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Pharmacogenomics: Foundation, Clinical Applications and Emerging Role of Artificial Intelligence in Personalized Medicine.

Author(s): [1]Kashish Uppal, [2]Sheikh Irshad Ul Haq,[2]Shafkat Hussain Malik, [3]Ruhit Ashraf*

[1]B. Pharmacy (Student), School of Pharmacy, Desh Bhagat University, Mandi Gobindgarh, Punjab, India

[2]Assistant Professor, School of Pharmacy, Desh Bhagat University, Mandi Gobindgarh, Punjab, India.

[3]Associate Professor, S. Lal Singh Memorial College of Pharmacy, Desh Bhagat University, Mandi Gobindgarh, Punjab, India.

Corresponding Author:

Ruhit Ashraf

Associate Professor

Email: ruwaidashraf1819@gmail.com

Abstract

Pharmacogenomics ties together pharmacology and genomics in order to know why individuals respond differently to the same drugs. It can be used to alter treatment to focus on truly personalized treatment rather than the standard treatment which has always been the one-size-fits-all method by examining how genetic variations influence the absorption of drugs, their metabolism, and their toxicity. This review will follow the development of pharmacogenomics, starting with the first experiments to examine the effects of single genes up to the more recent genome-wide methods, including the example of CYP2D6 and CYP2C19 polymorphisms, which contribute to the responses to metoprolol, clopidogrel, and warfarin. It further describes the molecular basis of pharmacogenomics such as SNP roles, CNVs and indels and discusses Pharmacoepigenomics and non-coding RNAs, which further dictate drug responses. This paper describes the differences in the population of major alleles and their clinical effects in major medicine fields like cardiology, oncology, psychiatry, neurology, pediatrics, and rare diseases. The examples of real-life applications of genetic testing, such as monogenic diabetes (MODY) and the treatment of hepatitis C, are instances of how genetic testing can enhance patient outcomes.

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The accuracy and speed of pharmacogenomic testing are being increased by new technologies like next-generation sequencing (NGS), CRISPR-based gene editing, and multi-omics. Taken together, all these developments are taking healthcare to a more predictive, preventive, and tailor-made future.

Keywords: Pharmacogenomics, CYP450; Pharmacoepigenomics, Next-Generation Sequencing (NGS), Artificial Intelligence,

1. Introduction

1.1 From genes to genomes: The Evolution of pharmacogenomics.

Pharmacogenetics refers to the impact of single gene on a person's response to drugs, for example- one patient gets toxicity from a standard dose while another patient does not. It focuses on the single genetic variations[1]. This field is originated in 1950s. The term pharmacogenetics was given by Friedrich Vogel of Heidelberg in Germany in 1959[2]. Certain people have single gene variations that affect how their body make certain enzymes. These enzymes variations are called polymorphism in which body don't handle medicines in usual ways[3]. This can cause unexpected side effects such as people who have n-acetyltransferase enzyme deficiency can get peripheral neuropathy or liver toxicity[4]. The discovery of CYP2D6 polymorphism and its effect on drug toxicity and other discoveries led to term Pharmacogenomics[5]. The cytochrome P450(CYP)2D6 enzyme is responsible for metabolism of up to 25% of all registered drugs[6]. The Metoprolol is most common drug used in cardiovascular diseases and 80% of this drug is metabolized by CYP2D6 enzyme and has significant effect of CYP2D6 genotype on the Pharmacokinetics of Metoprolol[7]. The CYP2D6 polymorphism led to different metabolizer phenotypes such as, poor metabolizers (PM) which effect the metoprolol and increase the risk of adverse effects such as bradycardia, hypotension so the patient requires dose reduction, Intermediate metabolizers (IM) which effects the metoprolol which reduces the enzyme activity and person may still experience some ADRs at normal dose, extensive metabolizers (EM) which has normal CYP2D6 activity and standard dosing in this case works well, Ultra Rapid metabolizers (UM) in which multiple copy of CYP2D6 or other gene occur, which give higher enzyme activity and reduce the therapeutic effect of Metoprolol and may require higher doses or alternative drug[8]. While Pharmacogenetics made groundbreaking contributions, it had limitations such as failed to explain about complex traits control by multiple genes, neglect the role of epigenetics and gene-gene/ environmental interactions etc.[9]. The term Pharmacogenomics was introduced in 1990s with rise of Human genome project[10]. Pharmacogenomics refers to the broader field which consider effect of multiple gene variations and it looks at the whole genome to understand how many genetic factors together shape drug absorption,

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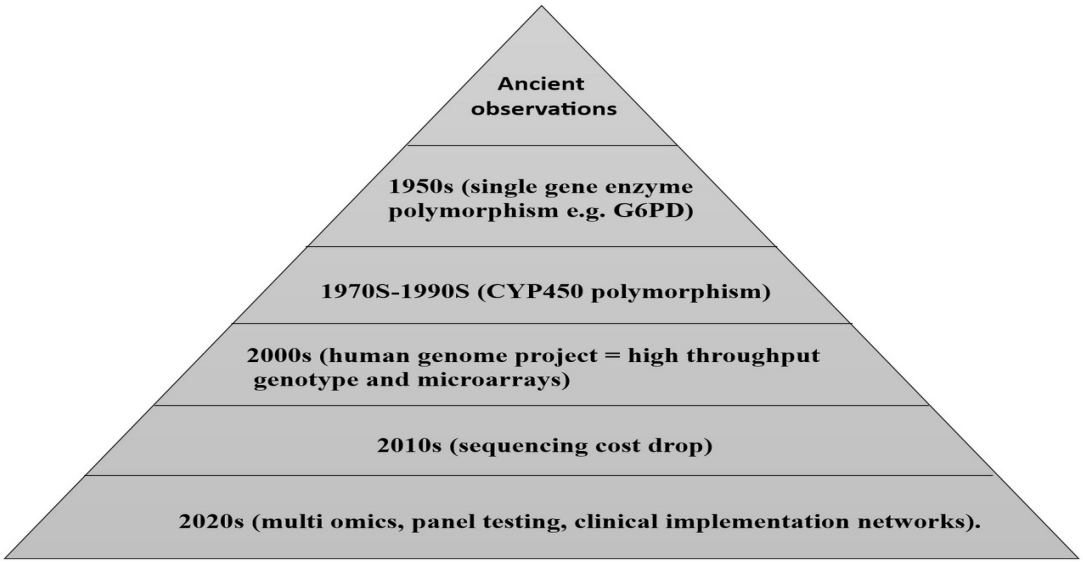
metabolism, targets, and side effects[1]. It combines pharmacology (which is the study of drugs) and genomics (which is the study of genes and their functions) to develop safe, effective, and personalized therapies[11]. Its aims to optimize drug selection and dosing by personalize treatment to the patient's genetic profile, thereby enhancing efficacy and minimizing toxicity[12]. With advancements in genome sequencing, artificial intelligence, and big data, pharmacogenomics is becoming more accessible and applicable[13].

