ConectGene TESTES GENÉTICOS

Laudo de Exame Farmacogenômico

Médico Responsável: Dr. Paulo Magno do Bem Filho
CRM ES 13785

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+55 21 2135 8716

PACIENTE: Jane Doe
DT. NASC.: 01/01/1985
REF.: LCG-1111111

COLETADO: 15/10/2018
RECEBIDO: 06/11/2018
REPORTADO: 20/11/2018

AMOSTRA: Bucal
MÉDICO: Não informado
PRÁTICA: Não informado

SUMÁRIO RÁPIDO

ANTIDEPRESSIVOS

Amitriptilina (TRYPTANOL®, AMYTRYL®, ELAVIL®) **Clomipramina** (ANAFRANIL®) **Desipramina** (NORPRAMIN®) **Doxepina** (SINEQUAN®) **Imipramina** (TOFRANIL ™) **Nortriptilina** (PAMELOR ™) **Trimipramina** (SURMONTIL®)	⚠ Evite o uso de tricíclicos. Se um tricíclico for prescrito, monitore o uso terapêutico do mediamento para ajustar da dose.
Citalopram (CIPRAMIL®, PROCIMAX®, CELEXA®) **Escitalopram** (LEXAPRO®)	⊘ Considere medicamento alternativo não metabolizado pelo CYP2C19.
Duloxetina (CYMBALTA®) **Sertralina** (ZOLOFT®)	✓ Considere a dosagem da bula se não houver contraindicações.
Paroxetina (AROPAX®, PAXIL®, PEXEVA®)	⊘ Considere medicamento alternativo não metabolizado pelo CYP2C19 ou considere a redução da dose.
Venlafaxina (EFFEXOR®)	⊘ Considere medicamento alternativo não metabolizado pelo CYP2D6.

IMPORTANTE

Este Sumário Rápido fornece uma visão geral da previsão de resposta do paciente. Estas informações estão baseadas somente nas informações do genótipo e não compõe o perfil completo do paciente. A detecção ou ausência de variantes genéticas não substitui a necessidade de monitoramento terapêutico. Antes de tomar decisões clínicas ou terapêuticas, médicos devem considerar a informação contida na seção Detalhada (disponível apenas em inglês), assim como prescrições atuais, histórico familiar, sintomas apresentados e outros fatores.

✓ Nenhuma observação negativa baseada no genótipo.

⚠ Genótipo pode apresentar maior risco ou menor efetividade. Prescreva com precaução.

⊘ Genótipo pode apresentar maior risco ou menor efetividade. Considere outro medicamento.

ConectGene TESTES GENÉTICOS

Pharmacogenetic Test Results

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PATIENT: Doe, Jane (F)
DOB: 1985-01-01
PATIENT ID:

COLLECTED: 10/15/2018
RECEIVED: 11/06/2018
REPORTED: 11/30/2018

SAMPLE TYPE: Buccal
PHYSICIAN: Not Provided
PRACTICE: Not Provided

ACCESSION: LGE-9290880938

QUICK SUMMARY

ANTIDEPRESSANTS	RESULTS
Amitriptyline (ELAVIL®) Clomipramine (ANAFRANIL®) Desipramine (NORPRAMIN®) Doxepin (SINEQUAN®) Imipramine (TOFRANIL™) Nortriptyline (PAMELOR™) Trimipramine (SURMONTIL®)	⚠ Avoid tricyclic use. If a tricyclic is warranted utilize therapeutic drug monitoring to guide dose adjustment.
Citalopram (CELEXA®) Escitalopram (LEXAPRO®)	⊘ Consider alternative drug not metabolized by CYP2C19.
Duloxetine (CYMBALTA®) Sertraline (ZOLOFT®)	✓ Consider label recommended dosage if no contraindication.
Paroxetine (PAXIL®, PEXEVA®)	⊘ Consider alternative drug not metabolized by CYP2D6 or consider reduced dose.
Venlafaxine (EFFEXOR®)	⊘ Consider alternative drug not metabolized by CYP2D6.

IMPORTANT

This Quick Summary provides a brief overview of the predicted response of the patient. This information is based solely on the genotype information and is not based on a complete patient profile. Detection or absence of variants does not replace the need for therapeutic monitoring. Physicians should consider the information contained in the Details section, as well as consider current prescriptions, family history, presenting symptoms, and other factors before making any clinical or therapeutic decisions.

✓ No negative assertions based on genotype.

⚠ Genotype may present increased risk or decreased effectiveness; prescribe with caution.

