

Polymorphisms of the L and M opsin genes and their consequences for colour vision

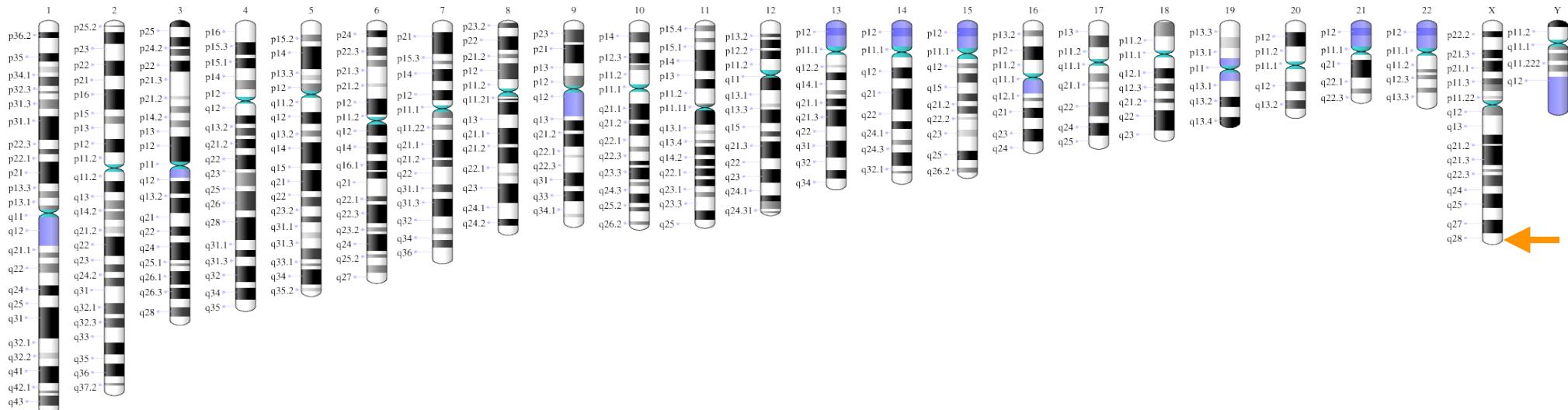
Jenny Boston; j.boston@sussex.ac.uk

Computational Color, 10th November 2020

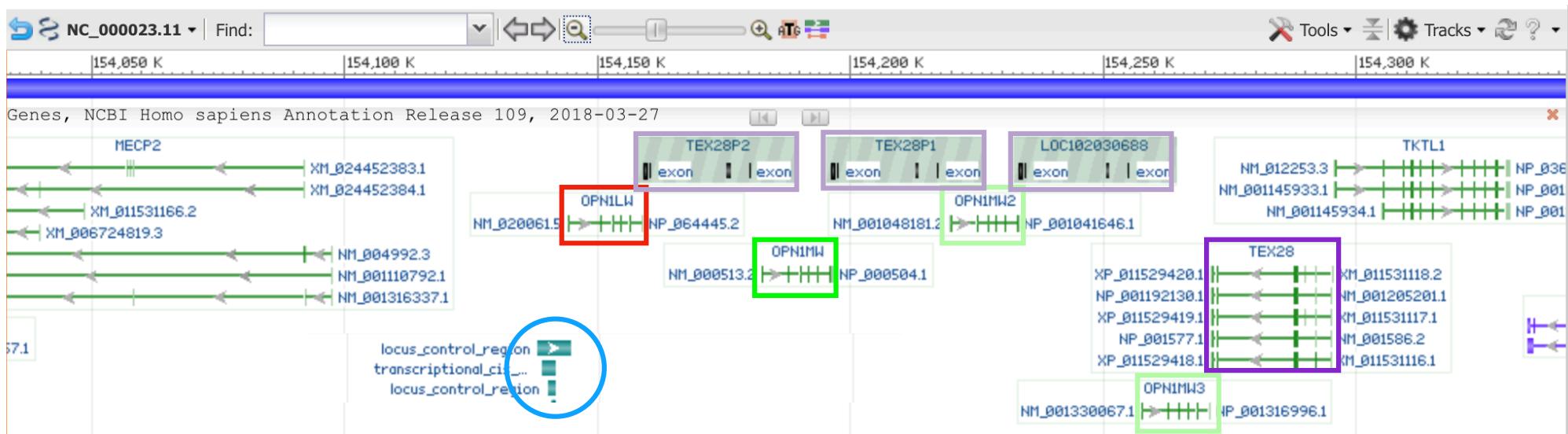
Outline

- Genetic basis of colour vision deficiency
- Modelling the colour vision of individual observers
- Diagnosing colour vision deficiency
- Individual differences in anomalous trichromacy and their impact on colour discrimination
- Postreceptoral compensation in anomalous trichromacy
- Notch filters as an aid to colour vision for anomalous trichromats
- Polymorphisms among normal trichromats
- Human tetrachromacy

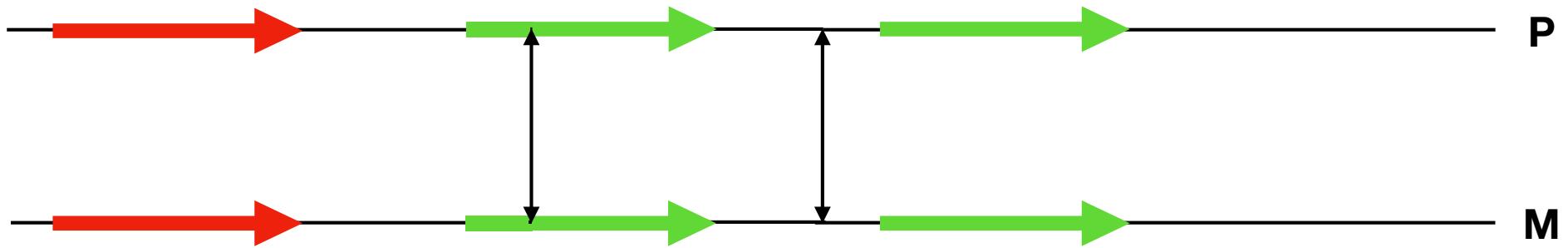
Location of the opsin genes



<https://www.ncbi.nlm.nih.gov/gene/5956>; alternatively <https://www.genome.ucsc.edu>