Figure 1: evolution of pharmacogenomics from 19s to 20th era.



1.2. Why pharmacogenomics matters: safety, efficacy, and cost.

Pharmacogenomics shows how genetic variations influence drug metabolism, efficacy, toxicity, ultimately guide clinicians to select the right drug with right dose to the right patient[14]. Personalized medicine represents the paradigm shifts from “one size fits all” approach towards customize drug plans for a person or a group based on their variability in genes, environment, and lifestyle[15]. The key contributions of pharmacogenomics in personalize medicine could be to maintain the safety, stability of the drug. it helps in drug development, genomics help identifying the large group of people belongs to small population that get benefited from a therapy, enabling target trials and faster regulatory approvals, it is Cost effective, avoid ineffective therapies and hospitalization from adverse reaction saves money[16]. It is used for

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Safer prescribing; genetic markers identify people at higher risk for severe adverse drug reactions[17]. Changing dose or choosing an alternative drug/ option can be safer and more effective. And also, can be used for better efficacy, some drugs only work in patients with particular genetic background. Testing can match right drug to the right patient[18].

2. Population variability in pharmacogenomic allele:

The pharmacogenomic study can be done individually as well as in population of different areas as well

Table 1: Given below is the global data collected which shows gene allele variant and its effect on drug in different populations.

Gene/marker	Key variant.	Clinical problems	Variant carriers.	References
CYP2C19	*2, *3(loss of function), *17(increased).	Affect Clopidogrel, PIPS, some antidepressants.	East Asians: PMs (10-15%) Europeans: PMs (2-5%) Africans: variable (10-20%).	[19]
CYP2D6	Multiple no function, decreased and gene dup alleles (PM/UM).	Affect many depressants, opioids.	Europeans: PMs (5-10%) Africans/ middle east: high diversity UMs (gene duplication)	[20]
CYP2C9/ VKORC1	CYP2C9*2, VKORC1-1639G>A.	Affect Warfarin dosing (risk bleeding)	European: CYP2C9*2, *3 more common East Asians: VKORC1A allele (high warfarin sensitivity). Africans: lower *2, *3 but another variant exist.	[21]
TPMT	TPMT*2, *3A, *3C.	Thiopurine toxicity risk	Europeans: (0-3%) Africans and some Asians:	[22]

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			variable.	
NUDT15	Lost variant (east Asians).	Thiopurine toxicity).	East Asians: actionable variant more common. Europeans/ Africans: rare.	[23]
DPYD	*2A, other LOF.	Fluoropyrimidine (severe toxicity).	European: low frequency actionable variant Africans/ Asians: variant spectrum differs.	[24]
HLA-B*57:01	HLA-B*57:01	Abacavir (hypersensitivity).	Europeans: carries 4-8% East Asians: low, Africans: variable.	[25]
HLA-B*15:02	HLA-B*15:02	Carbamazepine severe cutaneous reactions.	High in East Asia and some South Asians Europeans/ Africans: rare.	[26]
HLA-B*58:01	HLA-B*58:01	Allopurinol severe cutaneous reactions.	East Asians and some Asians: (higher %) Europeans/ Africans: lower.	[27]
SLCO1B1/ rs4149056	Decreased function allele.	Statin (Simvastatin) risks.	European: allele (10-15%). Asians/ Africans: frequency varies but variant is present.	[28]

3} Molecular basis of pharmacogenomics

The molecular basis of pharmacogenomics studies shows how variations in DNA affect the way drugs are processed and how they act in the body, variations such as single nucleotide polymorphisms (SNPs), insertions or deletion (indels), copy number variations (CNVs) can alter the function and structure of enzymes, targets and drug transporters[29]. These variations can

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affect pharmacodynamics and pharmacokinetics of drug. By linking gene variation to drug response, pharmacogenomics provides the scientific foundation for personalized medicines[30].

3.1. Genetic variations: Changes in the genes, protein structure, abundance or regulation all together affect how person responds to a medication[31]. Pharmacogenomics helps to predict the effective therapy in an individual, who will need dose changes and who is at high risk of adverse effects[16]. The most common type of genetic variation relevant to pharmacogenomics are single nucleotide polymorphisms in this, In certain population, a single base change at specific position in genome occur, another one is Insertion/ Deletion (indels): In this small insertion or deletion of base occur, within coding region-frameshift indels often produce nonfunctional protein, while within regulatory regions can change gene expression for example insertion disrupts a reading frame in drug metabolizing enzyme can abolish activity and produce poor metabolizers phenotypes, CNVs: Copy number variants: In this large, scale changes occur where DNA stretches are deleted or duplicated and change number of gene copies[32]. Classic example: CYP2D6 gene duplication can create ultra rapid metabolizers phenotype causing faster breakdown of drugs like codeine to morphine which will increase effect or toxicity and gene deletion of CYP2D6*5 which make poor metabolizers[33].