⊘ Genotype may present increased risk or decreased effectiveness; select alternative drug.

GENE SUMMARY

GENE	GENOTYPE	PHENOTYPE
CYP2D6	*3/*4	⊘ Poor Metabolizer
CYP2C19	*1/*17	⚠ Rapid Metabolizer
COMT	VAL/VAL	✓ Normal Stimulant Response

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DETAILED INFORMATION

Amitriptyline	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Amitriptyline. Patient may have increased metabolism of Amitriptyline when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Amitriptyline. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		
	ABCB1 rs2235015 C/A (HET)		Evidence ★
	Consider label recommended dosage of Amitriptyline if no contraindication.		
Citalopram	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts that the patient is a Rapid Metabolizer of Citalopram. The patient may have increased metabolism when compared to extensive metabolizers. Lower plasma concentrations will increase probability of pharmacotherapy failure. The CPIC Guideline recommends considering an alternative drug not predominantly metabolized by CYP2C19.		
	GRIK4 rs1954787 T/C (HET)		Evidence ★★★
	Consider label recommended dosage of Citalopram if no contraindication.		
	HTR2A rs7997012 A/A (WT)		Evidence ★★
	Consider label recommended dosage of Citalopram if no contraindication.		
	HTR2A rs6313 G/G (WT)		Evidence ★
	Patients with the wild-type genotype and major depression may have increased risk of heart palpitations when treated with citalopram as compared to patients with the homozygous genotype.		
	ABCB1 rs2235015 C/A (HET)		Evidence ★
	Consider label recommended dosage of Citalopram if no contraindication.		
Clomipramine	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Clomipramine. Patient may have increased metabolism of Clomipramine when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Clomipramine. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		

DETAILED INFORMATION

Desipramine	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Desipramine. Patient may have increased metabolism of Desipramine when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Desipramine. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		
Doxepin	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Doxepin. Patient may have increased metabolism of Doxepin when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Doxepin. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		
Duloxetine	DRD3 rs963468 G/A (HET)		Evidence ★
	Consider label recommended dosage of Duloxetine if no contraindication.		
Escitalopram	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts that the patient is a Rapid Metabolizer of Escitalopram. The patient may have increased metabolism when compared to extensive metabolizers. Lower plasma concentrations will increase probability of pharmacotherapy failure. The CPIC Guideline recommends considering an alternative drug not predominantly metabolized by CYP2C19.		
	HTR2C rs6318 C/C (HOM)		Evidence ★
	Male patients with this genotype and neuropathic pain may have increased pain relief when treated with escitalopram as compared to patients with the wild-type genotype.		
	CYP1A2 rs2069526 T/T (WT) CYP1A2 rs4646425 C/C (WT) HTR2A rs9316233 C/C (WT)	CYP1A2 rs4646427 T/T (WT) HTR2A rs6311 C/C (WT)	Evidence ★
	Consider label recommended dosage of Escitalopram if no contraindication.		

DETAILED INFORMATION

Imipramine	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Imipramine. Patient may have increased metabolism of Imipramine when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Imipramine. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		
Nortriptyline	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Nortriptyline. Patient may have increased metabolism of Nortriptyline when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.		
	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Nortriptyline. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.		
	GNB3 rs5443 C/C (WT)		Evidence ★
	Patients with the wild-type genotype and major depression who are treated with nortriptyline may have less improvement in neurovegetative symptoms and an increased likelihood of Sleep Initiation and Maintenance Disorders. These patients are at decreased risk for weight gain as compared to patients with the homozygous genotype.		
	ABCB1 rs1045642 G/G (HOM)		Evidence ★
	Consider label recommended dosage of Nortriptyline if no contraindication.		
Paroxetine	CYP2D6 *3/*4	*Poor metabolizer.*	Evidence ★★★
	The genotype predicts that the patient is a Poor Metabolizer of Paroxetine. The patient may have greatly reduced metabolism when compared to extensive metabolizers, and higher plasma concentrations may increase the probability of side effects. The CPIC Guideline recommends selecting alternative drug not predominantly metabolized by CYP2D6 or if paroxetine use warranted, consider a 50% reduction of recommended starting dose and titrate to response.		
	HTR1A rs6295 C/G (HET)		Evidence ★★
	Patients with the heterozygous genotype with panic disorder who are treated with paroxetine may have a reduced response at 4 weeks of treatment as compared to patients with the homozygous genotype.		