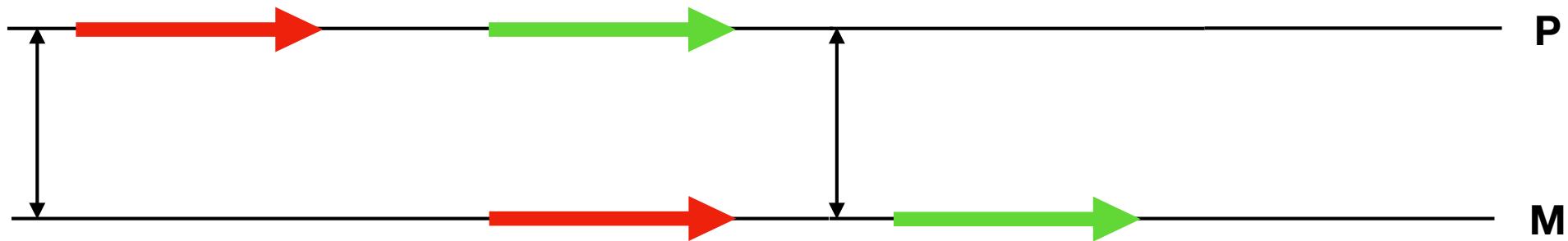
Genetic variation in the opsin genes



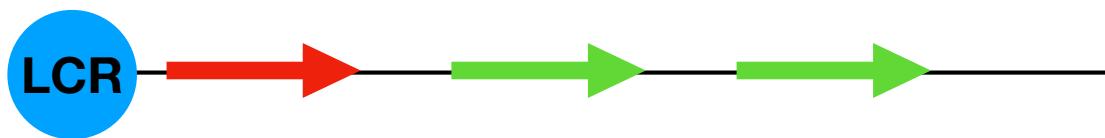