3.2. Pharmacokinetic genes: ADME (CYP450, UDP, SLCO1B1).

Variation in ADME gene strongly affects drug levels and exposures[34]. Cytochrome P450 (CYPS): In humans the major CYPs that metabolize the drugs are CYP1A2, CYP2B6, CYP2C9, CYP2C19, CYP2D6, CYP3A4/5[35]. Important clinical example of this is CYP2C19 variant. As it mentioned earlier Clopidogrel activation depends upon functional CYP2C19. LOF allele (2,3) reduces effectiveness and increase the risk of stent thrombosis and GOF allele (17) increases activity[36]. A person's gene type can help the doctors to decide if dose needs to be changed or if an alternative drug would work better, UDP-Glucuronosyltransferases (UGTs): UGTs conjugate or metabolites the drug with glucuronic acid, increase water solubility and promote excretion. Clinical examples include UGT1A1 28 allele lowers expression and reduces glucuronidation capacity. This affects the drug like Irinotecan. Testing for UGT1A1 variants can guide dose selection for Irinotecan[37], Drug transporters- SLCO1B1 and others: Transporters on the cell membranes help drug efflux and uptake. SCLO1B1 is a hepatic transporter that effect Statin clearance[38].

3.3. Pharmacodynamic genes: Receptors, targets (VKORC1, HER2, HLA-B*57:01): VKORC1 (vitamin k epoxide reductase complex 1) recycles vitamin K, which is necessary for gamma carboxylation of clotting factors. The Warfarin inhibits VKORC, common

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VKORC1 variant (1639G>A) reduces VKORC1 expression, increases the sensitivity to Warfarin. So, the individual having VKORC1 variant are sensitive to Warfarin and need lower dose[39], **HER2** is a protein that help the cells to grow and overexpression or amplification of HER2 drives growth in a subset of breast cancer. HER2 amplification predicts response to HER2 targeted therapies such as Trastuzumab and Pertuzumab[40], **HLA alleles:** HLA molecules present peptides to T-cells, certain HLA alleles present drug-protein adducts in ways that trigger immune responses. The important example include: HLA-B*57:01: strongly associated with abacavir hypersensitivity. Patient with positive for HLA-B*57:01 have high risk and should not receive abacavir, HLA-B*15:02-carbamazepine- associated Stevens Johnson Syndrome in Asian population etc. HLA variant does not alter the drug's target, instead determine immune surveillance and make them unique PD-type pharmacogenomic markers for hypersensitivity[41].

Variant type	Example of molecular change	Common functional outcomes	Clinical implications	References
HLA allele	HLA-B*57:01	Immune presentation change.	Abacavir hypersensitivity risks.	[25]
Promoter allele	UGTA1*28 (TA repeat)	Lower expression.	Irinotecan toxicity.	[42]
Gene amplification	HER2 amplification	Overexpression.	Candidate for HER2 targeted therapy.	[43]
SNP (splice)	CYP2C19*2	LOF	Reduce clopidogrel activation.	[44]
CNV duplication	CYP2D6 duplication	Increase enzyme activity.	Rapid metabolism of codeine- risks of toxicity.	[33]
Indel (frameshift)	Increase metabolic enzyme	LOF	PMs.	[45]
SNPs	CYP2C19*2	Reduce enzyme activity.	May require	[46]

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			reductio of dose of warfarin.	

Table 2: this table shows the different variant types with its example their functional outcomes and their clinical implications.

4. **Pharmacogenomics databases and resources:** PharmGKB is an open-source, web-based initiative to assemble information on the influence of genetic variations on drug responses as one of the primary resources that can be used by researchers and clinicians to practice personalized medicine. It can assist in determining the genetic markers of response to widely used drugs in cancer, cardiovascular and psychiatric diseases[47]. PharmVar brings a universal nomenclature to the major drug-metabolizing genes (especially the cytochrome P450 family) so that the genetic test results can be interpreted as precisely and reliably as possible[48]. Clinical Pharmacogenetics Implementation Consortium (CPIC) and the Dutch Pharmacogenetics Working Group (DPWG) are centers that apply translations of genetic findings into clinical practice, e.g. dose adjustment or alternative therapy[49]. Also, pharmacogenomic data is on the majority of approved drug labeling by the U.S. FDA, signifying that genetic testing is essential to prevent adverse effects or enhance the effectiveness of treatment[50]. Combined, these efforts improve the application of pharmacogenomics to clinical practice, improving patient care, which is safer, more effective, and even personalized[51].

5. **Pharmacogenomics in the Clinic: From Genes to healing:**

5.1. Cardiology: use of pharmacogenomics is very potential to enhance patient care by reducing adverse effects, healthcare related costs, etc. and also improves safety and efficacy of drug[16]. in this we will discuss the basic use of pharmacogenomic data of drug like Clopidogrel and Warfarin which are the most common drugs used in the prevention of cardiovascular disorders[52]. Antiplatelet therapy is use in the prevention of wide range of cardiovascular disorders but despite its effectiveness recurrent thrombotic events and adverse effects like bleeding remains an important clinical problem in cardiovascular medicines and there are large study evidences which suggest that the genetic variations effect the drug's pharmacokinetics and pharmacodynamics[53]. Clopidogrel (antiplatelet drug) is most prescribed to prevent ischemic events in patients of acute coronary syndrome (ACS) undergoing percutaneous coronary interventions (PCI)[54]. Despite its wide use 4% to 30% of patients do not respond to this drug in usual ways, resulting in high on treatment platelet

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reactivity (HTPR) and increase the rates of cardiovascular events. The PCI patients on Clopidogrel who exhibit HTPR are more likely to experience recurrent adverse clinical effects[55].

