin, sold under the trade names Adriamycin among others, is a chemotherapy medication used to treat cancer. This includes breast cancer, bladder cancer, Kaposi's sarcoma, lymphoma, and acute lymphocytic leukemia. It is often used together with other chemotherapy agents. Doxorubicin is given by injection into a vein. Common side effects include hair loss, bone marrow suppression, vomiting, rash, and inflammation of the mouth. Other serious side effects may include allergic reactions such as anaphylaxis, heart damage, tissue damage at the site of injection, radiation recall, and treatment-related leukemia. People often experience red discoloration of the urine for a few days. Doxorubicin is in the anthracycline and antitumor antibiotic family of medications. It works in part by interfering with the function of DNA.

EFAVIRENZ

Variant found:

- Gene: CYP2B6
- Marker: rs3745274
- Position: chr19:41512841
- Genotype: GT

Variant found:

- Gene: CYP2B6
- Marker: rs2279343
- Position: chr19:41515263
- Genotype: AG

Efavirenz (EFV), sold under the brand names Sustiva among others, is an antiretroviral medication used to treat and prevent HIV/AIDS. It is generally recommended for use with other antiretrovirals. It may be used for prevention after a needlestick injury or other potential exposure. It is sold both by itself and in combination as efavirenz/emtricitabine/tenofovir. It is taken by mouth once a day. Common side effects include rash, nausea, headache, feeling tired, and trouble sleeping. Some of the rashes may be serious such as Stevens-Johnson syndrome. Other serious side effects include depression, thoughts of suicide, liver problems, and seizures. It is not safe for use during pregnancy. It is a non-nucleoside reverse transcriptase inhibitor (NNRTI) and works by blocking the function of reverse transcriptase.

EPIRUBICIN

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Epirubicin is an anthracycline drug used for chemotherapy. It can be used in combination with other medications to treat breast cancer in patients who have had surgery to remove the tumor. It is marketed by Pfizer under the trade name Ellence in the US and Pharmorubicin or Epirubicin Ebewe elsewhere. Similarly to other anthracyclines, epirubicin acts by intercalating DNA strands. Intercalation results in complex formation which inhibits DNA and RNA synthesis. It also triggers DNA cleavage by topoisomerase II, resulting in mechanisms that lead to cell death. Binding to cell membranes and plasma proteins

may be involved in the compound's cytotoxic effects. Epirubicin also generates free radicals that cause cell and DNA damage. Epirubicin is favoured over doxorubicin, the most popular anthracycline, in some chemotherapy regimens as it appears to cause fewer side-effects. Epirubicin has a different spatial orientation of the hydroxyl group at the 4' carbon of the sugar - it has the opposite chirality - which may account for its faster elimination and reduced toxicity. Epirubicin is primarily used against breast and ovarian cancer, gastric cancer, lung cancer and lymphomas.

EXEMESTANE

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Exemestane, sold under the brand name Aromasin among others, is a medication used to treat breast cancer. It is a member of the class of antiestrogens known as aromatase inhibitors. Some breast cancers require estrogen to grow. Those cancers have estrogen receptors (ERs), and are called ER-positive. They may also be called estrogen-responsive, hormonally-responsive, or hormone-receptor-positive. Aromatase is an enzyme that synthesizes estrogen. Aromatase inhibitors block the synthesis of estrogen. This lowers the estrogen level, and slows the growth of cancers.

FENTANYL

Variant found:

- Gene: intergenic_non-coding
- Marker: rs2952768
- Position: chr2:208494234
- Genotype: CC

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Fentanyl, also spelled fentanil, is an opioid used as a pain medication and together with other medications for anesthesia. Fentanyl is also made illegally and used as a recreational drug, often mixed with heroin or cocaine. It has a rapid onset and effects generally last less than two hours. Medically, fentanyl is used by injection, as a patch on the skin, as a nasal spray, or in the mouth. Common side effects include vomiting, constipation, sedation, confusion, hallucinations, and injuries related to poor coordination. Serious side effects may include decreased breathing (respiratory depression), serotonin syndrome, low blood pressure, addiction, or coma. In 2016, more than 20,000 deaths occurred in the United States due to overdoses of fentanyl and analogues, half of all reported opioid-related deaths. Fentanyl works primarily by activating μ -opioid receptors. It is around 100 times stronger than morphine, and some analogues such as carfentanil are around 10,000 times stronger.

FLUOROURACIL

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Fluorouracil (5-FU), sold under the brand name Adrucil among others, is a medication used to treat cancer. By injection into a vein it is used for colon cancer, esophageal cancer, stomach cancer, pancreatic cancer, breast cancer, and cervical cancer. As a cream it is used for actinic keratosis, basal cell carcinoma, and skin warts. When used by injection most people develop side effects. Common side effects include inflammation of the mouth, loss of appetite, low blood cell counts, hair loss, and inflammation of the skin. When used as a cream, irritation at the site of application may occur. Use of either form in pregnancy may harm the baby. Fluorouracil is in the antimetabolite and pyrimidine analog families of medications. How it works is not entirely clear but believed to involve blocking the action of thymidylate synthase and thus stopping the production of DNA.

FLUOXETINE

Variant found:

- Gene: FKBP5
- Marker: rs4713916
- Position: chr6:35669983
- Genotype: AG

Fluoxetine, sold under the brand names Prozac and Sarafem among others, is an antidepressant of the selective serotonin reuptake inhibitor (SSRI) class. It is used for the treatment of major depressive disorder, obsessive-compulsive disorder (OCD), bulimia nervosa, panic disorder, and premenstrual dysphoric disorder. It may decrease the risk of suicide in those over the age of 65. It has also been used to treat premature ejaculation. Fluoxetine is taken by mouth. Common side effects include trouble sleeping, sexual dysfunction, loss of appetite, dry mouth, rash, and abnormal dreams. Serious side effects include serotonin syndrome, mania, seizures, an increased risk of suicidal behavior in people under 25 years old, and an increased risk of bleeding. If stopped suddenly, a withdrawal syndrome may occur with anxiety, dizziness, and changes in sensation. It is unclear if it is safe in pregnancy. If already on the medication, it may be reasonable to continue during breastfeeding. Its mechanism of action is not entirely clear but believed to be related to increasing serotonin activity in the brain.