DETAILED INFORMATION

	CYP1A2 rs2470890 C/T (HET) Patients with the heterozygous genotype and major depressive disorder who are treated with paroxetine may be less likely to experience remission as compared to patients with the wild-type genotype.	Evidence ★
	COMT rs4680 G/G (WT) *Normal stimulant response.* Patients with this genotype who have major depression may have a decreased response to paroxetine compared to the homozygous genotype.	Evidence ★
	HTR2A rs6313 G/G (WT) Patients with the wild-type genotype and depression who are treated with paroxetine may have an increased risk of adverse drug reactions as compared to patients with the heterozygous or homozygous genotype.	Evidence ★
	HTR1A rs10042486 C/T (HET) Patients with the heterozygous genotype and Major Depressive Disorder who are treated with paroxetine may have decreased response to treatment as compared to patients with the wild-type genotype.	Evidence ★
	CYP1A2 rs762551 C/A (HET) Patients with heterozygous genotype may require an increased dose of paroxetine and may have an increased risk of fatigue when treated with paroxetine as compared to patients with the wild-type genotype.	Evidence ★
	ABCB1 rs2235015 C/A (HET) CYP1A2 rs4646427 T/T (WT) DRD3 rs6280 C/T (HET) Consider label recommended dosage of Paroxetine if no contraindication.	Evidence ★
Sertraline	CYP2C19 *1/*17 *Rapid metabolizer.* HTR1A rs6295 C/G (HET) Consider label recommended dosage of Sertraline if no contraindication.	Evidence ★★★
Trimipramine	CYP2C19 *1/*17 *Rapid metabolizer.* The genotype predicts the patient is a CYP2C19 Rapid Metabolizer of Trimipramine. Patient may have increased metabolism of Trimipramine when compared to extensive metabolizers. The CPIC Guidelines recommends considering an alternative drug not metabolized by CYP2C19. If a tricyclic is warranted, utilize therapeutic drug monitoring to guide dose adjustments.	Evidence ★★★
	CYP2D6 *3/*4 *Poor metabolizer.* The genotype predicts the patient is a CYP2D6 Poor Metabolizer of Trimipramine. Patient may have a greatly reduced metabolism of tricyclics to less active compounds when compared to extensive metabolizers. Higher plasma concentrations will increase the probability of side effects. The CPIC Guidelines recommend avoiding tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a tricyclic is warranted, consider 50% reduction of recommended starting dose. Utilize therapeutic drug monitoring to guide dose adjustments.	Evidence ★★★
Venlafaxine	CYP2D6 *3/*4 *Poor metabolizer.* The genotype predicts that the patient is a Poor Metabolizer of venlafaxine. The Dutch Pharmacogenetics Working Group Guideline indicates that there is insufficient data to allow calculation of dose adjustment, and recommends selecting an alternative drug (e.g., citalopram, sertraline) or adjust dose to clinical response and monitor (O-desmethyl)venlafaxine plasma concentration.	Evidence ★★★

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	COMT rs4680 G/G (WT)	*Normal stimulant response.*	Evidence ★
	Patients with this genotype who are treated for Anxiety Disorders may have a decreased response to venlafaxine. However, patients with this genotype who are treated for Depressive Disorder may have an increased response to venlafaxine		
	HTR2A rs7997012 A/A (WT)		Evidence ★
	Patients with the wild-type genotype may be less likely to respond to venlafaxine as compared to patients with the heterozygous or homozygous genotype.		
	ABCB1 rs2235015 C/A (HET)	ABCB1 rs1045642 G/G (HOM)	Evidence ★
	Consider label recommended dosage of Venlafaxine if no contraindication.		

KEY FOR VARIANT-DRUG COMBINATION EVIDENCE

★★★ Replicated in multiple studies with statistical significance and strong effect size.
★★ Replicated in multiple studies with and without statistical significance and effect size may be minimal.
★ Not yet replicated or replicated but lacking clear evidence of an association.
Notable information is available and special considerations may be of interest when prescribing for this genotype.
Literature does not indicate additional risks, benefits, or prescription changes to consider for this genotype.