CYP2C19 is a major contributor in metabolism of Clopidogrel. It converts 2 oxo-Clopidogrel which is an intermediate metabolite into active metabolite which will bind to P2RY12 receptors and inhibit the platelet aggregation irreversibly[36]. Now the consistent studies have shown both loss of function (LOF) (CYP2C19*2, CYP2C19*3) and gain of function (GOF) (CYP2C19*17) have been associated with Clopidogrel efficacy. The CYP2C19 variant: individual who has CYP2C19*2 variant have poorer Clopidogrel response. This CYP2C19*2 specific higher platelet aggregation post Clopidogrel exposure was due to altered Clopidogrel metabolism resulting in reduction of Clopidogrel active metabolite[56]. Clopidogrel treated patient who carry at least one copy of CYP2C19*2 variant were more likely to experience major adverse cardiovascular events compared to individual who were homozygous for CYP2C19*1 allele. While CYP2C19*2 variant has been most extensively studied polymorphism with regards to Clopidogrel response variability, effort has been made to understand the impact of relatively common CYP2C19*17 allele. Studies suggest increase incidence of stent thrombosis in CYP2C19*2 allele carriers compare to noncarriers and indicate a strong association between CYP2C19*2 genotype and risk of stent thrombosis[57].

CYP2C19*17 variant: studies shows that individual carries CYP2C19*17 variant have better Clopidogrel response[58]. So instead of Clopidogrel new generation drugs like Prasugrel and Ticagrelor are used. Prasugrel is also a prodrug but unlike Clopidogrel, due to its chemical structure vast majority of parent compound is metabolized into active metabolite and has greater platelet P2Y12 inhibition[59]. Ticagrelor is a newer type of drug that is administered orally, which blocks P2Y12 receptor and get activated in body itself so it doesn’t need to go through bioactivation process[60]. The most common anticoagulant drug used is Warfarin, which acts by inhibiting an enzyme known as VKORC1, which triggers the clotting factors in blood[61]. Individuals do not react to Warfarin in a uniform manner due to the differences in genetics primarily I two genes namely, CYP2C9 and VKORC1. CYP2C9 assists in hydrolyzing Warfarin in the body and therefore individuals with some forms of this gene metabolize Warfarin at reduced rate, and require a reduced dose. VKORC1 influences the level of target enzyme produced, the variants in this case also have potential to alter the quantity of Warfarin required. Knowledge of such genetic variations may assist physicians in forecasting optimal dose to minimize risks of bleeding or clotting[62].

Table 3: This table shows the application of pharmacogenomics with gene variants leading to clinical problems in drugs used in cardiology.

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Drug/class	Gene variant	Clinical problem	Clinical applications	References
Clopidogrel	CYP2C19*2	LOF→ poor platelet inhibition	Switch to Prasugrel/ Ticagrelor.	[63]
	CYP2C19*3	No active metabolite→ stent Thrombosis risk.	Avoid Clopidogrel.	[64]
	CYP2C19*17	GOF→ excessive bleeding.	Use a lower dose or Consider alternative.	[65]
Warfarin	VKORC1-1639G>A	Increased sensitivity→ Bleeding risk.	Start with lower dose.	[66]
	CYP2C9*2	Slow metabolism→ Bleeding.	Reduce dose.	[39]
	CYP2C9*3	Very slow metabolism → toxicity.	Very slow starting dose needed.	[67]
Statin (Simvastatin)	SCLOB1*5	Myopathy rhabdomyolysis risk.	Use Pravastatin/ Rosuvastatin .	[68]
	SCLOB1*15	Transport defect→ Muscle toxicity.	Avoid Simvastatin, dose adjustments.	[38]

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5.2. Oncology: In the cancer treatment, pharmacogenomics is becoming really important because chemotherapy drugs are very powerful and risky. A well- known case is 5-flourouracil (5-FU), used in breast and colorectal cancers[69]. Normally, the DPYD gene makes an enzyme (DPD) that breaks this drug down. But some people have DPYD changes, like the *2A type, that reduce enzyme activity. For them, a normal dose can become dangerous, causing problems such as diarrhea, bone marrow damage, or even neurological issues. In such patients, doctors have lower dose or avoid the drug. genes like TYMS and MTHFR can also influence how safe and effective 5-FU is[70]. Other examples include breast cancer; Tamoxifen is affected by genetics. It needs to be activated by the enzyme CYP2D6. People with poor CYP2D6 function don't make enough of the active drug, so treatment may fail. In such cases, alternatives like Aromatase inhibitors are considered[71]. Overall, pharmacogenomics in oncology helps move away from 'one treatment for all' to more individualized care, reducing toxicity and improving outcomes[72].

Drug/ Class	Gene variant	Clinical Problems	Clinical Applications.	References
Fluoropyrimidines (5-FU, Capecitabine, Tegafur) Methotrexate	DPYD*2A	severe myelosuppression, diarrhea.	Avoid or reduced dose of 5-FU/ Capecitabine.	[73]
	**DPYD13, 9B, HapB3	Toxicity.	Choose non-fluoropyrimidine regimen if high risk.	[74]
	MTHFR 677>T	Higher toxicity, variable survival.	adjust folate related therapy.	[75]
	MTHFR1298>C	altered MTX metabolism.	Monitor toxicity, supportive care.	[75]
Thiopurines (6-MP, Azathioprine, Thioguanine)	TPMT*2	Risk of myelosuppression.	Dose reduction.	[76]
	**TPMT3A, 3C	High Thioguanine nucleotide buildup→ bone marrow	Genotype based dose adjustment.	[77]

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		suppression.		
	NUDT15*2	Severe cytopenia.	Reduce dose or avoid thiopurines.	[78]
Irinotecan	UGT1A1*28	Severe neutropenia.	Lower dose or close monitoring.	[42]
	UGT1A1*6	Diarrhea, neutropenia.	Reduce dose or use alternatives.	[79]

Table 4: this table shows the clinical application of pharmacogenomics in different gene variant leading to clinical problem for drugs used in oncology.

Psychiatry: Many psychiatric medicines are broken down in the liver by enzymes, mainly the cytochrome P450 family. Changes in these genes affect how quickly or slowly the drug is removed from the body[80]. CYP2C19 is one of the key gene. It affects medicines like Citalopram, Escitalopram, and Sertraline. Poor metabolizers may get more side effects, while Ultra-rapid ones may see no improvement at all[81]. In these cases, doctors switch to safer alternatives such as Fluoxetine. Altogether, pharmacogenomics makes psychiatric treatment less of a trial-and-error process and helps patients get the right medicine faster[82].

