Diagnosing tritan defects in a clinical environment

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Abstract
Tritan color vision defects have been found in four individuals of a five member family, revealing an autosomal dominant mode of inheritance with a variation of test results within the family. Three males have been observed to be carrying both inherited tritan and deutan defects. Several popular color vision tests are shown to differ in the ability to detect color defective individuals. Arrangement color vision tests failed to detect deutan defects and various plate tests are solely designed to screen for red/green defects. Primary interest was given to those standard tests used in the clinical setting.

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DIAGNOSING TRITAN DEFECTS IN A CLINICAL ENVIRONMENT

By

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JEFFREY FRIES
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Advisor:

Robert Yolton, Ph.D., O.D.
BIOGRAPHIES

Jeffrey Fries graduated with a Bachelor of Science degree from Moorhead State University in Moorhead, Minnesota. In May of 1997 Jeffrey will graduate from Pacific University College of Optometry with a Doctor of Optometry degree. In the future, Jeffrey hopes to join a group practice somewhere in the western Unites States.

Nicole Kish graduated with a Bachelor of Science degree from Eastern Illinois University in Charlestown, Illinois. In May of 1997 Nicole will graduate from Pacific University College of Optometry with a Doctor of Optometry degree. In the future, Nicole plans to practice in the Dallas/Ft. Worth, Texas area and obtain her pilots license.
ABSTRACT

Tritan color vision defects have been found in four individuals of a five member family, revealing an autosomal dominant mode of inheritance with a variation of test results within the family. Three males have been observed to be carrying both inherited tritan and deutan defects. Several popular color vision tests are shown to differ in the ability to detect color defective individuals. Arrangement color vision tests failed to detect deutan defects and various plate tests are solely designed to screen for red/green defects. Primary interest was given to those standard tests used in the clinical setting.
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Abstract
Tritan color vision defects have been found in four individuals of a five member family, revealing an autosomal dominant mode of inheritance with a variation of test results within the family. Three males have been observed to be carrying both inherited tritan and deutan defects. Several popular color vision tests are shown to differ in the ability to detect color defective individuals. Arrangement color vision tests failed to detect deutan defects and various plate tests are solely designed to screen for red/green defects. Primary interest was given to those standard tests used in the clinical setting.

Introduction
The familial nature of color vision defects has been known for some time. Went and Pronk (1985) conducted a study on the genetics of tritan defects and included a historical review of our knowledge on color vision. There are at least two main gene loci on the X chromosome involved with color vision: the deutan (green) locus and the protan (red) locus. The third group of color vision defects is the tritan (blue) defect which is inherited via an autosomal dominant gene(s) (Went and Pronk 1985).

Individuals with congenital red-green color defects comprise about 10 percent of the U.S. male population. Red-green defects have X-chromosome linked recessive inheritance meaning that the defect is carried on the sex chromosomes. The genes occur at a locus on the X chromosome for which no corresponding gene occurs on the Y chromosome (Pokorny et al. 1981; Alpern, M. 1983). Therefore, red-green defects are distributed among the male population.

According to a study by Kalmus (1955), the frequency of tritan defects is about 1 in 20,000 in the general population, but Went and Pronk (1985) estimated the incidence to be as high as 2 per 1000 based on results from three familial screening series. Tritan defects show autosomal dominant
inheritance; the trait is carried on any except the sex chromosomes (Pokorny et al. 1981). Therefore, tritan defects have an equal sex distribution (Went and Pronk 1985; Alpern 1976).

Before the early 1970's, researchers have suggested that inherited tritan defects do not exist but are mimicked by mild optic atrophy. This view has been proven incorrect by the studies of Smith et al. (1973), Went et al. (1974), and van de Merendonk and Went (1980) (Went and Pronk 1985).

The main problem with detecting tritan defects is that many of the commonly used color vision tests, such as the Ishihara Pseudo-Isochromatic Plate Test, are only designed to detect red-green defects. The AOHRR Plate Test, Farnsworth F2 Tritan Plate Test, and the City University Color Plate Test screen for tritan defects, but only the City University test is currently produced. (Went and Pronk 1985; Pokorny et al. 1981). The arrangement tests, such as the Farnsworth Panel D-15 Test and the Farnsworth-Munsell 100-Hue Test, are designed to detect protan, deutan, and tritan defects (Pokorny et al. 1981). An anomaloscope can permit a definitive diagnosis of color vision defects; however, anomaloscopes are primarily found in research settings and most clinicians do not have access to an instrument (Pokorny et al. 1981).