FUROSEMIDE

Variant found:

- Gene: ADD1
- Marker: rs4961
- Position: chr4:2906707
- Genotype: GT

Furosemide, sold under the brand name Lasix among others, is a medication used to treat fluid build-up due to heart failure, liver scarring, or kidney disease. It may also be used for the treatment of high blood pressure. It can be taken by injection into a vein or by mouth. When taken by mouth, it typically begins working within an hour, while intravenously, it typically begins working within five minutes. Common side effects include feeling lightheaded with standing, ringing in the ears, and sensitivity to light. Potentially serious side effects include electrolyte abnormalities, low blood pressure, and hearing loss. Blood tests are recommended regularly for those on treatment. Furosemide is a type of loop diuretic that works by decreasing the reabsorption of sodium by the kidneys.

GLIBENCLAMIDE

Variant found:

- **Gene:** KCNJ11
- **Marker:** rs5219
- **Position:** chr11:17409572
- **Genotype:** CC

Glibenclamide, also known as glyburide, is an antidiabetic drug in a class of medications known as sulfonylureas, closely related to sulfonamide antibiotics. It was developed in 1966 in a cooperative study between Boehringer Mannheim (now part of Roche) and Hoechst (now part of Sanofi-Aventis). It is used in the treatment of type 2 diabetes. It is not as good as either metformin or insulin in those who have gestational diabetes. This medication is a major cause of medication-induced hypoglycemia. The risk is greater than with other sulfonylureas. Cholestatic jaundice is noted. Glibenclamide may be not recommended in those with G6PD deficiency, as it may cause acute hemolysis. The medication works by binding to and inhibiting the ATP-sensitive potassium channels (KATP) inhibitory regulatory subunit sulfonylurea receptor 1 (SUR1) in pancreatic beta cells. This inhibition causes cell membrane depolarization, opening voltage-dependent calcium channels. This results in an increase in intracellular calcium in the pancreatic beta cell and subsequent stimulation of insulin release. After a cerebral ischemic insult, the blood-brain barrier is broken and glibenclamide can reach the central nervous system. Glibenclamide has been shown to bind more efficiently to the ischemic hemisphere. Moreover, under ischemic conditions SUR1, the regulatory subunit of the KATP- and the NCCa-ATP-channels, is expressed in neurons, astrocytes, oligodendrocytes, endothelial cells and by reactive microglia.

GLICLAZIDE

Variant found:

- **Gene:** KCNJ11
- **Marker:** rs5219
- **Position:** chr11:17409572
- **Genotype:** CC

Gliclazide, sold under the brand name Diamicon among others, is an anti-diabetic medication used to treat diabetes mellitus type 2. It is used when dietary changes, exercise, and weight loss are not enough. It is taken by mouth. Side effect may include low blood sugar, vomiting, abdominal pain, rash, and liver problems. Use by those with significant kidney problems, liver problems, or who are pregnant is not recommended. Gliclazide is in the sulfonylurea family of medications. It works mostly by increasing the release of insulin.

GLIMEPIRIDE

Variant found:

- Gene: KCNJ11
- Marker: rs5219
- Position: chr11:17409572
- Genotype: CC

Glimepiride, sold under the trade name Amaryl among others, is a medication used to treat diabetes mellitus type 2. It is less preferred than metformin. Use is recommended together with diet and exercise. It is taken by mouth. Glimepiride takes up to three hours for maximum effect and lasts for about a day. Common side effects include headache, nausea, and dizziness. Serious side effects may include low blood sugar. Use in during pregnancy and breastfeeding is not recommended. It works mainly by increasing the amount of insulin released from the pancreas. It is classified as a second-generation sulfonylurea.

GLIPIZIDE

Variant found:

- Gene: KCNJ11
- Marker: rs5219
- Position: chr11:17409572
- Genotype: CC

Glipizide, sold under the trade name Glucotrol among others, is an anti-diabetic medication of the sulfonylurea class used to treat type 2 diabetes. It is used together with a diabetic diet. It is not indicated for use by itself in type 1 diabetes. It is taken by mouth. Effects generally begin within half an hour and can last for up to a day. Common side effects include nausea, diarrhea, low blood sugar, and headache. Other side effects include sleepiness, skin rash, and shakiness. The dose may need to be adjusted in those with liver or kidney disease. Use during pregnancy or breastfeeding is not recommended. It works by stimulating the pancreas to release insulin and increases tissue sensitivity to insulin.

HALOPERIDOL

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Haloperidol, marketed under the trade name Haldol among others, is a typical antipsychotic medication. Haloperidol is used in the treatment of schizophrenia, tics in Tourette syndrome, mania in bipolar disorder, nausea and vomiting, delirium, agitation, acute psychosis, and hallucinations in alcohol withdrawal. It may be used by mouth, as an injection into a muscle, or intravenously. Haloperidol typically works within thirty to sixty minutes. A long-acting formulation may be used as an injection every four weeks in people with schizophrenia or related illnesses, who either forget or refuse to take the medication by mouth. Haloperidol may result in a movement disorder known as tardive dyskinesia which may be permanent. Neuroleptic malignant syndrome and QT interval prolongation may occur. In older people with psychosis due to dementia it results in an increased risk of death. When taken during pregnancy it may result in problems in the infant. It should not be used in people with Parkinson's disease.

LATANOPROST

Variant found:

- Gene: PTGFR
- Marker: rs3753380
- Position: chr1:78956432
- Genotype: CC

Latanoprost, sold under the brand name Xalatan among others, is a medication used to treat increased pressure inside the eye. This includes ocular hypertension and open angle glaucoma. It is applied as eye drops to the eyes. Onset of effects is usually within four hours, and they last for up to a day. Common side effects include blurry vision, redness of the eye, itchiness, and darkening of the iris. Latanoprost is in the prostaglandin analogue family of medication. It works by increasing the outflow of aqueous fluid from the eyes through the uveoscleral tract.