Drug/ class	Gene/ variant	Clinical problem	Clinical application	References.
SSRIs (Paroxetine, Fluoxetine, Escitalopram)	CYP2D6 (PM)	High level of drug→ side effects.	Lower dose or switch drug.	[83]
	CYP2D6 Ultrarapid metabolizers (UM)	Subtherapeutic effect.	Alternative antidepressant.	[84]
	CYP2C19 (PM)	High plasma concentration of Escitalopram/ Citalopram.	Reduce dose.	[85]
	CYP2C19 (UM)	Poor response, treatment failure.	Use non- CYP2C19 metabolized SSRI.	[86]

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TCA (Amitriptyline, Nortriptyline)	CYP2D6 (PM)	Toxic plasma levels.	Reduce dose.	[87]
	CYP2D6 (UM)	Lack of efficacy.	Switch antidepressants.	[84]
	CYP2C19 (PM)	Slow metabolism, more side effects.	Reduce dose.	[88]
Antipsychotic (Risperidone, Haloperidol)	CYP2D6 PM	High levels→ EPS, Sedation.	Lower dose or change agent.	[89]
	CYP2D6 (UM)	Ineffective due to rapid clearance.	Alternative drug.	[90]
Mood stabilizer (Valproate)	POLO variants	Risk of liver failure in children.	Avoid Valproate.	[91]

Table 5: this table show the application of pharmacogenomics in different gene variant leading to clinical problems for drugs used in psychiatry.

Neurology: In neurology, pharmacogenomics mainly helps to avoid severe side effects of anti-epileptic drugs[92]. for example, patients with the HLA-B*15:02 gene are at very high risk of developing Steven-Johnson syndrome when given Carbamazepine, especially in Asian populations[93]. By testing for this gene before treatment, doctors can choose safer alternatives like Levetiracetam or Valproate[93]. Even in movement disorders, the COMT Val158Met variant can influence how patients respond to Levodopa, which is important for Parkinsons disease management. Overall, neurology uses pharmacogenomics to guide both safety and effectiveness of therapy[94].

Drug/ class	Gene variant	Clinical problem	Clinical application	References.
Carbamazepine, Oxcarbazepine	HLA-B*15:02	SJS/TEN.	Avoid instead use Levetiracetam/ Valproate.	[95]

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	HLA-A*31:01	Hypersensitivity syndrome.	Avoid in carriers.	[25]
Phenytoin	CYP2C9*2	High Phenytoin levels→ toxicity.	Dose reduction.	[96]
	CYP2C9*3	Very slow clearance→ toxicity.	Lower dose or alternative AED.	[97]
	HLA-B*15:02	SJS/TEN.	Avoid in carriers.	[98]
Levodopa	COMT VAL158Met	Variability I dopamine metabolism.	Adjust dose or add COMT inhibitor.	[94]
Valproate	POLO mutations	Fatal hepatotoxicity.	Contraindicated.	[99]

Table 6: this table shows the application of pharmacogenomics in different gene variant leading to clinical problems for drugs used in neurology.

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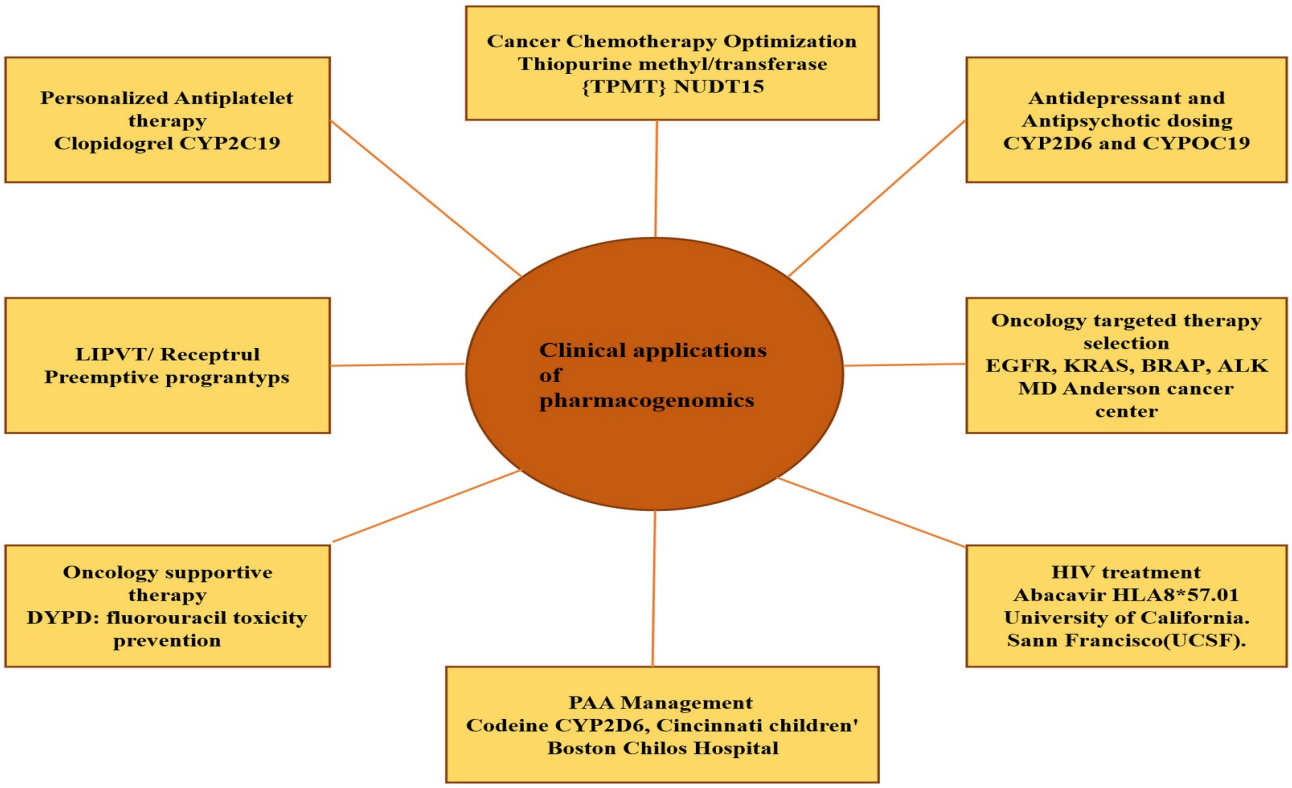