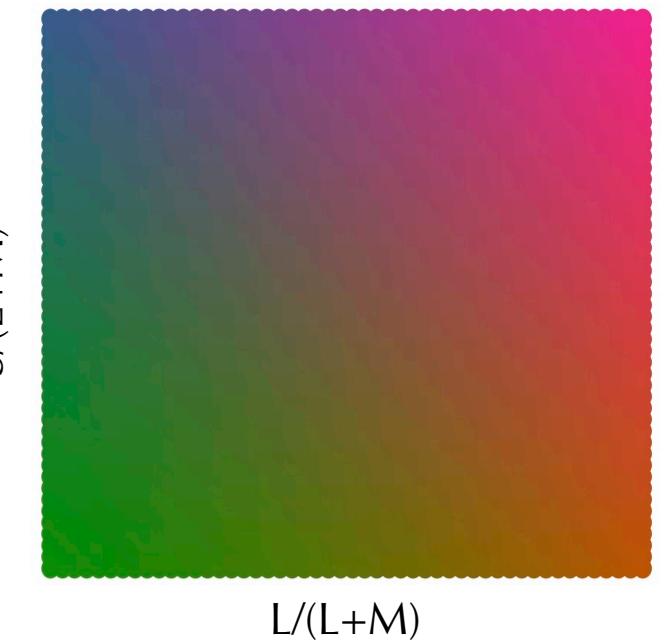
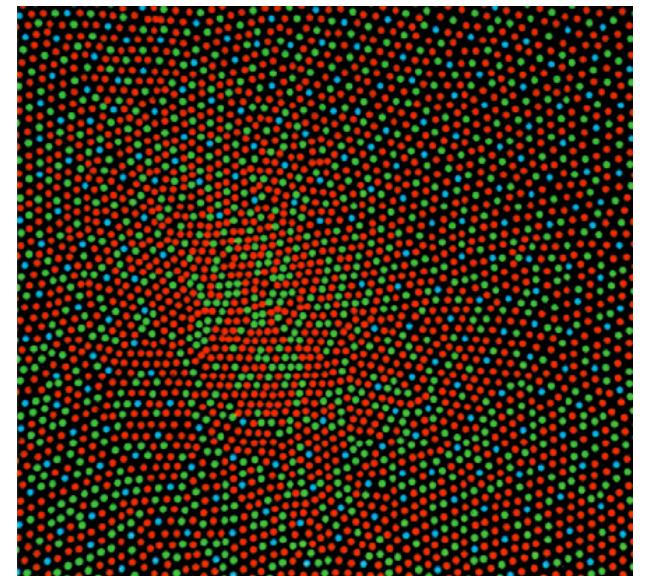
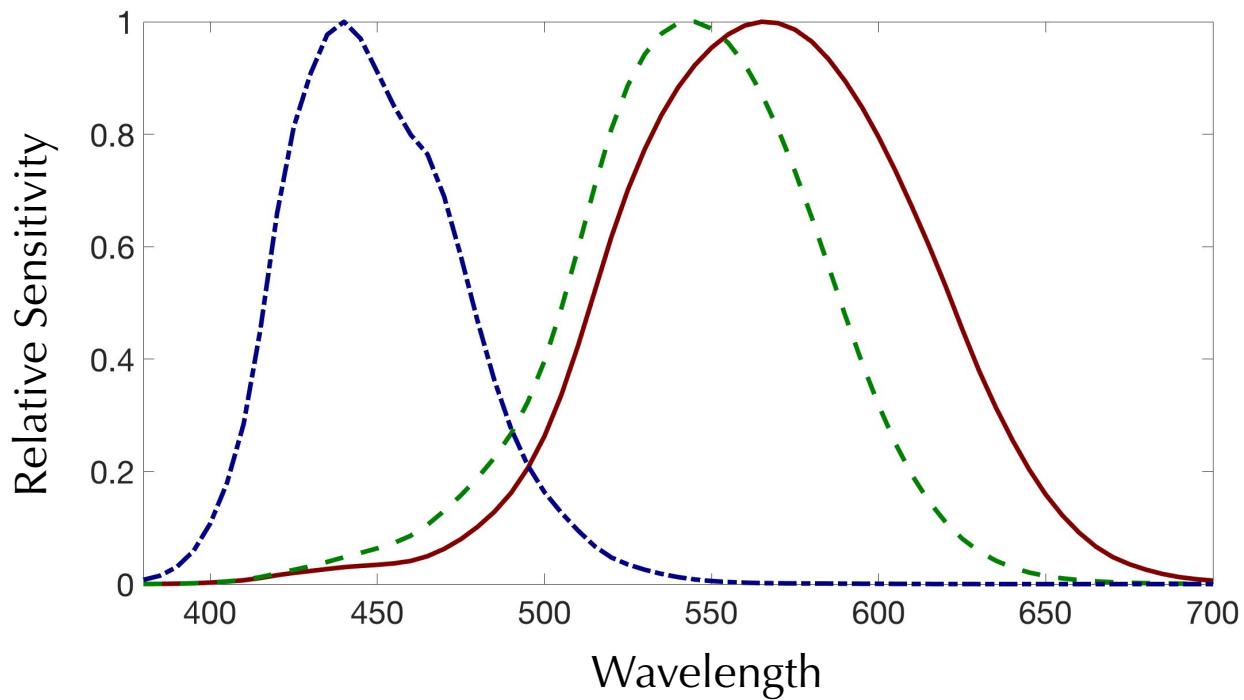
from Hunt and Carvalho (2016)