LORAZEPAM

Variant found:

- Gene: UGT2B15
- Marker: rs1902023
- Position: chr4:69536084
- Genotype: CC

Lorazepam, sold under the brand name Ativan among others, is a benzodiazepine medication. It is used to treat anxiety disorders, trouble sleeping, active seizures including status epilepticus, alcohol withdrawal, and chemotherapy-induced nausea and vomiting. It is also used during surgery to interfere with memory formation and to sedate those who are being mechanically ventilated. While it can be used for severe agitation, midazolam is usually preferred. It is also used, along with other treatments, for acute coronary syndrome due to cocaine use. It can be given by mouth or as an injection into a muscle or vein. When given by injection onset of effects is between one and thirty minutes and effects last for up to a day. Common side effects include weakness, sleepiness, low blood pressure, and a decreased effort to breathe. When given intravenously the person should be closely monitored. Among those who are depressed there may be an increased risk of suicide. With long-term use, larger doses may be required for the same effect. Physical dependence and psychological dependence may also occur. If stopped suddenly after long-term use, benzodiazepine withdrawal syndrome may occur. Older people more often develop adverse effects. In this age group lorazepam is associated with falls and hip fractures. Due to these concerns, lorazepam use is generally only recommended for up to two to four weeks.

METHADONE

Variant found:

- Gene: CYP2B6
- Marker: rs3745274
- Position: chr19:41512841
- Genotype: GT

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Methadone, sold under the brand name Dolophine among others, is an opioid used for opioid maintenance therapy in opioid dependence, and for pain. Detoxification using methadone can either be done relatively rapidly in less than a month or gradually over as long as six months. While a single dose has a rapid effect, maximum effect can take five days of use. The pain relieving effects last about six hours after a single dose, similar to morphine's. After long term use, in people with normal liver function, effects last 8 to 36 hours. Methadone is usually taken by mouth and rarely by injection into a muscle or vein. Side effects are similar to those of other opioids. Commonly these include dizziness, sleepiness, vomiting, and sweating. Serious risks include opioid abuse and a decreased effort to breathe. Abnormal heart rhythms may also occur due to a prolonged QT interval. The number of deaths in the United States involving methadone poisoning declined from 4,418 in 2011 to 3,300 in 2015. Risks are greater with higher doses. Methadone is made by chemical synthesis and acts on opioid receptors.

MIRTAZAPINE

Variant found:

- Gene: FKBP5
- Marker: rs4713916
- Position: chr6:35669983
- Genotype: AG

Mirtazapine, sold under the brand name Remeron among others, is an antidepressant primarily used to treat depression. Its full effect may take more than four weeks to occur, with some benefit possibly as early as one to two weeks. Often it is used in depression complicated by anxiety or trouble sleeping. It is taken by mouth. Common side effects include increased weight, sleepiness, and dizziness. Serious side effects may include an increased suicide among children, mania, and low white blood count. Withdrawal symptoms may occur with stopping. It is not recommended together with an MAO inhibitor. It is unclear if use during pregnancy is safe. How it works is not clear but may involve blocking certain adrenergic and serotonin receptors. Chemically, it is a tetracyclic antidepressant (TeCA). It also has strong antihistamine effects.

MORPHINE

Variant found:

- Gene: Intergenic_non-coding
- Marker: rs2952768
- Position: chr2:208494234
- Genotype: CC

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Morphine is a pain medication of the opiate family which is found naturally in a number of plants and animals. It acts directly on the central nervous system (CNS) to decrease the feeling of pain. It can be taken for both acute pain and chronic pain. It is frequently used for pain from myocardial infarction and during labor. It can be given by mouth, by injection into a muscle, by injection under the skin, intravenously, injection into the space around the spinal cord, or rectally. Maximum effect is reached after about 20 minutes when given intravenously and after 60 minutes when given by mouth, while duration of effect is 3–7 hours. Long-acting formulations also exist. Potentially serious side effects include decreased respiratory effort and low blood pressure. Morphine is addictive and prone to abuse. If the dose is reduced after long-term use, opioid withdrawal symptoms may occur. Common side effects include drowsiness, vomiting, and constipation. Caution is advised when used during pregnancy or breast feeding, as morphine may affect the baby. Morphine was first isolated between 1803 and 1805 by Friedrich Sertürner. This is generally believed to be the first isolation of an active ingredient from a plant. Merck began marketing it commercially in 1827. Morphine was more widely used after the invention of the hypodermic syringe in 1853–1855. Sertürner originally named the substance morphium after the Greek god of dreams, Morpheus, as it has a tendency to cause sleep.

NEVIRAPINE

Variant found:

- Gene: CYP2B6
- Marker: rs3745274
- Position: chr19:41512841
- Genotype: GT

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Variant found:

- Gene: CCHCR1
- Marker: rs746647
- Position: chr6:31114182
- Genotype: AG

Nevirapine (NVP), marketed under the trade name Viramune among others, is a medication used to treat and prevent HIV/AIDS, specifically HIV-1. It is generally recommended for use with other antiretroviral medication. It may be used to prevent mother to child spread during birth but is not recommended following other exposures. It is taken by mouth. Common side effects include rash, headache, nausea, feeling tired, and liver problems. The liver problems and skin rash may be severe and should be checked for during the first few months of treatment. It appears to be safe for use during pregnancy. It is a non-nucleoside reverse transcriptase inhibitor (NNRTI) and works by blocking the function of reverse transcriptase.

OLANZAPINE

Variant found:

- Gene: MC4R

- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Olanzapine, sold under the trade name Zyprexa among others, is an atypical antipsychotic primarily used to treat schizophrenia and bipolar disorder. For schizophrenia, it can be used for both new onset disease and long term maintenance. It is taken by mouth or by injection into a muscle. Common side effect include weight gain, movement disorders, dizziness, feeling tired, constipation, and dry mouth. Other side effects include low blood pressure with standing, allergic reactions, neuroleptic malignant syndrome, high blood sugar, seizures, gynecomastia, and tardive dyskinesia. In older people with dementia, its use increases the risk of death. Use in the later part of pregnancy may result in a movement disorder in the baby for some time after birth. Although how it works is not entirely clear, it blocks dopamine and serotonin receptors. It is classed as an atypical antipsychotic.

ONDANSETRON

Variant found:

- Gene: ABCB1
- Marker: rs2032582
- Position: chr7:87160618
- Genotype: CC

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Ondansetron, marketed under the brand name Zofran, is a medication used to prevent nausea and vomiting caused by cancer chemotherapy, radiation therapy, or surgery. It is also useful in gastroenteritis. It has little effect on vomiting caused by motion sickness. It can be given by mouth, or by injection into a muscle or into a vein. Common side effects include diarrhea, constipation, headache, sleepiness, and itchiness. Serious side effects include QT prolongation and severe allergic reaction. It appears to be safe during pregnancy but has not been well studied in this group. It is a serotonin 5-HT₃ receptor antagonist. It does not have any effect on dopamine receptors or muscarinic receptors.

