# ANOMALOUS TRICHROMATISM

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October 13, 2025



RECOMMENDED CITATION

mohammad looti (2025). *ANOMALOUS TRICHROMATISM*. PSYCHOLOGICAL SCALES. Retrieved from https://scales.arabpsychology.com/?p=44319

# ANOMALOUS TRICHROMATISM

**Primary Disciplinary Field(s):** Ophthalmology, Sensory Neuroscience, Genetics

## 1. Core Definition and Nomenclature

**Anomalous trichromatism** represents the most common form of inherited color vision deficiency, characterized by the presence of all three standard types of cone cells in the retina, but with a critical defect in the spectral sensitivity of one of those photopigments. Unlike dichromacy, where an entire class of cones is absent, individuals with anomalous trichromatism retain the ability to perceive three primary colors (trichromacy), but their perception is faulty or skewed because the absorption spectrum of one photopigment is shifted closer to another. This shift creates abnormal overlaps in the visual processing pathways, leading to difficulties in distinguishing specific hues, most frequently within the red-green spectrum.

This condition is distinguished from normal trichromacy by the requirement for an atypical mixture of primary colors to match a given test color, as demonstrated in psychophysical testing. The sensory experience is often described as a reduction in the richness or saturation of specific colors, rather than outright blindness to them. This distinguishes it profoundly from monochromacy or achromatopsia, which involve complete failure of color discrimination.

The condition is frequently referred to by the synonymous terms **anomalopia** or **anomalous trichromasy** in clinical and academic literature. The specific type of anomaly dictates which colors are confused and the overall severity of the impairment, although the overarching principle remains a functional misalignment rather than a structural absence of retinal photoreceptors.



## 2. Biological Basis: The Spectral Shift

Normal human trichromacy relies on three types of retinal cone photoreceptors, often denoted S, M, and L, corresponding to peak sensitivities in the short (blue), medium (green), and long (red) wavelengths, respectively. In anomalous trichromatism, the fundamental issue lies in the molecular structure of the opsin protein within one set of cones. This structural alteration, typically due to a genetic mutation, causes the cone to absorb light maximally at a wavelength slightly different from the norm.

For instance, if the L-cone (red-sensitive) photopigment is shifted closer to the M-cone (green-sensitive) spectrum, the difference between the signals generated by red and green light becomes minimal. Because the brain interprets color based on the *ratio* of stimulation received from the L and M cones, this increased spectral overlap makes the differentiation of red and green hues extremely challenging. The closer the two spectral sensitivity curves are, the more severe the anomaly and the more functionally impaired the individual becomes.

The high prevalence of red-green anomalies is attributable to the genetic clustering of the L and M opsin genes on the X chromosome. These genes are highly homologous, making them susceptible to errors like unequal crossing over during meiosis. This genetic mechanism often results in the creation of hybrid genes that encode photopigments with shifted spectral peaks, thereby establishing the basis for anomalous trichromatism.

## 3. Classification of Anomalous Trichromatism

Anomalous trichromatism is systematically categorized based on which of the three cone types possesses the functionally altered photopigment. The three main classifications are protanomaly, deuteranomaly, and, much less commonly, tritanomaly.

### 3.1. Protanomaly

**Protanomaly** is characterized by a defect in the L-cone (long-wavelength/red) mechanism. Specifically, the L-cone photopigment is shifted toward shorter wavelengths, closer to the M-cone peak. Because the L-cones are also responsible for perceived brightness in the red spectrum, individuals with protanomaly experience a noticeable darkening of red hues, a phenomenon known as protanopia. This condition requires a disproportionately high amount of red light to be mixed with green light to match a standard yellow, indicating a reduced sensitivity to the long-wavelength end of the spectrum.

### 3.2. Deuteranomaly

**Deuteranomaly** involves a defect in the M-cone (medium-wavelength/green) mechanism. The M-cone photopigment is shifted toward longer wavelengths, closer to the L-cone peak. Deuteranomaly is generally the mildest and most common form of color vision deficiency. Individuals with this condition typically do not experience the darkening of red hues seen in protanomaly, but they still struggle significantly with distinguishing reds and greens due to the high degree of spectral overlap between the two cone types.

### 3.3. Tritanomaly

**Tritanomaly** is a much rarer form involving the S-cone (short-wavelength/blue) mechanism. This type involves confusion between blue and yellow hues. Unlike protanomaly and deuteranomaly, which are X-linked, tritanomaly is typically inherited in an autosomal dominant pattern. Because blue-yellow vision defects are less common and often less functionally limiting than red-green defects, tritanomaly receives less clinical attention.