The goal of this project was to administer color vision tests to members of a genetically related family in order to: 1) diagnose any color vision defects, 2) detect an inheritance pattern in the color vision defects, and 3) determine which standard tests are most effective in the diagnosis of color vision defects. With the exception of the anomaloscope, which was required in this study for the definitive diagnoses of color vision defects, all color vision tests were chosen based on their standard availability in the clinical setting.

Methods
The motivation for the study came from a subject who reported that he, his father, and two brothers were color defective to varying degrees. The subject, who will be referred to as son 1, reported an eight year history of recreational drug use; however, military and vocational color vision tests were failed by this subject prior to the drug use. A complete vision examination revealed no signs of pathology; the fundi and optic discs of son 1 appeared normal with no disc pallor, abnormal macular pigmentation, or degenerative macular changes. Uncorrected Snellen visual acuities were 20/20 (6/6) OU, OD, OS distance and near. Based on these results and the subject's color vision test
history, it was concluded that the color defect reported by the subject was not secondary to any pathology or drug use.

The five member family consisted of the 65 year old father, 62 year old mother, and son 1, son 2, and son 3 who were 35, 30, and 44 years old, respectively. In addition to son 1, complete vision examinations were performed on the father and mother. Sons 2 and 3 were given partial examinations which included near and far visual acuity determinations, near and far cover tests, versions, pupil testing, confrontational fields, and direct ophthalmoscopy. Visual acuity was measured using distance and near Snellen charts.

All family members had best corrected visual acuities of at least 20/20 (6/6) OD, OS, OU with no signs of ocular pathology. None of the subjects reported an acute onset of their color vision defect, but all of the males did recall being aware of color confusions during childhood.

The following color vision tests were used: Ishihara Pseudo-Isochromatic Plate Test, Farnsworth Panel D-15 arrangement test (D-15), Farnsworth-Munsell 100-Hue arrangement test (100-Hue), and the Pickford-Nicolson Anomaloscope.

The Ishihara plate test was administered using a full-spectrum lamp for illumination. Each family member was tested monocularly at a distance of 75 cm with each of the 17 plates held at approximately a right angle to the line of sight.

The D-15 test was administered to all family members, each eye separately, using a full spectrum lamp for illumination. The mother was excluded from the remaining tests based on her error-free results on the previous two tests.

The 100-Hue is designed to test hue discrimination among individuals with normal color vision and to measure the areas of color confusion in color defective individuals (Pokorny et al. 1981). Testing conditions of the 100-Hue test included monocular viewing and full spectrum illumination. The color axis graph was plotted for the four male members of the family and an Error Score was computed according to the instructions provided with the test.

Anomaloscopes are optical instruments in which the observer must manipulate stimulus control knobs to match two colored fields with respect to color and brightness. The Pickford-Nicolson anomaloscope is a filter type
An anomaloscope that uses broad-band filters to provide primary and test wavelengths. The Pickford-Nicolson anomaloscope is capable of determining the Rayleigh match (red/green), the Engelking-Trendelenburg match (blue/green), and the Pickford-Lakowski match (yellow/blue) (Pokorny et al. 1981).

Anomaloscope testing was performed with the four male members of the family using instrument instructions. The instrument presents a circular split field with the color and brightness of both halves varied independently by the use of interference and neutral density filters (Pokorny et al. 1981; Pickford and Lakowski 1960). The test field appears in the left half of the split field; its radiance can be varied from dark at scale setting zero to its maximum at scale setting 82. The primary mixture appears in the right half with the relative proportion of the primaries adjusted continuously. The mixture ratios are read from a scale on the top of the instrument. The subject is asked to report when the two halves appeared equal or different in color. This adjustment process was repeated until a reliable range of equality settings was found for each filter combination. Using the high and low scale readings for each equal color mixture, a Mid-Matching Point (MMP) was calculated and compared to the mean for the particular color match. An absolute difference between the mean and the MMP of 2 standard deviations (SD's) indicates a color deficiency (Pickford and Lakowski 1960).

