



Clinical Implementation of Pharmacogenomics

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Center for Personalized Medicine

Coordinates predictive/diagnostic and therapeutic services along with education & research

Molecular Pathology

- *Tumor sequencing*
- *Hot spot NGS panel*
- *Microarray*

Clinics

- *Medical Genetics*
- *Oncology*
- *Pharmacogenomics*
- *Cardiology*
- *Neurology*
- *Diabetes*

Systemic integration

- *Pharmacogenomics*
- *Genomic results display*

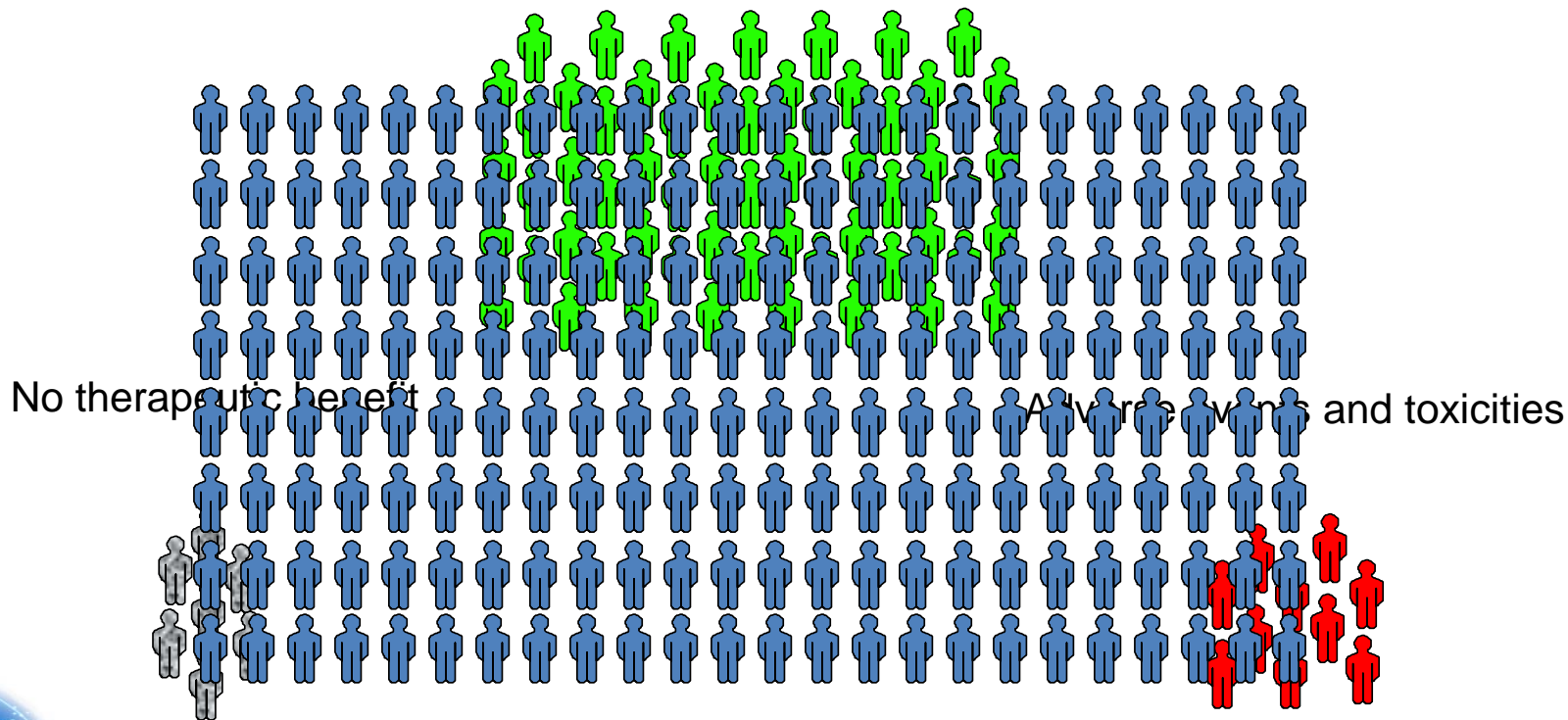
Education

- *Physicians*
- *Nurses*
- *Pharmacists*
- *Employees*
- *Patients*

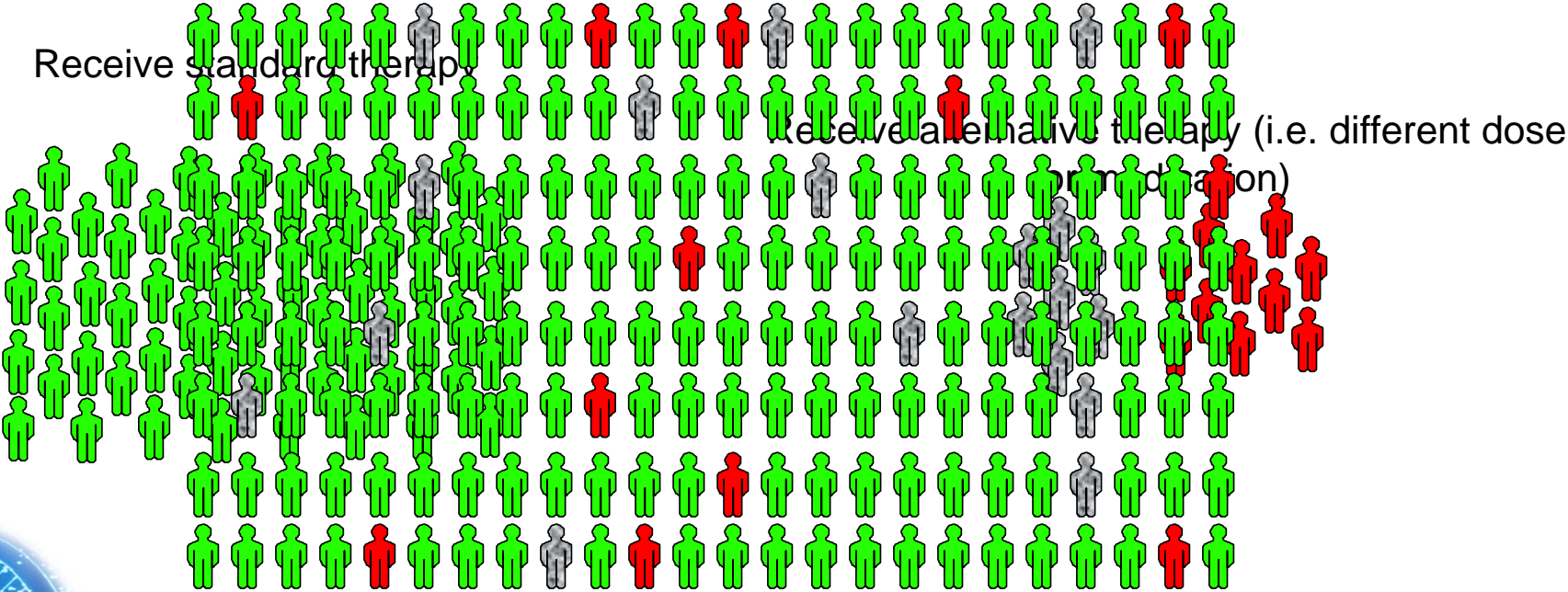
Research

Clinical Biorepository Translational

Benefit with little to no toxicity



Genetic testing can identify these patients before they take the medications



Pharmacogenetics helps clinicians choose between therapeutic equals