Figure 2

Rare disease treated by enabling orphan drug development: Pharmacogenomics has transformed the treating of rare diseases by enabling the development and precise use of orphan drug tailored to individual genetic variants, for instance, cystic fibrosis is caused by mutations in the CFTR gene, and the drugs like Ivacaftor specifically target the GD551D CFTR confirms this mutation show dramatic improvements in lung function and quality of life, while those without mutation do not benefit. Another key example is Spinal Muscular Atrophy (SMA), where gene therapies like Zolgensma (onasemnogene abeparvovec) and precision RNA modulators such as Nusinersen (Spinraza) are only given after confirming SMN1 gene deletions or mutations by genomic analysis, offering many children a chance to thrive who previously face poor prognosis[100].

5.3. Real life cases:

Case 1: Type 2 Diabetes Mellitus- Sulfonyl urea in monogenetic diabetes (MODY)

Example of a real-life case of a teenage girl from UK, named Sophie began experiencing symptoms of high blood sugar at the age of 14. Her father and grandmother were also diagnosed

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with diabetes when they were young and they were treated with insulin. Sophie was also placed initially on insulin therapy for presumed type 1 diabetes, but her blood sugar level was erratic and she hate daily injections. Now because of her young age and strong family history, her doctor suspected a rare genetic form of diabetes[101]. Sophie had a genetic test called targeted gene sequencing panels using next-generation sequencing (NGS) and found to have a novel variant in HNF1A gene (this gene controls how the pancreas make insulin but the people who have this gene variant their pancreas still work but do not respond properly)- diagnosing her with MODY (maturity onset Diabetes of young type-3)[102]. Now with diagnosis, her doctor switched Sophie from insulin to a sulfonylurea tablet (Glibenclamide). She responded amazingly-with perfect blood sugar control and no longer needing insulin shots. Sophie's quality of life improved, she was able to manage her diabetes with a daily pill, and her case is used in worldwide literature as a textbook example of PGx based intervention[103].

Case 2: Hepatitis C impact of 1L-10 Genotype

Another example of a real-life case, a boy named Ahmed, a 32 years old man from Tunisia, was living with chronic Hepatitis C infection and concerned about treatment success since his brother has failed interferon therapy. Now the problem is Hepatitis C cures are unreliable; interferon therapy can cause months of side effects for the little benefit. Ahmed's doctor ordered a genetic test which is single nucleotide polymorphism (SNP) genotyping assay to analyze his 1L-10 gene. His genotype showed a low expression of 1L-10 meaning he was genetically mor likely to respond well to interferon therapy. 1L-10 helps regulate the immune system. Some people naturally produce more or less 1L-10, those with lower levels fight hepatitis C more effectively when treated with interferon. Now knowing Ahmed had good response genotype, his doctor recommended the standard interferon and Ribavirin therapy. He completed treatment and cleared the virus after 6 months, unlike his brother who had a poor response genotype. Ahmed avoided unnecessary treatment delays, saved money, and had much higher chance of cue thanks to PGx testing. This approach is increasingly used and documented in clinical studies from North Africa, the Middle East, and Asia[104].

6. **Patient engagement, equity and Challenges in pharmacogenomics (ELSI):** Pharmacogenomics (PGx) is the attempt to make treatment more individualized with the help of the knowledge of gene responses to drugs[105]. Its adoption however is still unsatisfactory particularly in developing nations because of the expensive nature of carrying tests, inadequate infrastructure, support policies and incompetence among health practitioners[106]. There are also significant ethical, legal, and social issues (ELSI), such as the privacy problems, the possibility of genetic information abuse, and the risk of discrimination[107]. Lopsided access may further enhance healthcare disparity where only

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rich patients would be able to afford the costs of PGx tests[108]. It is necessary to establish the public trust, maintain open communication, and implement powerful data protection laws[109]. The national genome programs in countries such as UK and France demonstrate that the government is able to support, fund and educate the program. The developing countries ought to place emphasis on affordability, capacity building, and the implementation of the application of the PGx to the major diseases such as the cancer, HIV, and heart diseases. Caring about the ethics, justice, and access, PGx can make treatment safer, less expensive, and change the global healthcare[110].

7. **Implementation of pharmacogenomics in hospital setting:** Although pharmacogenomics (PGx) is gaining more and more clinical evidence, its application is still underdeveloped in hospitals worldwide, which requires a systematic adoption of the use of this method in hospitals. It starts with assessment of the evidence of PGx, the most relevant drugs and the actionable drug-gene pairs and variant alleles are selected[111]. These selections are guided by data provided by several experts including PharmGKB, the Clinical Pharmacogenetics Implementation Consortium (CPIC), the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA). Following the prioritization, it may be followed by panel testing, and the results may be incorporated into the electronic health records (EHRs) and clinical decision support systems (CDSS)[112]. The implementation will involve action on the part of the major stakeholders: regulatory agencies to accept and regulate the use of the PGx tests, hospital administrations to fund and manage such tests, pharmacy and therapeutics committees to consider priority drugs, laboratories to perform genotyping and cooperate with IT and pharmacy, IT departments to integrate the results, healthcare providers to implement such results in treatment and educate patients, and payers to enable reimbursement[113]. The patient engagement and feedback are needed to optimize the practice. FDA has also facilitated the exposure of PGx with public tables of drug-gene interactions, including those between HLA-B57:01 and Abacavir, dose modifications during DPYD, as well as antiplatelet therapy by CYP2C19 variants. Most recent label changes can be seen through 2024 which show the growing nature of the applications of the PGx in oncology, cardiology, psychiatry and infectious diseases[114]**.**