Table 1: Anomaloscope Mean and Subject Standard Deviations

<table>
<thead>
<tr>
<th>SUBJECT</th>
<th>COLOR MIXTURE</th>
<th>MEAN</th>
<th>STANDARD DEVIATION</th>
<th>MMP</th>
<th># of SD from MEAN</th>
</tr>
</thead>
<tbody>
<tr>
<td>Father</td>
<td>R/G</td>
<td>37.40</td>
<td>1.25</td>
<td>35.50</td>
<td>2.00</td>
</tr>
<tr>
<td>Father</td>
<td>G/B</td>
<td>46.90</td>
<td>1.47</td>
<td>35.00</td>
<td>8.00</td>
</tr>
<tr>
<td>Father</td>
<td>Y/B</td>
<td>38.60</td>
<td>2.33</td>
<td>34.00</td>
<td>2.00</td>
</tr>
<tr>
<td>Son 1</td>
<td>R/B</td>
<td>37.40</td>
<td>1.25</td>
<td>44.00</td>
<td>5.00</td>
</tr>
<tr>
<td>Son 1</td>
<td>G/B</td>
<td>46.90</td>
<td>1.47</td>
<td>35.00</td>
<td>8.00</td>
</tr>
<tr>
<td>Son 1</td>
<td>Y/B</td>
<td>38.60</td>
<td>2.33</td>
<td>30.00</td>
<td>3.70</td>
</tr>
<tr>
<td>Son 2</td>
<td>R/G</td>
<td>37.40</td>
<td>1.25</td>
<td>40.00</td>
<td>2.00</td>
</tr>
<tr>
<td>Son 2</td>
<td>G/B</td>
<td>46.90</td>
<td>1.47</td>
<td>34.50</td>
<td>8.00</td>
</tr>
<tr>
<td>Son 2</td>
<td>Y/B</td>
<td>38.60</td>
<td>2.33</td>
<td>35.50</td>
<td>1.30</td>
</tr>
<tr>
<td>Son 3</td>
<td>R/G</td>
<td>37.40</td>
<td>1.25</td>
<td>42.50</td>
<td>4.00</td>
</tr>
<tr>
<td>Son 3</td>
<td>G/B</td>
<td>46.90</td>
<td>1.47</td>
<td>26.00</td>
<td>14.00</td>
</tr>
<tr>
<td>Son 3</td>
<td>Y/B</td>
<td>38.60</td>
<td>2.33</td>
<td>26.50</td>
<td>5.00</td>
</tr>
</tbody>
</table>
Results

The number of SD's from the mean for each subject on the anomaloscope are displayed in Table 1. The father and all of his offspring displayed a green/blue (G/B) defect; the largest of which was a deviation of 14 units for son 1. Additionally, sons 1 and 3 were found to have red/green (R/G) and yellow/blue (Y/B) defects on the anomaloscope. Figure 1 shows that the largest deviation from the mean was associated with a G/B defect for all subjects.

Figure 1: Subject Standard Deviations on Anomaloscope

![Graph showing standard deviations for different subjects and color defects](image)

The scores and/or results for each color vision test are included in Table 2. The table shows that the mother and father passed the Ishihara plate test; however, all three sons failed this red/green screening test. The Ishihara test result for son 2 is in contrast to the anomaloscope results and will be addressed later.

Son 1 failed to perceive the figures or numbers on 14 of the 17 plates and was best described as a strong deuteranope. Son 2 and son 3 missed a total of 9 and 7 plates, respectively, and were described as mild deuteranopes based on Ishihara scoring instructions. The red/green sensitive Ishihara test successfully detected all of the red/green defects, but provided no information regarding tritan defects. This study was limited in the fact that a plate test designed to screen for all color defects was not included. This limitation was
compensated for by the inclusion of the Pickford-Nicolson Anomaloscope in the color testing protocol.