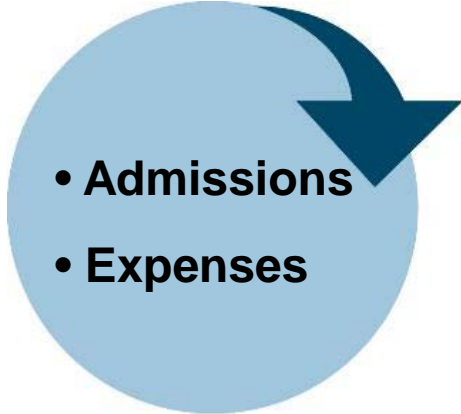
Safer and more effective drug treatment

Increased adherence to drug therapy

Decreased hospitalizations

Decreased health care costs

- 
- Safety
 - Efficacy
 - Compliance

- 
- Admissions
 - Expenses

```

#PatientName=XXXXXX
#DMETfile=DMET_8170.dmet_GT.txt
#TubeNumber=8170
#PatientID=(0000)02XXXX
#SampleType=PGEN DNA
#TranslationFile=DMET_Plus.v1.20101104DRAFT.full.translation
#AnnotationFile=DMET_Plus.v1.20090910.dc_annot.csv
#ReporterBuild=0.8.5
#VerifiedList=VerifiedbyAffy_Nov08.marker.list.txt
PharmGKB link http://www.pharmgkb.org/do/serve?objId=PA128&objCls=Gene

```

```

Independent Copy Number 2
Called Interpretation Code UNIQ+UNK
Called Diplotypes Possible *1/*41
Called Novel Diplotypes Possible *2/UNK
Copy Number Corrected Alleles NA
Number Non-reference Probe Sets 5