8. **Emerging technologies in Pharmacogenomics:** The imminent sequencing technologies (NGS), whole genome sequencing, CRISPR, genome editing, pharmacogenomics (PGx), and multi-omics are ushering the medical field into the era of actual personalization[115]. Genetic variations which impact drug response and enable the detection of differences in dose accuracy and side effects which would in fact reduce the drug side effects are now detectable fast and cheaply through NGS and genome sequencing[116]. CRISPR takes this a step further

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since the technology allows precise correction of mutated genes, which promises to bring fresh hope to the treatment of cancer and rare genetic diseases[4]. Multi-omics is a subset of genomics, proteomics, and metabolomics, among others, that can be used to give a holistic perspective of drug processing, and AI can be used to inform treatment decisions by examining large amounts of data. Despite these barriers such as cost, data complexity and ethics, these innovations are defining the future of precision medicine[117].

9. **Artificial intelligence and Machine learning in Pharmacogenomics:** Artificial Intelligence (AI) is defined as computer systems that can handle tasks traditionally handled by human intelligence, and Machine Learning (ML) is a sub-discipline of AI and allows such systems to learn and provide predictions without need of explicit programming[118]. Leveraging AI and ML in pharmacogenomics the study of how genetic variation affects the way different people respond to drugs, AI and ML are changing the field by constructing predictive models based on large molecular, clinical and genetic data sets[119]. Single-gene markers were the markers that traditional pharmacogenomics used to work with, but AI and ML now have access to complex, multidimensional information, including DNA sequences, gene expression, protein interactions, and clinical histories, to make genuinely personalized treatment suggestions[120]. This is a significant change of the old-fashioned one-size-fits-all approach towards precision medicine[121]. In the field of oncology, an AI-based system called PANCDR (Precise Anti-Cancer Drug Response) uses adversarial neural networks to determine how cancer patients will react to the action of chemo drugs[122]. With brain tumor patients tested, and tested validated using the Cancer Genome Atlas (TCGA) database, PANCDR has been found to be much more accurate than conventional methods by bridging the gap between patient outcomes in the laboratory and in reality, on patients[123]. Equally, AI is transforming mental health care by forecasting the reaction to antidepressants and mood stabilizers using genomic data and age of the patient, as well as their symptoms and a medical history[124]. These models help psychiatrists to select the best medications to use in major depressive and bipolar disorders at the first instance, eliminating trial and error treatment[125]. Drug repurposing AI is also being applied to discover novel therapeutic applications of known drugs, by applying more sophisticated network modeling, combining both genetic and spatial genome data, to find novel interactions between drugs and diseases. These systems are able to forecast that a drug that was approved to treat a specific disease will work effectively on a disease with a similar genetic mechanism and hasty drug discovery. These advances are motivated by several machine learning approaches: supervised learning supervised learning approaches, such as random forests and support vector machines (SVMs), rely on known data on drug-response relationships to make predictions in new patients; deep learning deep learning models (such as convolutional neural networks

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(CNNs)) process rich genomic and time-series clinical data; unsupervised learning approaches cluster patients by their genetic similarities to demonstrate new response patterns; and reinforcement learning is underway to find optimal combinations of multiple drugs and treatment regimens[126]. The Next-Generation Sequencing (NGS), Whole Genome Sequencing (WGS), Electronic Health Records (EHRs) and multi-omics, which encompass genomics, proteomics and metabolomics, are some of the various data types included in modern pharmacogenomics. Natural Language Processing (NLP) systems identify information within scientific literature and clinical records and Clinical Decision Support Systems (CDSS) consume this combined information to make prescription recommendations within real time that are informed by genetics[127]. Hospitals are currently integrating AI-based CDSS to recommend the best drugs and dosage depending on the genetic composition of the patient, detect possible adverse drug reactions, and avoid any harmful interactions[128]. These advances notwithstanding, there are a number of challenges. Another significant concern is the diversity and bias of the data as the majority of pharmacogenomic databases are characterized with European genetic data, which is not applicable to other populations[129]. Present attempts are directed at creating more diverse datasets to make it fair and inclusive. There are legal and ethical issues that have to be addressed, too; regulatory bodies are attempting to build the framework of AI-based solution verification, and privacy protection is also paramount[130]. Such approaches as federated learning enable the training of models in different institutions, avoiding the transfer of sensitive patient information to maintain confidentiality[131]. Another challenge is the integration with the healthcare systems; this will be successful only with user-friendly interfaces, clear AI suggestions, and appropriate training of the healthcare providers[132]. On the whole, AI and pharmacogenomics are converging, turning medicine into a resource-driven and predictive and personal field of study, allowing more specific therapies based on the genetic background of a specific patient and marking the beginning of a new era of precision healthcare[133].