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REPORTED GENOTYPES

This panel performs genotyping analysis on key genes and variant hot-spot regions, focused on analyzing loci documented as altering the effectiveness of drug metabolism. Key genotyping results include the following:

ABCB1
rs1045642:G/G Hom
rs2235015:C/A Het

COMT
rs4680:G/G Wild

CYP1A2
rs2069526:T/T Wild
rs2470890:C/T Het
rs4646425:C/C Wild
rs4646427:T/T Wild
rs762551:C/A Het

CYP2C19
CYP2C19 *1/*17
rs4244285:G/G Wild
rs4986893:G/G Wild
rs28399504:A/A Wild
rs56337013:C/C Wild
rs72552267:G/G Wild
rs72558186:T/T Wild
rs41291556:T/T Wild
rs17884712:G/G Wild
rs6413438:C/C Wild
rs55640102:A/A Wild
rs12248560:C/T Het

CYP2D6
CYP2D6 *3/*4
rs16947:G/G Hom
rs1135840:G/C Het
rs35742686:T/- Het
rs1135824:T/T Wild
rs1065852:G/A Het
rs3892097:C/T Het
rs5030655:A/A Wild
rs5030867:T/T Wild
rs5030865:C/C Wild
rs5030656:CTT/CTT Wild
rs5030863:C/C Wild
rs5030862:C/C Wild
rs72549357:C/C Wild
rs28371706:G/G Wild
rs59421388:C/C Wild
rs769258:C/C Wild
rs28371725:C/C Wild
rs28371696:C/C Wild
rs28371717:C/C Wild

DRD3
rs6280:C/T Het
rs963468:G/A Het

GNB3
rs5443:C/C Wild

GRIK4
rs1954787:T/C Het

HTR1A
rs10042486:C/T Het
rs6295:C/G Het

HTR2A
rs7997012:A/A Wild
rs9316233:C/C Wild
rs6313:G/G Wild
rs6311:C/C Wild

HTR2C
rs6318:C/C Hom

Reported genotype calls are all displayed with respect to the positive DNA strand. Variants indicated as homozygous (Hom) or heterozygous (Het) differ from the GRCh37/hg19 reference sequence (Wild). This report is limited to the following star-alleles: CYP2D6: *1, *2, *3, *3B, *4, *5, *6C, *6, *7, *8, *9, *10, *11, *12, *14A, *14B, *15, *17, *29, *33, *35A, *41, *45A & *46. CYP2C19: *1, *2, *3, *4B, *4, *5, *6, *7, *8, *9, *10, *12 & *17. CYP2C9: *1, *2 & *3. CYP3A5: *1 & *3. CYP3A4: *1, *8, *11, *12, *13, *16, *17 & *22. TPMT: *1, *2, *3A, *3C, *3B & *4. DPYD: *1, *2, *4, *5, *13 & rs67376798A. Any genotype identified as a default star-allele (CYP2D6 *2, CYP2C19 *1, CYP2C9 *1, CYP3A5 *3, CYP3A4 *1, TPMT *1, DPYD *1) indicates the absence only of the other alleles listed and does not imply that other variants in the gene are absent. Full allele deletions and duplications are only analyzed for the CYP2D6 gene. This test does not report polymorphisms other than those specifically listed, and mutations in other genes associated with drug metabolism will not be detected. Rare diagnostic errors may occur if variations occur in primer site locations.

DISCLAIMER

The information presented on this report is provided as supplementary health information. The results presented are intended for use by a physician, pharmacist or other healthcare professional to advise a patient on the use of prescribed medications. This test is not a 510k cleared test, but managed by CMS and FDA under the Clinical Laboratory Improvement Amendment (CLIA) as a LDT. The ordering physician is responsible for the diagnosis and management of disease and decisions based on the data provided. Results are dependent on adequate specimen collection and processing.

METHODOLOGY

Genomic DNA is extracted from dry buccal swabs using magnetic particle processing. DNA from patient samples are amplified with primers specific for ABCB1, COMT, CYP1A2, CYP2C19, CYP2D6, DRD3, GNB3, GRIK4, HTR1A, HTR2A & HTR2C using Nested Patch PCR (Varley, et. al.). Positive and negative controls are used with each run. Patient samples, positive, and negative controls are sequenced using a MiSeq (Illumina). Sequences are analyzed using alignment and base call algorithms with Kailos Blue Software for the presence or absence of single nucleotide base changes, insertions and deletions. LR-PCR utilized for confirmation of CYP2D6 duplications and deletions. Results and recommendations are compiled as part of a medical report.

Genetic testing was performed in the Kailos Genetics CLIA facility at 601 Genome Way; Huntsville, Al. 35806. CLIA#: 01D2016114. Medical Director: Ronald McGlennen MD, FCAP, FACMG, ABMG.

This report was reviewed and approved for release by CLIA Lab Manager & Supervisor: Michele R. Erickson-Johnson, PhD, MB (ASCP)^{CM}

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