OXALIPLATIN

Variant found:

- Gene: XRCC1
- Marker: rs25487
- Position: chr19:44055726
- Genotype: CC

Oxaliplatin, sold under the brand name Eloxatin, is a cancer medication used to treat colorectal cancer. Often it is used together with fluorouracil and folinic acid (leucovorin) in advanced cancer. It is given by injection into a vein. Common side effects include numbness, feeling tired, nausea, diarrhea, and low blood cell counts. Other serious side effects include allergic reactions. Use in pregnancy is known to harm the baby. Oxaliplatin is in the platinum-based antineoplastic family of medications. It is believed to work by blocking the duplication of DNA.

OXAZEPAM

Variant found:

- Gene: UGT2B15
- Marker: rs1902023
- Position: chr4:69536084
- Genotype: CC

Oxazepam is a short-to-intermediate-acting benzodiazepine. Oxazepam is used for the treatment of anxiety and insomnia and in the control of symptoms of alcohol withdrawal syndrome. It is a metabolite of diazepam, prazepam, and temazepam, and has moderate amnesic, anxiolytic, anticonvulsant, hypnotic, sedative, and skeletal muscle relaxant properties compared to other benzodiazepines.

OXYCODONE

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Oxycodone, sold under brand name OxyContin among others, is an opioid medication used for treatment of moderate to severe pain. It is usually taken by mouth, and is available in immediate release and controlled release formulations. Onset of pain relief typically begins within 15 minutes and lasts for up to six hours with the immediate release formulation. In the United Kingdom, it is available by injection. Combination products are also available with paracetamol (acetaminophen) or aspirin. Common side effects include constipation, nausea, sleepiness, dizziness, itching, dry mouth, and sweating. Severe side effects may include addiction, respiratory depression (a decreased effort to breathe), and low blood pressure. Those allergic to codeine may also be allergic to oxycodone. Use of oxycodone in early pregnancy appears relatively safe. Opioid withdrawal may occur if rapidly stopped. Oxycodone acts by activating the μ -opioid receptor. When taken by mouth, it has roughly 1.5 times the effect of the equivalent amount of morphine.

PACLITAXEL

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Paclitaxel (PTX), sold under the brand name Taxol among others, is a chemotherapy medication used to treat a number of types of cancer. This includes ovarian cancer, breast cancer, lung cancer, Kaposi sarcoma, cervical cancer, and pancreatic cancer. It is given by injection into a vein. There is also an albumin-bound formulation. Common side effects include hair loss, bone marrow suppression, numbness, allergic reactions, muscle pains, and diarrhea. Other serious side effects include heart problems, increased risk of infection, and lung inflammation. There are concerns that use during pregnancy may cause birth defects. Paclitaxel is in the taxane family of medications. It works by interference with the normal function of microtubules during cell division.

PALIPERIDONE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Paliperidone, sold under the brand name Invega among others, is an atypical antipsychotic. It is marketed by Janssen Pharmaceutica. Invega is an extended release formulation of paliperidone that uses the OROS extended release system to allow for once-daily dosing. Paliperidone is mainly used to treat schizophrenia and schizoaffective disorder. Paliperidone palmitate is a long-acting injectable formulation of paliperidone palmitoyl ester indicated for once-every 28 days injection after an initial titration period.

PAROXETINE

Variant found:

- Gene: HTR1A
- Marker: rs6295
- Position: chr5:63258565
- Genotype: CG

Variant found:

- Gene: FKBP5
- Marker: rs4713916
- Position: chr6:35669983
- Genotype: AG

Paroxetine, sold under the brand names Paxil and Seroxat among others, is an antidepressant of the selective serotonin reuptake inhibitor (SSRI) class. It is used to treat major depressive disorder, obsessive-compulsive disorder, panic disorder, social anxiety disorder, posttraumatic stress disorder, generalized anxiety disorder and premenstrual dysphoric disorder. It has also been used in the treatment of hot flashes due to menopause and premature ejaculation. It is taken by mouth. Common side effects include drowsiness, dry mouth, loss of appetite, sweating, trouble sleeping, and sexual dysfunction. Serious side effects may include suicide in those under the age of 25, serotonin syndrome, and mania. While rate of side effects appear similar compared to other SSRIs and SNRIs, antidepressant discontinuation syndromes may occur more often. Use in pregnancy is not recommended while use during breastfeeding is relatively safe. It believed to work by blocking the re-uptake of the chemical serotonin by neurons in the brain.

PEGINTERFERON ALFA-2A

Variant found:

- Gene: IFNL3, IFNL4
- Marker: rs12979860
- Position: chr19:39738787
- Genotype: CT

Variant found:

- Gene: IFNL3
- Marker: rs11881222
- Position: chr19:39734923
- Genotype: AG

Pegylated interferon alfa-2a, sold under the brand name Pegasys among others, is medication used to treat hepatitis C and hepatitis B. For hepatitis C it is typically used together with ribavirin and cure rates are between 24 and 92%. For hepatitis B it may be used alone. It is given by injection under the skin. Side effects are common. They may include headache, feeling tired, depression, trouble sleeping, hair loss, nausea, pain at the site of injection, and fever. Severe side effects may include psychosis, autoimmune disorders, blood clots, or infections. Use with ribavirin is not recommended during pregnancy. Pegylated interferon alfa-2a is in the alpha interferon family of medications. It is pegylated to protect the molecule from breakdown.

PEGINTERFERON ALFA-2B

Variant found:

- Gene: VDR
- Marker: rs2228570
- Position: chr12:48272895
- Genotype: GG

Variant found:

- Gene: IFNL3, IFNL4
- Marker: rs12979860
- Position: chr19:39738787
- Genotype: CT

Variant found:

- Gene: IFNL3
- Marker: rs11881222
- Position: chr19:39734923
- Genotype: AG

Pegylated interferon alfa-2b, sold under the brand name PegIntron among others, is a medication used to treat hepatitis C and melanoma. For hepatitis C it is typically used with ribavirin and cure rates are between 33 and 82%. For melanoma it is used in addition to surgery. It is given by injection under the skin. Side effects are common. They may include headache, feeling tired, mood changes, trouble sleeping, hair loss, nausea, pain at the site of injection, and fever. Severe side effects may include psychosis, liver problems, blood clots, infections, or an irregular heartbeat. Use with ribavirin is not recommended during pregnancy. Pegylated interferon alfa-2b is in the alpha interferon family of medications. It is pegylated to protect the molecule from breakdown.