```

Probe set ID	Affy Verified	Genome	Position	dbSNP	RS ID	Genotype	Call	Contributes To Alleles	Descri
AM_12261	Y	Ch22:40853887	rs16947	C/T	Ref/Var	*2,*8,*11,*12,*14A,*14B,*17,*19,*20,*21,*29,*40,*41,*4			
AM_12257	Y	Ch22:40853749	rs28371725	G/A	Ref/Var	*41		CYP2D6*41_2988G>A(SpliceDefect)	
AM_15502	Y	Ch22:40854118	rs11358364	G/T	Ref/Var	-		CYP2D6_1581G>C(V136V)	
AM_12277	Y	Ch22:40854118	rs11358364	G/C	Ref/Var	-		CYP2D6_1581G>C(V136V)	
AM_12247	Y	Ch22:40852557	rs1135840	G/C	Ref/Var	S486T		CYP2D6_4180G>C(S486T)	
Number Reference Probes	5								
Probe Set ID	Affy Verified	Genome	Position	dbSNP	RS ID	Genotype	Call	Contributes To Alleles	Descri
AM_12285	Y	Ch22:40856638	rs1065852	C/C	Ref/Ref	*4,*10,*14A,*56B,*64		CYP2D6_100C>T(P34S)	
AM_12284	Y	Ch22:40856638	rs1065852	T/T	Ref/Ref	*2,*124G>A(G42R)			
AM_12283	N	Ch22:40856600	rs72549357	T/T	Ref/Ref	*15		CYP2D6*15_137inst	
AM_12281	Y	Ch22:40855856	rs5030863	G/G	Ref/Ref	*11		CYP2D6*11_883G>C(SpliceDefect)	
AM_12280	Y	Ch22:40855716	rs28371706	C/C	Ref/Ref	*17,*40,*64		CYP2D6_1023C>T(T107I)	
AM_12278	N	Ch22:40855078	rs61736512	G/G	Ref/Ref	*29		CYP2D6*29_1659G>A(V136I)	
AM_12276	Y	Ch22:40855030	rs5030655	T/T	Ref/Ref	*6		CYP2D6*6_1707delT	
AM_12275	N	Ch22:40854979	Ch22:40854979	rs5030865	G/G	Ref/Ref	*14A,*14B,*8	CYP2D6*14or*8_	
AM_12274	Y	Ch22:40854891	rs3892097	G/G	Ref/Ref	*4		CYP2D6*4_1846G>A(SpliceDefect)	
AM_12272	Y	Ch22:40854873	rs72549356	-/-	Ref/Ref	*40		CYP2D6*40_1863ins(TTTCGCCCC)2	
AM_12270	Y	Ch22:40854763	rs72549354	-/-	Ref/Ref	*20		CYP2D6*20_1973insG	
AM_12268	Y	Ch22:40854195	rs72549353	AACT/AACT	Ref/Ref	*19		CYP2D6*19_2539delAACT	
AM_12267	Y	Ch22:40854188	rs35742686	A/A	Ref/Ref	*3		CYP2D6*3_2549delA	
AM_12266	Y	Ch22:40854157	rs72549352	-/-	Ref/Ref	*21		CYP2D6*21_2573insC	
AM_12265	Y	Ch22:40854147	rs72549351	GACT/GACT	Ref/Ref	*38		CYP2D6*38_2587delGACT	
AM_12264	Y	Ch22:40854120	rs5030656	AGA/AGA	Ref/Ref	*9		CYP2D6*9_2615delAAG	
AM_12259	Y	Ch22:40853802	rs5030867	A/A	Ref/Ref	*7		CYP2D6*7_2935A>C(H324P)	
AM_12258	Y	Ch22:40853787	rs72549349	G/G	Ref/Ref	*44		CYP2D6*44_2950G>C(SpliceDefect)	
AM_12255	Y	Ch22:40853554	rs59421388	G/G	Ref/Ref	*29		CYP2D6*29_3183G>A(V338M)	
AM_12254	Y	Ch22:40853536	rs72549347	C/C	Ref/Ref	*56A,*56B		CYP2D6*56_3201C>T(R344X)	
AM_12252	Y	Ch22:40853477	rs72549346	-/-	Ref/Ref	*42		CYP2D6*42_3259insGT	
AM_12248	Y	Ch22:40852603	rs1135836	T/T	Ref/Ref	*18		CYP2D6*18_4125dupGTGCCCACT	
AM_15506	N	Ch22:40858920	rs28360521	G/G	Ref/Ref	-		CYP2D6_-2178G>A	
AM_15503	N	Ch22:40858703	Ch22:40858703	-	C/C	Ref/Ref	-	CYP2D6_-1961C>G>A	
AM_12291	Y	Ch22:40858326	rs1080985	C/C	Ref/Ref	-		CYP2D6_-1584C>G	

Her CYP2C19 genotype is consistent with an increased risk of therapeutic failure with paroxetine. Please consider alternative therapies when initiating drug therapy to treat depression.

