Mechanism of Color Vision: When light enters the eye through the cornea and passes through the aqueous humour, lens, and vitreous humour, it arrives at the retina. Cone cells contain the plasma membrane of the cone cells. At each photopigment’s peak sensitivity, approximately five photons (of the correct wavelength) are required to isomerize 11-cis retinal and trigger a reaction in the photopsin. Figure two is a representation of how 11-cis retinal is isomerized. (OMIM)

At lower sensitivities, more photons are required to trigger a signal. Different sensitivities are mediated by the apoprotein’s interaction with the retinal. This is determined by the structure of the apoprotein. For a given set of photons of the same wavelength, many electrical signals will be produced by the photopigments sensitive to that color. The more signals sent for a color determines what color is perceived. (Rabin; OMIM)

Deuteranomaly:

In deuteranomaly, a hybrid red-green pigment exists in place of the green pigment, OPN1MW. This pigment is the anomalous form of the normal green pigment. As a result, the medium-wave cone pigment is more similar to red (fig. 3) and thus less sensitive to green. Fewer green signals are sent, and the appearance of green in images is weaker. Other colors between the red and green wavelengths are also distorted because of their longer-wavelength peak sensitivity. (NCBI)

DIAGNOSIS and INHERITANCE PATTERNS:

Diagnosis: The diagnosis of deuteranomaly (and other types of color blindness) is performed through an Ishihara test like the one shown above. There are also other similar Ishihara tests which provide the same diagnosis. Another type of test that has been developed to diagnose deuteranomaly is called “American Optical HRR pseudochromatic plates” (Motulski and Deeb).

Inheritance Patterns: The gene for the green cone pigment OPN1MW is at locus Xq28, at the far end of the long arm of the X chromosome. The mutated allele is a hybrid form of OPN1MW and OPN1LW, the genes for green and red pigments. The inheritance pattern for the two alleles is X-linked recessive. The reason why males are more likely to inherit the mutated allele is because they inherit only one X-chromosome which comes from their mother (who must have or carry the disease to transmit it to their offspring).

Females who have two X-chromosomes become carriers only if they inherit the allele, otherwise they are unaffected. The diagram on the following slide should outline a few case scenarios in which red-green color blindness is inherited. (OMIM; NCBI)

References:
2. “1KPW: Firstglance in Jmol.” Jmol <http://video.google.com/videoplay?docid=2623091881173699730&q=color+blindness+&ei=5ZIrSbdiI8qCqAPJ0PCk&start=30&nstart=100&nump=100>
3. Carbonaceous, Interactive with Bobby J, Wyeth, and Rafael C. Color Blindness, 15 May 2008 <http://videos.google.com/videoplay?docid=2623091881173699730&q=color+blindness+&ei=5ZIrSbdiI8qCqAPJ0PCk&start=30&nstart=100&nump=100>
Pedigree for Deuteranomaly: Red-Green Color Blindness

KEY

- Red Male Affected
- Male Unaffected
- Female Affected
- Female Unaffected
- Female Carrier
Deuteranomaly Video

http://video.google.com/videoplay?docid=2623091881173699730&q=color+blindness+&ei=5ZIrSK62NYeIrgKwm-iNCg&hl=en
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