PLATINUM BASED THERAPIES

Variant found:

- Gene: XRCC1
- Marker: rs25487
- Position: chr19:44055726
- Genotype: CC

Platinum is a chemical element with symbol Pt and atomic number 78. It is a dense, malleable, ductile, highly unreactive, precious, silverish-white transition metal. Its name is derived from the Spanish term platino, meaning "little silver". Compounds containing platinum, such as cisplatin, oxaliplatin and carboplatin, are applied in chemotherapy against certain types of cancer.

PRAVASTATIN

Variant found:

- Gene: KIF6
- Marker: rs20455
- Position: chr6:39325078
- Genotype: GG

Pravastatin, sold under the brand name Pravachol among others, is a statin medication, used preventing cardiovascular disease in those at high risk and treating abnormal lipids. It should be used together with diet changes, exercise, and weight loss. It is taken by mouth. Common side effects include joint pain, diarrhea, nausea, headaches, and muscle pains. Serious side effects may include rhabdomyolysis, liver problems, and diabetes. Use during pregnancy may harm the baby. Like all statins, pravastatin works by inhibiting HMG-CoA reductase, an enzyme found in liver that plays a role in producing cholesterol.

QUETIAPINE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Quetiapine, sold under the trade name Seroquel among others, is an atypical antipsychotic used for the treatment of schizophrenia, bipolar disorder, and major depressive disorder. It is also used as a sleep aid due to its sedating effect, but this use is not recommended. It is taken by mouth. Common side effects include sleepiness, constipation, weight gain, and dry mouth. Other side effects include low blood pressure with standing, seizures, a prolonged erection, high blood sugar, tardive dyskinesia, and neuroleptic malignant syndrome. In older people with dementia, its use increases the risk of death. Use in the third trimester of pregnancy may result in a movement disorder in the baby for some time after birth. Quetiapine is believed to work by blocking a number of receptors including serotonin and dopamine.

RADIOTHERAPY

Variant found:

- Gene: TANC1
- Marker: rs10497203
- Position: chr2:159851423
- Genotype: AC

Variant found:

- Gene: TANC1
- Marker: rs264631
- Position: chr2:159950865
- Genotype: GG

Variant found:

- Gene: TANC1
- Marker: rs264651
- Position: chr2:159929431
- Genotype: AG

Variant found:

- Gene: TANC1
- Marker: rs6432512
- Position: chr2:159899913
- Genotype: TT

Variant found:

- Gene: TANC1
- Marker: rs264588
- Position: chr2:159936391
- Genotype: AA

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Variant found:

- Gene: TANC1
- Marker: rs7582141
- Position: chr2:159899489
- Genotype: TT

Radiation therapy or radiotherapy, often abbreviated RT, RTx, or XRT, is therapy using ionizing radiation, generally as part of cancer treatment to control or kill malignant cells and normally delivered by a linear accelerator. Radiation therapy may be curative in a number of types of cancer if they are localized to one area of the body. It may also be used as part of adjuvant therapy, to prevent tumor recurrence after surgery to remove a primary malignant tumor (for example, early stages of breast cancer). Radiation therapy is synergistic with chemotherapy, and has been used before, during, and after chemotherapy in susceptible cancers. The subspecialty of oncology concerned with radiotherapy is called radiation oncology. Radiation therapy is commonly applied to the cancerous tumor because of its ability to control cell growth. Ionizing radiation works by damaging the DNA of cancerous tissue leading to cellular death. To spare normal tissues (such as skin or organs which radiation must pass through to treat the tumor), shaped radiation beams are aimed from several angles of exposure to intersect at the tumor, providing a much larger absorbed dose there than in the surrounding, healthy tissue. Besides the tumour itself, the radiation fields may also include the draining lymph nodes if they are clinically or radiologically involved with tumor, or if there is thought to be a risk of subclinical malignant spread. It is necessary to include a margin of normal tissue around the tumor to allow for uncertainties in daily set-up and internal tumor motion. These uncertainties can be caused by internal movement (for example, respiration and bladder filling) and movement of external skin marks relative to the tumor position. Radiation oncology is the medical specialty concerned with prescribing radiation, and is distinct from radiology, the use of radiation in medical imaging and diagnosis. Radiation may be prescribed by a radiation oncologist with intent to cure ("curative") or for adjuvant therapy. It may also be used as palliative treatment (where cure is not possible and the aim is for local disease control or symptomatic relief) or as therapeutic treatment (where the therapy has survival benefit and it can be curative). It is also common to combine radiation therapy with surgery, chemotherapy, hormone therapy, immunotherapy or some mixture of the four. Most common cancer types can be treated with radiation therapy in some way. The precise treatment intent (curative, adjuvant, neoadjuvant therapeutic, or palliative) will depend on the tumor type, location, and stage, as well as the general health of the patient. Total body irradiation (TBI) is a radiation therapy technique used to prepare the body to receive a bone marrow transplant. Brachytherapy, in which a radioactive source is placed inside or next to the area requiring treatment, is another form of radiation therapy that minimizes exposure to healthy tissue during procedures to treat cancers of the breast, prostate and other organs. Radiation therapy has several applications in non-malignant conditions, such as the treatment of trigeminal neuralgia, acoustic neuromas, severe thyroid eye disease, pterygium, pigmented villonodular synovitis, and prevention of keloid scar growth, vascular restenosis, and heterotopic ossification. The use of radiation therapy in non-malignant conditions is limited partly by worries about the risk of radiation-induced cancers.

RIBAVIRIN

Variant found:

- Gene: VDR
- Marker: rs2228570
- Position: chr12:48272895
- Genotype: GG

Variant found:

- Gene: IFNL3, IFNL4
- Marker: rs12979860
- Position: chr19:39738787
- Genotype: CT

Variant found:

- Gene: IFNL3

- **Marker:** rs11881222
- **Position:** chr19:39734923
- **Genotype:** AG

Ribavirin, also known as tribavirin, is an antiviral medication used to treat RSV infection, hepatitis C, and viral hemorrhagic fever. For hepatitis C, it is used in combination with other medications such as simeprevir, sofosbuvir, peginterferon alfa-2b or peginterferon alfa-2a. Among the viral hemorrhagic fevers it is used for Lassa fever, Crimean-Congo hemorrhagic fever, and Hantavirus infection but not Ebola or Marburg. Ribavirin is taken by mouth or inhaled. Common side effects include feeling tired, headache, nausea, fever, muscle pains, and an irritable mood. Serious side effects include red blood cell breakdown, liver problems, and allergic reactions. Use during pregnancy results in harm to the baby. Effective birth control is recommended for both males and females for at least 7 months during and after use. The mechanism of action of ribavirin is not entirely clear.