Pharmacogenetic translation process

Genotype

Examples: rs16947 CT

Haplotype

Examples: *1, *2, *3, *17

Diplotype

Examples: *1/*1, *1/*2, *2/*2

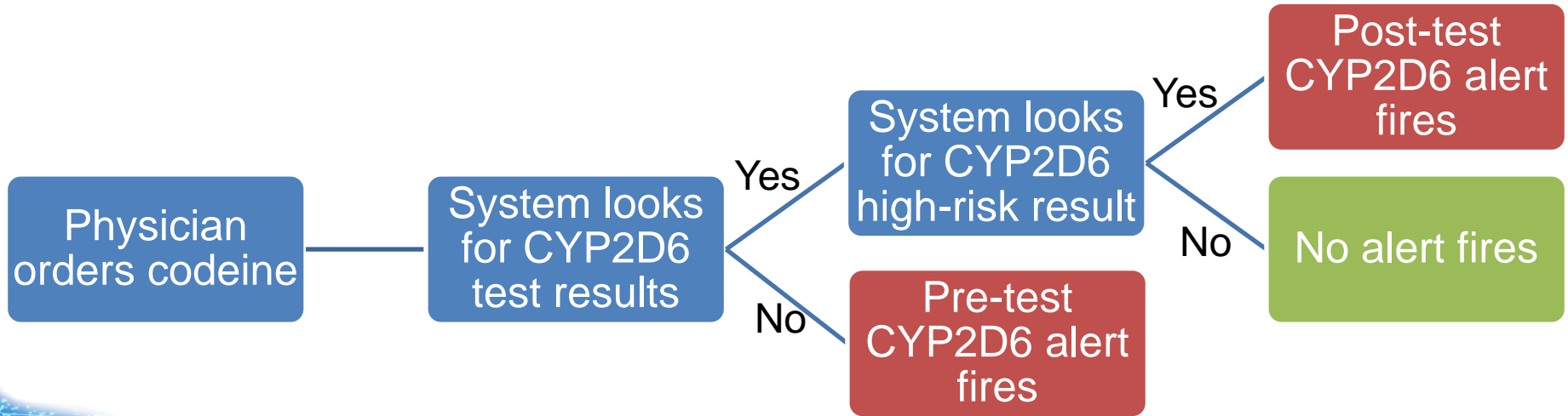
Phenotype

Example: poor metabolizer (PM)

Therapeutic Recommendation



One Drug/One Gene CDS Decision Tree



NorthShore Post-test Alert - 2

BestPractice Advisory - Stasinos,Pgx

ActX Genomic alert

Amitriptyline



Increased risk of side effects. Recommend decreasing dose.

Supporting Evidence: **Strong** | Patient Data Reliability: **A** | Genotype: *CYP2C19*2 / CYP2C19*2*

Additional Information

Remove the following orders?

Remove

Keep

Amitriptyline 100 MG PO Tab Disp-30 Tab, R-11, QHS starting 6/28/2016, E-Prescribing, Take 1 Tab by mouth every night at bedtime.
Amitriptyline

The following actions have been applied:

✓ Sent: This advisory has been sent via In Basket



Acknowledge Reason

past t/e

recommended

interaction

OTHER

✓ Accept

External Viewer

PGX STASINOS | Born 01/17/1959 | Age: 57 | F

Medications Risks Conditions Carrier Status

Check Medication Medication List

See [list](#) of what we check for.
Use Check Medication tab to look up by brand name.

Amitriptyline
Increased risk of side effects. Recommend decreasing dose.
Supporting Evidence: **Strong** | Patient Data Reliability: **A** | Genotype: **CYP2C19*2 / CYP2C19*2**
Common brand names: domical, duo-vil, elavil, endep, etrafon, lentizol, limbitrol, triavil, vanatrip

[Additional Information](#)

Carisoprodol
Increased risk of side effects. Recommend monitoring for toxicity.
Supporting Evidence: **Strong** | Patient Data Reliability: **A** | Genotype: **CYP2C19*2 / CYP2C19*2**
Common brand names: arisoprodol, carisoma, soma, soma compound, soma compound with codeine, soprodal, vanadom



Need discrete, standardized data as a foundation for CDS!