## 4. Clinical Manifestations and Subjective Experience

The primary clinical manifestation of red-green anomalous trichromatism is difficulty in differentiating hues that fall along the red-green axis. This impairment is not absolute; most everyday tasks involving gross color discrimination remain manageable, but subtle differences in saturation or shade can lead to errors. The severity of the manifestation is highly dependent on the degree of spectral shift in the anomalous photopigment; mild cases may go undiagnosed, while severe cases closely mimic dichromacy.

A specific characteristic noted in the literature, particularly in severe cases of protanomaly and deuteranomaly, is the relationship between color intensity and discriminability. Individuals may find it easier to distinguish between duller or desaturated shades of red and green, but the difficulty intensifies when presented with brilliant or highly saturated colors. This paradox occurs because highly intense, bright colors flood both the overlapping L and M cones simultaneously, exacerbating the ambiguity of the resulting signal ratio. Conversely, duller shades utilize a lower saturation input, sometimes providing just enough differential signal for the brain to process a distinction.

Furthermore, color constancy--the ability to perceive colors reliably under different light sources--is often impaired. Since the anomalous cone system relies on highly specific spectral input, changes in ambient lighting conditions (such as moving from natural sunlight to fluorescent light) can drastically alter the perceived color balance, increasing the likelihood of misidentification of critical signals, such as indicator lights or warning signs.

## 5. Etiology and Molecular Genetics

The genetics underlying protanomaly and deuteranomaly are rooted in the X chromosome, explaining why approximately 8% of Caucasian males are affected, compared to only 0.5% of females. The genes for the M and L opsins (OPN1LW and OPN1MW) reside adjacently on the X chromosome (Xq28). This genetic arrangement is highly unstable due to the sequence similarity between the two genes.

The creation of anomalous photopigments typically arises from two molecular mechanisms. First, unequal homologous recombination during meiosis can lead to the formation of hybrid fusion genes, where parts of the L-opsin gene are spliced with parts of the M-opsin gene, resulting in a novel photopigment with a shifted spectral peak. Second, point mutations within the existing L or M opsin genes can cause minor amino acid substitutions that subtly alter the physical structure of the photopigment, thereby changing its maximum absorption wavelength.

The inheritance pattern is X-linked recessive. Males only possess one X chromosome; thus, a single anomalous gene is sufficient to express the condition. Females, having two X chromosomes, are usually carriers unless they inherit the defective gene on both chromosomes or experience adverse X-inactivation (lyonization), making full expression of the condition in females rare.

## 6. Diagnosis and Psychophysical Testing

Diagnosis of anomalous trichromatism requires sophisticated psychophysical testing, as simple screening tools often fail to differentiate between a severe anomaly and true dichromacy.

**Screening Tests:** The most common initial screening involves pseudoisochromatic plates, such as the Ishihara plates. These plates rely on figures embedded in backgrounds composed of dots of different colors and varying brightness. While effective at detecting the presence of a red-green defect, they cannot accurately categorize the type (protan vs. deutan) or the severity of the anomaly.
**Quantitative Diagnosis:** The gold standard for definitive diagnosis and quantification of severity is the **anomaloscope**, historically the Rayleigh equation match. The patient is asked to mix specific ratios of red light and green light until the mixture exactly matches a reference yellow light. Normal trichromats will accept only a narrow, specific range of red/green ratios. Anomalous trichromats, depending on the direction and degree of their spectral shift, will require highly abnormal ratios (either too much red for protanomals or too much green for deuteranomals) to perceive a match. This test provides a precise quantification of the anomaly coefficient.

## 7. Significance and Impact

Anomalous trichromatism has significant implications for daily life and occupational fitness. While often perceived as a minor inconvenience, it can pose serious safety risks in environments where rapid and accurate color discrimination is vital. For example, interpreting traffic signals, distinguishing color-coded wiring, or reading medical diagnostic tests that rely on color changes can become hazardous.

Furthermore, specific careers, particularly those involving transportation (pilot, train driver), electrical engineering, graphic design, and certain military roles, impose strict requirements for normal color vision, leading to occupational exclusion for individuals with moderate to severe anomalous trichromatism. Therefore, the condition is not merely a perceptual difference but a determinant of functional capacity in a visually complex world. Given its high prevalence, understanding and accommodating anomalous trichromacy is essential for public safety and educational policy.

## Further Reading

Color vision deficiency (Wikipedia)

Trichromacy (Wikipedia)
Cone Cell (Wikipedia)
Ishihara Test (Wikipedia)
Rayleigh Equation (Wikipedia)