RISPERIDONE

Variant found:

- **Gene:** MC4R
- **Marker:** rs489693
- **Position:** chr18:57882787
- **Genotype:** AA

Risperidone, sold under the brand name Risperdal among others, is an antipsychotic. It is used to treat schizophrenia, bipolar disorder, and irritability associated with autism. It is taken either by mouth or by injection into a muscle. The injectable version is long-acting and lasts for about two weeks. Common side effects include movement problems, sleepiness, dizziness, trouble seeing, constipation, and increased weight. Serious side effects may include the potentially permanent movement disorder tardive dyskinesia, as well as neuroleptic malignant syndrome, an increased risk of suicide, and high blood sugar levels. In older people with psychosis as a result of dementia, it may increase the risk of dying. It is unclear if it is safe for use in pregnancy. Risperidone is an atypical antipsychotic. Its mechanism of action is not entirely clear, but is believed to be related to its action as a dopamine antagonist and serotonin antagonist.

RITUXIMAB

Variant found:

- **Gene:** FCGR3A
- **Marker:** rs396991
- **Position:** chr1:161514542
- **Genotype:** AC

Rituximab, sold under the brand name Rituxan among others, is a medication used to treat certain autoimmune diseases and types of cancer. It is used for non-Hodgkin's lymphoma, chronic lymphocytic leukemia, rheumatoid arthritis, granulomatosis with polyangiitis, idiopathic thrombocytopenic purpura, pemphigus vulgaris, myasthenia gravis and Epstein-Barr virus-positive mucocutaneous ulcers. It is given by slow injection into a vein. Common side effects, which often occur within two hours of the medication being given, include rash, itchiness, low blood pressure, and shortness of breath. Other severe side effects include reactivation of hepatitis B in those previously infected, progressive multifocal leukoencephalopathy, and toxic epidermal necrolysis. It is

unclear if use during pregnancy is safe for the baby. Rituximab is a chimeric monoclonal antibody against the protein CD20, which is primarily found on the surface of immune system B cells. When it binds to this protein it triggers cell death.

SALBUTAMOL

Variant found:

- Gene: CRHR2
- Marker: rs7793837
- Position: chr7:30726777
- Genotype: AT

Variant found:

- Gene: ADRB2
- Marker: rs1042713
- Position: chr5:148206440
- Genotype: AA

Salbutamol, also known as albuterol and marketed as Ventolin among other brand names, is a medication that opens up the medium and large airways in the lungs. It is used to treat asthma, including asthma attacks, exercise-induced bronchoconstriction, and chronic obstructive pulmonary disease (COPD). It may also be used to treat high blood potassium levels. Salbutamol is usually used with an inhaler or nebulizer, but it is also available as a pill and intravenous solution. Onset of action of the inhaled version is typically within 15 minutes and lasts for two to six hours. Common side effects include shakiness, headache, fast heart rate, dizziness, and feeling anxious. Serious side effects may include worsening bronchospasm, irregular heartbeat, and low blood potassium levels. It can be used during pregnancy and breastfeeding, but safety is not entirely clear. It is a short-acting β_2 adrenergic receptor agonist which works by causing relaxation of airway smooth muscle.

SALMETEROL

Variant found:

- Gene: ADRB2
- Marker: rs1042713
- Position: chr5:148206440
- Genotype: AA

Salmeterol is a long-acting β_2 adrenergic receptor agonist (LABA) used in the maintenance and prevention of asthma symptoms and maintenance of chronic obstructive pulmonary disease (COPD) symptoms. Symptoms of bronchospasm include shortness of breath, wheezing, coughing and chest tightness. It is also used to prevent breathing difficulties during exercise (exercise-induced bronchoconstriction). It was patented in 1983 and came into medical use in 1990. It is marketed as Serevent in the US. It is available as a dry powder inhaler that releases a powdered form of the drug. It was previously available as a metered-dose inhaler (MDI) but was discontinued in the US in 2002. It is still available as an MDI in the UK as of 2013.

SILDENAFIL

Variant found:

- Gene: GNB3
- Marker: rs5443
- Position: chr12:6954875
- Genotype: TT

Sildenafil, sold as the brand name Viagra among others, is a medication used to treat erectile dysfunction and pulmonary arterial hypertension. It is unclear if it is effective for treating sexual dysfunction in women. It is taken by mouth or injection into a vein. Onset is typically within 20 minutes and lasts for about 2 hours. Common side effects include headaches, heartburn, and flushed skin. Caution is advised in those with cardiovascular disease. Rare but serious side effects include a prolonged erection that can lead to damage to the penis, vision problems, and hearing loss. Sildenafil should not be taken by people on nitrates such as nitroglycerin (glycerin trinitrate), as this may result in a serious drop in blood pressure. Sildenafil acts by blocking phosphodiesterase 5 (PDE5), an enzyme that promotes breakdown of cGMP, which regulates blood flow in the penis. It requires sexual arousal, however, to work. It also results in dilation of the blood vessels in the lungs.

SIMVASTATIN

Variant found:

- Gene: ABCB1
- Marker: rs2032582
- Position: chr7:87160618
- Genotype: CC

Simvastatin, marketed under the trade name Zocor among others, is a lipid-lowering medication. It is used along with exercise, diet, and weight loss to decrease elevated lipid levels. It is also used to decrease the risk of heart problems in those at high risk. It is taken by mouth. Common side effects include constipation, headaches, and nausea. Serious side effects may include muscle breakdown, liver problems, and increased blood sugar levels. A lower dose may be needed in people with kidney problems. There is evidence of harm to the developing baby when taken during pregnancy and it should not be used by those who are breastfeeding. It is in the statin class of medications and works by decreasing the manufacture of cholesterol by the liver.