Genetic variation in the opsin genes

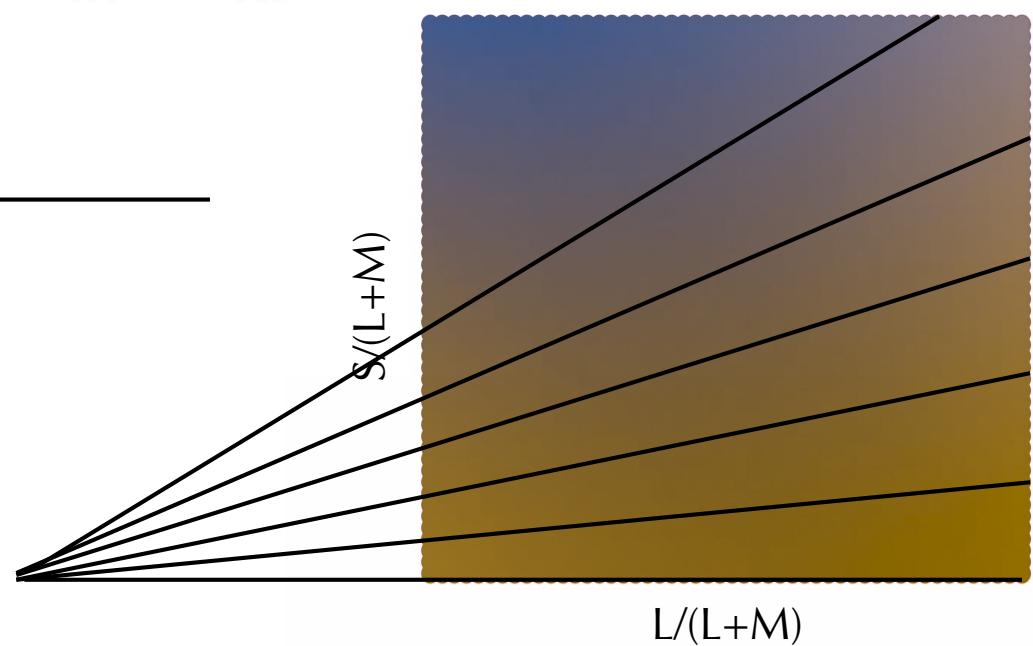
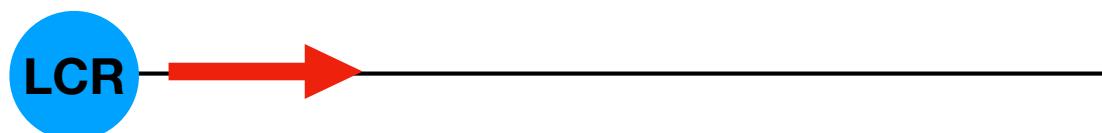