**Table 2: Color Vision Test Results**

<table>
<thead>
<tr>
<th>SUBJECT</th>
<th>ISHIHARAa</th>
<th>D-15</th>
<th>100-HUE Error Score+</th>
<th>PICKFORD-NICOLSON</th>
</tr>
</thead>
<tbody>
<tr>
<td>MOTHER</td>
<td>P</td>
<td>P</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>FATHER</td>
<td>P (0 errors)</td>
<td>P (one tritan reversal)</td>
<td>F (182 OD, 211 OS)</td>
<td>G/B defect</td>
</tr>
<tr>
<td>SON 1</td>
<td>F (14 errors)</td>
<td>P</td>
<td>P (76 OD, 85 OS)</td>
<td>R/G, G/B, and Y/B defects</td>
</tr>
<tr>
<td>SON 2</td>
<td>F (9 errors)</td>
<td>P (one tritan reversal)</td>
<td>F (118 OD, 70 OS)</td>
<td>G/B defect</td>
</tr>
<tr>
<td>SON 3</td>
<td>F (7 errors)</td>
<td>P (one tritan reversal)</td>
<td>F (178 OD, 193 OS)</td>
<td>R/G, G/B, and Y/B defects</td>
</tr>
</tbody>
</table>

F = fail   P = Pass   X = Not Tested   + = axis not definite   a = 17 plates used

The mother and son 1 performed the D-15 test with no errors. The father, son 2, and son 3 each had a major crossover parallel to the tritan axis line and varying numbers of minor crossovers. The D-15 scoring instructions state that the test is failed if a subject makes two or more major crossovers that are parallel to an axis line on the scoring sheet (Pokorny et al. 1981). By this criteria, all passed the test; however, the father, son 2, and son 3 repeated the major crossover parallel to the tritan axis on subsequent trials of the D-15.

No definite color confusion axis on plots of the 100-Hue test error scores was found for any of the subjects. The father, son 2, and son 3 failed the test. However, son 1 passed the test and, therefore, the anomaloscope was needed for detection of the tritan defect.
Discussion
All subjects display variation in the phenotypic expression of their color vision defects. Speculation on a color defect inheritance pattern is based solely on the test results of the family. Unfortunately, there is no written record on the color vision status of the father and mother's deceased parents and anamnesis of the family was inconclusive.

All tritan sensitive color vision tests classified the father as a tritan defective. The three sons undoubtedly inherited the autosomal dominant tritan defect from the father. The term tritan defect was used as the diagnosis for this project. Other terms, such as tritanopia and tritanomaly are commonly found in the literature. However, Went and Pronk (1985) state that such a distinction, which is justified for the X-chromosome inherited red/green defects, is incorrect for the autosomal dominant blue/green defects; the designation of tritan defect should therefore be preferred.

The color normal mother is a carrier of an X-chromosome linked recessive red/green defect. In addition to a tritan defect, all sons inherited varying amounts of a deutan defect from the mother.

The Ishihara plate test screened son 1 as a strong deuteranope, whereas son 2 and son 3 were described as mild deuteranopes; a description supported by the anomaloscope for only son 1 and son 3. Son 2 narrowly passed the red/green portion of the anomaloscope. None of the congenital tritan defective subjects studied by Smith et al. (1973) failed a single red/green color plate and no other congenital tritans have performed any differently from color normal subjects on such plate tests (Kalmus 1955; Sperling 1960; Walls 1964; Schmidt 1970). Therefore, based on the Ishihara test results and other published research, son 2 can be described as deutan defective regardless of the anomaloscope results.

The finding of all three sons with a combination of an autosomal dominant tritan defect and varying amounts of an X-chromosome inherited deutan defect is rare. In a random population, the frequency of double defective male individuals might be approximately 1.5 per 10,000 (Went and Pronk 1985). However, since this family has a tritan gene, the chance of finding a double defective male is much higher; it should be comparable to the chance of finding a red/green defective male in the general population (Went and Pronk 1985).
tests and would not have been detected as tritan defective without anomalouscope testing. Went and Pronk (1985) hypothesized that the arrangement tests (D-15 and 100-Hue) are unreliable for the diagnosis of a tritan defect and the variability of color vision test results among family members with tritan defects has been previously reported by Pokorný et al. (1981). Moreover, the D-15 failed to detect a deutan defect in any of the three sons.

The Ishihara plate test successfully detected all deutan defects. However, son 1 would not have been diagnosed as having a tritan defect using standard clinical color vision tests without the inclusion of a tritan sensitive color plate test.

The fact that tritan defects in this family were diagnosed was primarily due to the fact that an anomaloscope was available. It is possible that tritan defects could be clinically diagnosed more frequently if clinicians used color vision tests designed to detect all color vision defects.
References


Walls, G. L. Notes of four tritanopes, Vision Res. 1964; 4: 3.