SIROLIMUS

Variant found:

- Gene: CYP3A5
- Marker: rs776746
- Position: chr7:99270539
- Genotype: TT

Sirolimus, also known as rapamycin, is a macrolide compound that is used to coat coronary stents, prevent organ transplant rejection and to treat a rare lung disease called lymphangioleiomyomatosis. It has immunosuppressant functions in humans and is especially useful in preventing the rejection of kidney transplants. It inhibits activation of T cells and B cells by reducing their sensitivity to interleukin-2 (IL-2) through mTOR inhibition. It is produced by the bacterium *Streptomyces hygroscopicus* and was isolated for the first time in 1972 by Surendra Nath Sehgal and colleagues from samples of *Streptomyces*.

hygroscopicus found on Easter Island. The compound was originally named rapamycin after the native name of the island, Rapa Nui. Sirolimus was initially developed as an antifungal agent. However, this use was abandoned when it was discovered to have potent immunosuppressive and antiproliferative properties due to its ability to inhibit mTOR. It was approved by the US Food and Drug Administration in September 1999 and is marketed under the trade name Rapamune by Pfizer (formerly by Wyeth).

SPIRONOLACTONE

Variant found:

- Gene: ADD1
- Marker: rs4961
- Position: chr4:2906707
- Genotype: GT

Spironolactone, sold under the brand name Aldactone among others, is a medication that is primarily used to treat fluid build-up due to heart failure, liver scarring, or kidney disease. It is also used in the treatment of high blood pressure, low blood potassium that does not improve with supplementation, early puberty in boys, acne and excessive hair growth in women, and as a part of feminizing hormone therapy in transgender women. Spironolactone is taken by mouth. Common side effects include electrolyte abnormalities, particularly high blood potassium, nausea, vomiting, headache, rashes, and a decreased desire for sex. In those with liver or kidney problems, extra care should be taken. Spironolactone has not been well studied in pregnancy and should not be used to treat high blood pressure of pregnancy. It is a steroid that blocks the effects of the hormones aldosterone and testosterone and has some estrogen-like effects. Spironolactone belongs to a class of medications known as potassium-sparing diuretics.

TACROLIMUS

Variant found:

- Gene: CYP3A5
- Marker: rs776746
- Position: chr7:99270539
- Genotype: TT

Variant found:

- Gene: CYP3A5
- Marker: rs776746
- Position: chr7:99270539
- Genotype: TT

Tacrolimus, also known as fujimycin or FK506, is an immunosuppressive drug used mainly after allogeneic organ transplant to lower the risk of organ rejection. It achieves this by inhibiting the production of interleukin-2, a molecule that promotes the development and proliferation of T cells, which are vital to the body's learned (or adaptive) immune response. Tacrolimus is also used in the treatment of other T cell-mediated diseases such as eczema (for which it is applied to the skin in a medicated ointment), severe refractory uveitis after bone marrow transplants, exacerbations of minimal change disease, Kimura's disease, and the skin condition vitiligo. Chemically it is a 23-membered macrolide lactone that was first discovered in 1987 from the fermentation broth of a Japanese soil sample that contained the bacterium *Streptomyces tsukubaensis*. Tacrolimus is also used to treat dry eye syndrome in cats and dogs.

TAMOXIFEN

Variant found:

- Gene: CYP19A1
- Marker: rs4646
- Position: chr15:51502844
- Genotype: AC

Tamoxifen, sold under the brand name Nolvadex among others, is a medication that is used to prevent breast cancer in women and treat breast cancer in women and men. It is also being studied for other types of cancer. It has been used for Albright syndrome. Tamoxifen is typically taken daily by mouth for five years for breast cancer. Serious side effects include a small increased risk of uterine cancer, stroke, vision problems, and pulmonary embolism. Common side effects include irregular periods, weight loss, and hot flashes. It may cause harm to the baby if taken during pregnancy or breastfeeding. It is a selective estrogen-receptor modulator (SERM) and works by decreasing the growth of breast cancer cells. It is of the triphenylethylene group.

TELAPREVIR

Variant found:

- Gene: IFNL3, IFNL4
- Marker: rs12979860
- Position: chr19:39738787
- Genotype: CT

Telaprevir (VX-950), marketed under the brand names Incivek and Incivo, is a pharmaceutical drug for the treatment of hepatitis C co-developed by Vertex Pharmaceuticals and Johnson & Johnson. It is a member of a class of antiviral drugs known as protease inhibitors. Specifically, telaprevir inhibits the hepatitis C viral enzyme NS3/4A serine protease. Telaprevir is only indicated for use against hepatitis C genotype 1 viral infections and has not been proven to have an effect on or being safe when used for other genotypes of the virus. The standard therapy of pegylated interferon and ribavirin is less effective on genotype 1.

TRAMADOL

Variant found:

- Gene: ABCB1
- Marker: rs1045642
- Position: chr7:87138645
- Genotype: GG

Tramadol, sold under the brand name Ultram among others, is an opioid pain medication used to treat moderate to moderately severe pain. When taken by mouth in an immediate-release formulation, the onset of pain relief usually begins within an hour. It is also available by injection. It may be sold in

combination with paracetamol (acetaminophen) or as longer-acting formulations. Common side effects include constipation, itchiness, and nausea. Serious side effects may include seizures, increased risk of serotonin syndrome, decreased alertness, and drug addiction. A change in dosage may be recommended in those with kidney or liver problems. It is not recommended in those who are at risk of suicide or in those who are pregnant. While not recommended in women who are breastfeeding, those who take a single dose should not generally stop breastfeeding. Tramadol acts by binding to μ -opioid receptors on neurons. It is also a serotonin-norepinephrine reuptake inhibitor (SNRI). It is converted in the liver to O-desmethyltramadol, an opioid with stronger binding to the μ -opioid receptor.

TRASTUZUMAB

Variant found:

- Gene: FCGR2A
- Marker: rs1801274
- Position: chr1:161479745
- Genotype: GG

Variant found:

- Gene: FCGR3A
- Marker: rs396991
- Position: chr1:161514542
- Genotype: AC

Trastuzumab, sold under the brand name Herceptin among others, is a monoclonal antibody used to treat breast cancer. Specifically it is used for breast cancer that is HER2 receptor positive. It may be used by itself or together with other chemotherapy medication. Trastuzumab is given by slow injection into a vein and injection just under the skin. Common side effects include fever, infection, cough, headache, trouble sleeping, and rash. Other severe side effects include heart failure, allergic reactions, and lung disease. Use during pregnancy may harm the baby. Trastuzumab works by binding to the HER2 receptor and slowing down cell duplication.