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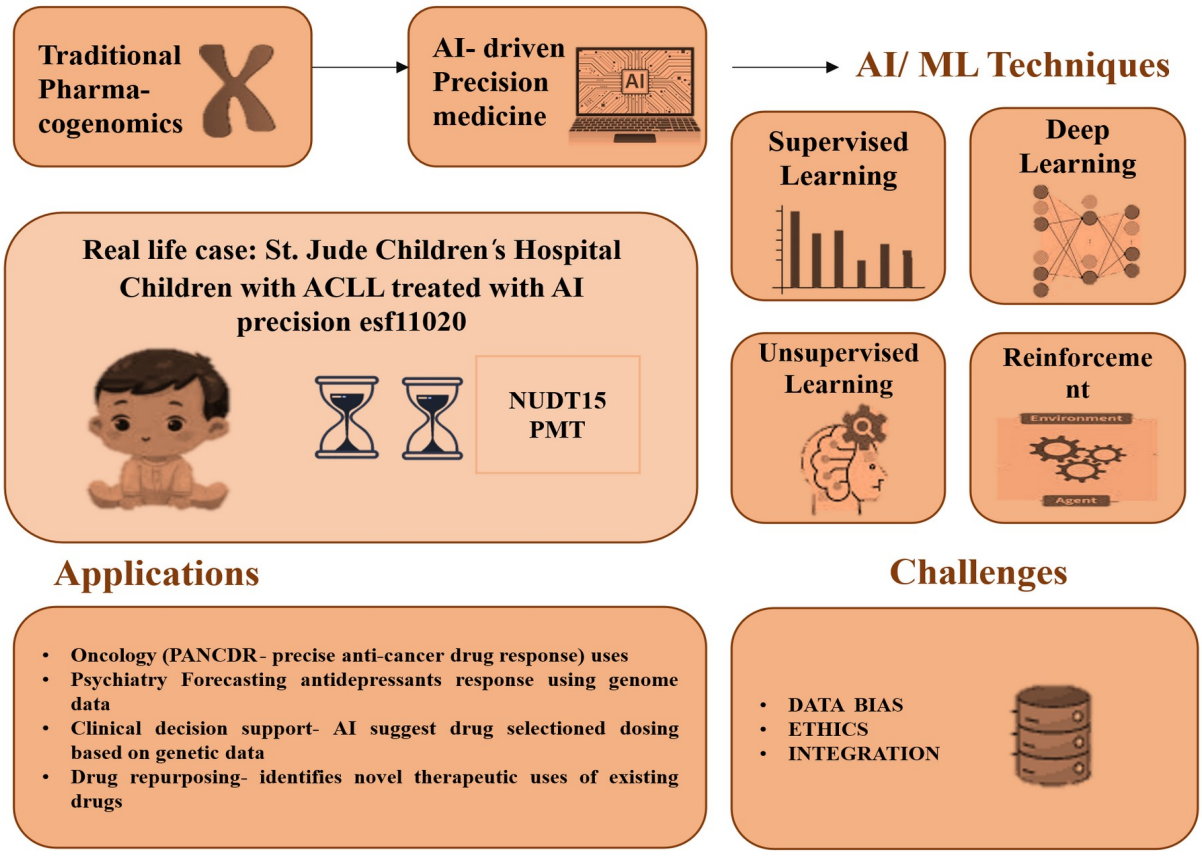

Figure 3: Artificial intelligence in pharmacogenomics.

9.1. Real life cases: now let's take an example of use of AI powered pharmacogenomics for childhood leukemia. Children diagnosed with acute lymphoblastic leukemia (ALL) at St. Jude children's research hospital in the USA. The kids with leukemia get strong chemotherapy. Some have severe side effects, and others don't respond well, often due to hidden genetic differences[134]. Now the hospital uses AI and ML to analyze huge sets of genetic and medical data from these children. The computer programme looks for patterns in the genes such as TPMT and NUDT15, that affect how each child's body handles chemotherapy drugs. then AI predict which children is more likely to get serious side effects or not respond to certain medications[135]. Treatment: now if a child is found to have a genetic variant that makes them sensitive to a particular drug, the dose is lowered or new drug is used which prevents bad reaction and helping cure the cancer. This approach is already being use, helping hundreds of children get better treatment with fewer side effects[134].

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Ongoing research at St. Jude is expanding the AI system for more types of cancer and more drugs, aiming for safer, tailored therapies for young patients everywhere[136].

10. future direction of Pharmacogenomics:

Future pharmacogenomics will involve a high-level technology, integration of large-scale data and a wider clinical application[137]. Genome and exome sequencing will allow more accurate and timely diagnosis of common and rare diseases as they will be cheaper and faster[138]. AI and machine learning will be critical in decoding the genomic data, drug reactions, and the individual-based treatment[139]. The international biobanks will enlarge the variety of the population, enhance the exchange of data, and endorse the pharmacogenomic conventions applicable worldwide[140]. The innovations based on AI will increase drug repurposing, gene therapy, and customized treatments of rare illnesses[141]. Nevertheless, there is still a challenge of providing fair access, patient privacy, and healthcare professional education[142]. New edge AI and adaptive learning systems will make it possible to provide real-time treatment recommendations based on genomics even in resource-limited environments[143]. In the future, AI-enabled pharmacogenomics enables healthcare to become proactive and precision-oriented in 2026 and further, and genomic literacy becomes significant in the future healthcare environment[144].

Conclusion:

Pharmacogenomics has come out of its single-gene-concentrated approach to understanding the impact the entire genome has on pharmacodynamics. It has gained significance now, as genomic testing is essential in various fields of medicine such as cardiology, oncology, psychiatry, neurology, and even pediatrics where safer, more accurate, and more effective therapeutic results become possible. Genetic diversity is a population-based study which also shows that interethnic differences in allele frequencies are significantly high and thus there is a need to have globally inclusive datasets to ensure equitable health care benefits. Recent technologies, including next-generation sequencing (NGS), whole-genome sequencing and the CRISPR-based gene editing, are expanding the clinical field of use at an astronomical pace. Besides this, Pharmacoepigenomics and non-coding RNAs demonstrate that gene expression regulation that is not confined to the primary DNA sequence has a significant impact on the response of patients to treatment. Artificial Intelligence (AI) and Machine Learning (ML) are however facilitating the most significant change in the field. These technologies are used to predict pharmacologic reactions and real-time individualization of treatment using enormous genomic and clinical data. Artificial intelligence-based solutions are already improving patient safety through predicting adverse reactions and improving clinical decision-making. Despite such issues as financial expenses, ethical

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issues, and scarce infrastructure, the trend of precision medicine is undeniable. The combination of AI and pharmacogenomics is an indicator of a significant breakthrough in a healthcare future that is more precise, efficient, fair, safer, and focused on a unique genetic and molecular situation of an individual.

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