The Clinical Testing Laboratories, Inc.
Genesis Center - A
MSC3ARP Box 30001
3655 Research Dr
Las Cruces, NM 88003
Tel. (575) 646-3465
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A copy was sent to: Dr. Jay Neitz, PhD
University of Washington
Dept. of Ophthalmology
1959 NE Pacific St
Seattle, WA 98195

PHOTORECEPTOR VISUAL CHROMACY GENOTYPING REPORT

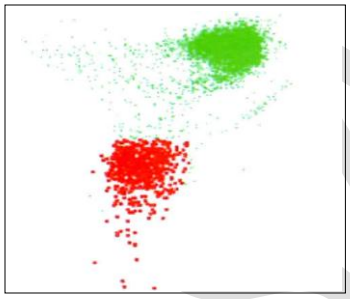
OID: U10-0000

Patient Information		**Facility Information**	
Patient Name:	**John Doe**	Facility Name:	Clinic
Patient SSN:	123-45-6789	Address:	123 Main Street
Specimen Id:	SMT00001	City/State/Zip:	Anywhere, 98765
Date of Collection:	Friday, July 02, 2011	Physician:	Ordering Physician
Date of Birth:	Sept 21, 1972	NPI:	987654321
Sex:	Male	Account Number:	200999

SPECIMEN INFORMATION

Specimen Type:	Cheek cells	**Received Date/ Time:**	7/5/2011 / 10:03AM
Collected Date:	7/2/2011	**Final Report Date/Time:**	7/6/2011 / 12:45PM

DIAGNOSIS: DEUTERANOMALOUS TRICHROMAT



Ex. Picture of Patients Photoreceptor Goes Here

Comment: Your genetic results indicate that you have an extra red photopigment gene inserted between the normal red and green photopigment genes. This pushes your green photopigment gene out of position and what would be normally green cones express a red photopigment instead. Thus, instead of having the red, green and blue cones required for normal color vision you have two populations of cone photoreceptors that express different red opsin genes and a third cone class that expresses blue opsin. The photopigments expressed in your two class of red cone absorb light in slightly different parts of the spectrum. This gives you a reduced form of red-green color vision called **Deuteranomalous Trichromacy**.

Electronically signed by:
Staff CLIA MD
CLIA ID# 32D1086620

Disclaimer:
Laboratory specimens were analyzed using a MALDI-TOF (from Sequenom, Inc.) running 2 multiplex assays which together detects 5 nucleotide variants in a multiplex polymerase chain reaction and allele-specific primer extension format. The performance of the assay for the opsin genes for use with the MALDI-TOF MassArray System was validated by the Clinical Testing Laboratories, Inc. and developed by Genevolve Vision Diagnostics®, Inc. sold under the Eyedox™ by Dr. Neitz brand.

This assay has not been approved by the FDA (Food and Drug Administration). The assay is for clinical use and should not be viewed as experimental or for "research use only".

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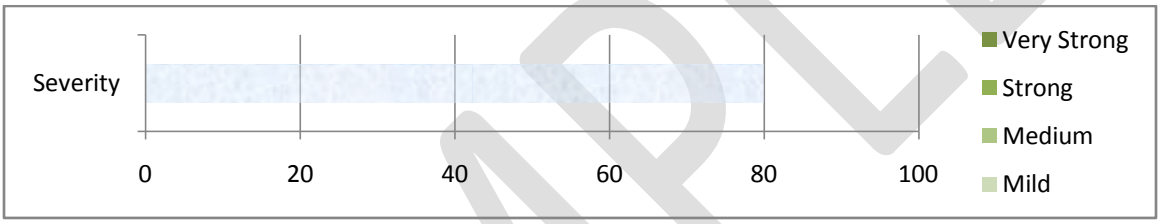
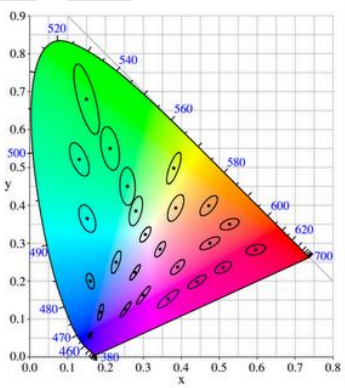
SEVERITY: MEDIUM

Deuteranomalous Trichromacy is by far the most common form of congenital color vision deficiency; it affects about 1 out of every 20 men in the United States and 1 out of every 400 women. The sensations of red and green depend on the red and green cones. People who are missing either the red or green cones do not have the sensations of either red or green experienced by normal trichromats. You do have the capability to sense both red and green. This ability is based on the differences in spectral sensitivity between your two red cone types. In people with your condition, the amount of red-green color vision depends how different the absorption spectra of your cones are. Your genetic results indicate that your red cones are moderately far apart. This results in a **Medium** loss of red and green sensations.



The Above Chart is a Representation – Actual Chart May Appear Different

Deuteranomalous trichromacy is considered to be a stationary disorder; it does not get better or worse. Some times deuteranomalous trichromats are incorrectly labeled as "red-green color blind." This is inaccurate. You sense both red and green and can tell pure red apart from pure green as well as anyone.



Ex. Customized Mac Adams Ellipse

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TREATMENT PLAN: DEUTERANOMALOUS TRICHROMAT

However, you have trouble distinguishing some colors that have a small amount of red or green in them that can be seen by normal trichromats but are below threshold for your eye. A common example is distinguishing olive green from brown. Olive green is close to brown but has a small amount of greenish hue. Your lower sensitivity to green makes these two colors very difficult to distinguish. Similarly, burnt orange is close to brown but has a little red in it that may be below your detection threshold. Thus, burnt orange, olive green and brown all look similar to you. Very light pastel greens and pastel pinks that are nearly white or gray also cause a problem for you because the amount of green or red may be below your threshold. The difference between blue and purple is that purple has some amount of red hue in it. You have trouble distinguishing purple from blue especially shades of bluish-purple that have only a small amount of red hue in them. The same is true of your ability to tell greenish blues from blue because of your relative insensitivity to the green hue in them.