VENLAFAXINE

Variant found:

- Gene: FKBP5
- Marker: rs4713916
- Position: chr6:35669983
- Genotype: AG

Venlafaxine, sold under the brand name Effexor among others, is an antidepressant medication of the serotonin-norepinephrine reuptake inhibitor (SNRI) class. It is used to treat major depressive disorder (MDD), generalized anxiety disorder (GAD), panic disorder, and social phobia. It is taken by mouth. Common side effects include loss of appetite, constipation, dry mouth, dizziness, sweating, and sexual problems. Severe side effects include an increased risk of suicide, mania, and serotonin syndrome. Antidepressant withdrawal syndrome may occur if stopped. There are concerns that use during the later part of pregnancy can harm the baby. How it works is not entirely clear but it is believed to involve alterations in neurotransmitters in the brain.

WARFARIN

Variant found:

- Gene: VKORC1
- Marker: rs7294
- Position: chr16:31102321
- Genotype: CT

Variant found:

- Gene: VKORC1
- Marker: rs2359612
- Position: chr16:31103796
- Genotype: AG

Warfarin, sold under the brand name Coumadin among others, is a medication that is used as an anticoagulant (blood thinner). It is commonly used to treat blood clots such as deep vein thrombosis and pulmonary embolism and to prevent stroke in people who have atrial fibrillation, valvular heart disease or artificial heart valves. Less commonly it is used following ST-segment elevation myocardial infarction (STEMI) and orthopedic surgery. It is generally taken by mouth but may also be used by injection into a vein. The common side effect is bleeding. Less common side effects may include areas of tissue damage and purple toes syndrome. Use is not recommended during pregnancy. It is recommended that the effects of warfarin typically be monitored by checking prothrombin time (INR) every one to four weeks. Many other medications and dietary factors can interact with warfarin, either increasing or decreasing its effectiveness. The effects of warfarin may be reversed with phytonadione (vitamin K1), fresh frozen plasma, or prothrombin complex concentrate. Warfarin decreases blood clotting by blocking an enzyme called vitamin K epoxide reductase that reactivates vitamin K1. Without sufficient active vitamin K1, clotting factors II, VII, IX, and X have decreased clotting ability. The anticlotting protein C and protein S are also inhibited but to a lesser degree. A few days are required for full effect to occur and these effects can last for up to five days.

ZIPRASIDONE

Variant found:

- Gene: MC4R
- Marker: rs489693
- Position: chr18:57882787
- Genotype: AA

Ziprasidone, sold under the brand name Geodon among others, is an atypical antipsychotic that is used for the treatment of schizophrenia as well as acute mania and mixed states associated with bipolar disorder. Its immediate release intramuscular injection form is approved for acute agitation in people with schizophrenia. Ziprasidone is also used off-label for depression, bipolar maintenance, and post-traumatic stress disorder (PTSD). Common side effects include dizziness, drowsiness, dry mouth, and twitches. Although it can also cause weight gain, the risk is much lower than for other antipsychotics.

ENALAPRIL

Variant found:

- Gene:NR3C2
- Marker:rs5522
- Position:4:149357475
- Genotype: TT

Genotype TT is associated with increased response to enalapril in people with hypertension as compared to genotypes CC + CT.

Enalapril is a prodrug belonging to the angiotensin-converting enzyme (ACE) inhibitor drug class that works on the renin-angiotensin-aldosterone system, which is responsible for the regulation of blood pressure and fluid and electrolyte homeostasis. Enalapril was first approved by the FDA for the management of hypertension, heart failure, and asymptomatic left ventricular dysfunction.

OMEPRAZOLE

Variant found:

- Gene:ABCB1
- Marker:rs1045642
- Position: chr7:87138645
- Genotype: GG

Genotype GG is associated with decreased concentrations of Omeprazole in infants with Gastroesophageal Reflux as compared to genotype AG + AA.

Omeprazole belongs to the group of proton pump inhibitors (or PPIs). It is defined as a gastroprotective and anti-ulcer drug as it works by reducing the production of stomach acid. Omeprazole comes in the form of hard gastro-resistant capsules and the route of administration is oral. The use is indicated in the treatment of various ailments and diseases, both in adult patients and in children, such as: gastroesophageal reflux disease (GERD), gastric and duodenal ulcers, ulcers caused by *Helicobacter pylori* in combination with other drugs, treatment and prevention ulcers caused by FANS (non-steroidal anti-inflammatory drugs) and treatment of Zollinger-Ellison syndrome.

GLOSSARY

ALLELE

An allele is a variant form of a gene that is located at a specific position, or genetic locus, on a specific chromosome. Humans have two alleles at each genetic locus, with one allele inherited from each parent.

CHROMOSOME

A chromosome is a condensed thread-like structure of DNA that carries hereditary information, or genes. Human cells have 22 chromosome pairs plus two sex chromosomes, giving a total of 46 per cell.

GENOME

A genome is an organism's complete set of DNA, including all of its genes. Each genome contains all of the information needed to build and maintain that organism. In humans, a copy of the entire genome—more than 3 billion DNA base pairs—is contained in all cells that have a nucleus.

GENOTYPE	The genetic makeup of an individual organism. It may also refer to just a particular gene or set of genes carried by an individual. The genotype determines the phenotype, or observable traits of the organism.
ODDS RATIO	The odds ratio is a way of comparing whether the odds of a certain outcome is the same for two different groups. In this report, the odds ratio estimates the probability of a condition occurring in a group of people with a certain genetic variant compared to a group of people without that variant. An odds ratio of 1 means that the two groups are equally likely to develop the condition. An odds ratio higher than 1 means that the people with the genetic variant are more likely to develop the condition, while an odds ratio of less than 1 means that the people with the variant are less likely to develop the condition.
PHENOTYPE	A description of an individual's physical characteristics, including appearance, development and behaviour. The phenotype is determined by the individual's genotype as well as environmental factors.
POPULATION ALLELE FREQUENCY	The allele frequency represents the incidence of a variant in a population. Alleles are variant forms of a gene that are located at the same position, or genetic locus, on a chromosome.
SNP	Single nucleotide polymorphisms, frequently called SNPs, are the most common type of genetic variation among people. A SNP is a variation in a single nucleotide that occurs at a specific position in the genome.