People with your condition are able to accommodate to their color vision deficiency especially once they are aware of how their color vision differs from normal, as outlined here. Some of the hazards are a reduced ability to tell when you, or someone else, is getting a sunburn because of your reduced ability to detect small amounts of red hue. It is also harder for you to tell when meat is cooked by its color. Your sense of what colors look appealing may differ from people with normal color vision. Your ability to do critical tasks that require you to distinguish subtly different colors especially if there is a demand to make those discriminations very quickly is reduced. It is not usually associated with any other eye problems except the incidence of nearsightedness may be slightly higher.

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OCCUPATIONAL: DEUTERANOMALOUS TRICHROMAT

Professions that require perfect color vision do not employ deuteranomalous trichromats. There is also some amount of discrimination against deuteranomalous trichromats because some old-fashion tests of color vision do not separate people with red-green color anomalies from those with red-green colorblindness. However, some employers use tests that do distinguish deuteranomalous trichromacy from more complete forms of red-green color blindness (called dichromacy) and they only reject the dichromats. In the future, as genetic testing for red-green colorblindness becomes the standard, there will be less confusion among employers about red-green color blindness verses deuteranomalous trichromacy.

FAMILY HISTORY: DEUTERANOMALOUS TRICHROMAT

Deuteranomalous trichromacy, like all forms of red-green color vision deficiency, is X-chromosome linked. Boys inherit it from their mothers and pass it on to their girl children who will not be colorblind unless they also get a gene for a deutan-type color vision defect from their mothers.

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INTRODUCTION: COLOR VISION GENETICS

For our color vision, we rely on a class of cells in our retinas called cones. There are three types of cones, differentiated by the wavelengths of light to which they are most sensitive, roughly corresponding to blue, green, and red. The technical names of these cells are short-wavelength sensitive or *S-cones* for the blue cones, medium-wavelength sensitive or *M-cones* for the green, and long-wavelength sensitive or *L-cones* for the red. People who have functional cells of all three types have normal color vision, termed *trichromacy*, while those lacking any of the three have a congenital color vision deficiency.

The key genes that determine cone type are called *opsin genes*, which come in three types corresponding to each of the three cone cell types. The opsin genes each encode the amino acid sequence of a protein that is involved in the absorption of photons by the cone cell, a step that begins the process that underlies our vision. Each cone has only one of the three opsin genes switched on, and it is the presence of this opsin that determines which type of cone a cell becomes. If any opsin gene has a mutation that makes it nonfunctional or if it is missing entirely, this will lead to an absence of the corresponding cone type from the retina and thus to color vision deficiency.

The S-cone opsin gene is located on chromosome 7 and is not often mutated. Color vision disorders resulting from defective S-cones are very rare, affecting only about one in ten thousand people. Conversely, mutations involving the M-cone opsin gene and L-cone opsin gene are very common, with approximately one in twelve men and one in two hundred women affected by some form of red-green color blindness in the United States. The gender difference is due to the fact that the M-cone opsin gene and the L-cone opsin gene are located together on the X chromosome, giving women two opportunities to have functional copies of both genes while men have only one opportunity. Because of the rarity of S-cone opsin gene mutations, this genetic assay focuses on the M-cone and L-cone opsin genes.

The opsin genes that are expressed in the M and L cones are among the most variable genes in the human genome. This means that rather than a single M-cone opsin gene and single L-cone opsin gene, there exists a family of each in which each member has a slightly different wavelength of light that it absorbs best. Not only are there many different versions of the M- and L-cone opsin genes, but a single X chromosome can have multiple copies of either gene.

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INTRODUCTION CONTINUED: COLOR VISION GENETICS

Usually, the L-cone opsin genes precede M-cone opsin genes on the chromosome. Among people with normal color vision, a single L-cone opsin gene can be followed on the X-chromosome by as few as one M-cone opsin gene or as many as five or six. The most common arrangement of genes is one L-cone opsin gene followed by two M-cone opsin genes.

As long as the array of opsin genes can become, only the first two genes are ever switched on. Thus people with multiple L-cone opsin genes at the beginning of the array will not have M-cone cells in their retinas no matter how many M-cone opsin genes they possess. People with S-cones and two different L-cone subtypes in their retinas have *deuteranomalous* color vision. People who lack L-cone opsin genes but who have multiple, different M-cone opsin genes will have S-cones and two different M-cone subtypes in their retinas and have *protanomalous* color vision. Both deuteranomalous and protanomalous people will have some ability to discriminate a wider range of colors, depending on how different their two L- or two M-cone subtypes are.

People with only a single opsin gene (or multiple copies of single version of the gene) on their X chromosome are called *dichromats*, referring to the fact that there will only be two cone types present in their retinas. Dichromats with only an M opsin gene are called *protanopes* and will have only S- and M-cones. Dichromats with only an L opsin gene are called *deuteranopes* and will have only S- and L-cones. Dichromacy is the most severe form of red-green color blindness, with only variations of yellow and blue hues distinguishable.

The genetic assay determines how many of each type of cone opsin gene is present on your X-chromosome and can indicate whether multiple copies of either M- or L- cone opsin genes, if present, are different versions of the gene with different peaks of light absorption. From this, we can tell if you have normal color vision, protanamolous color vision, deuteranomalous color vision, protanopia, or deuteranopia.