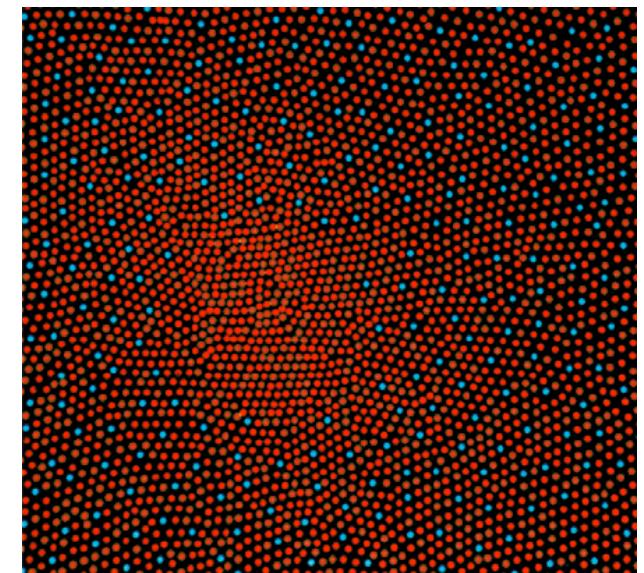
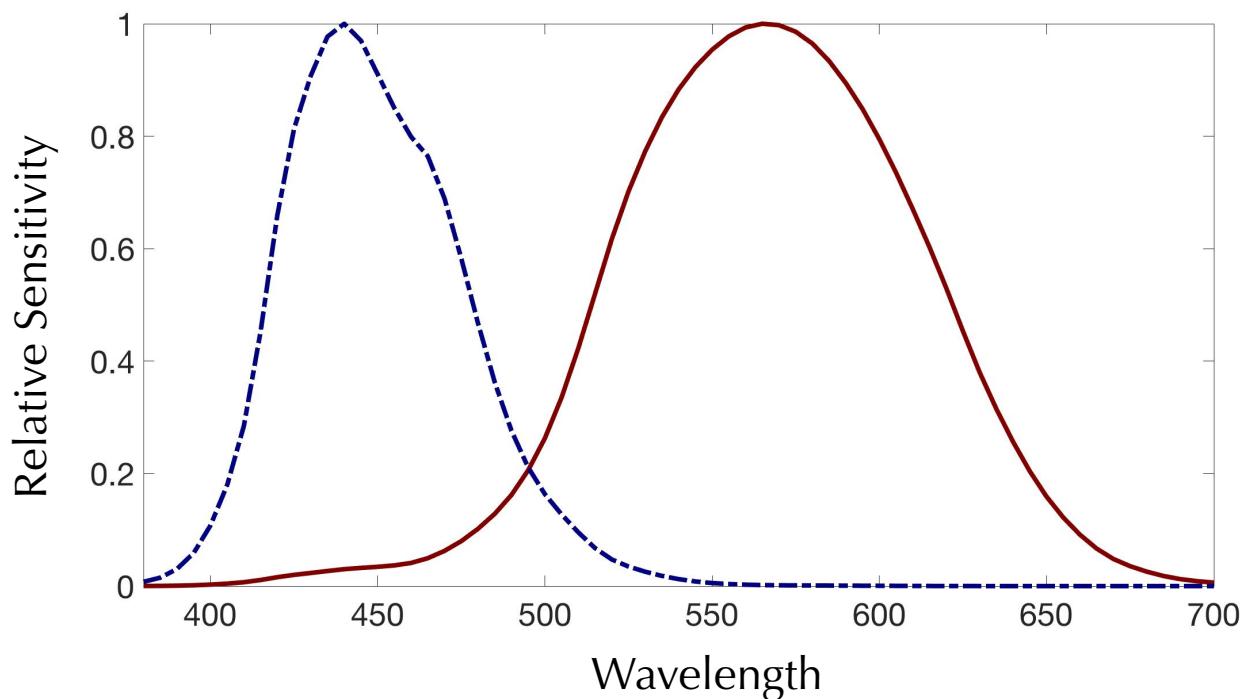
- OPN1LW or OPN1MW may be missing



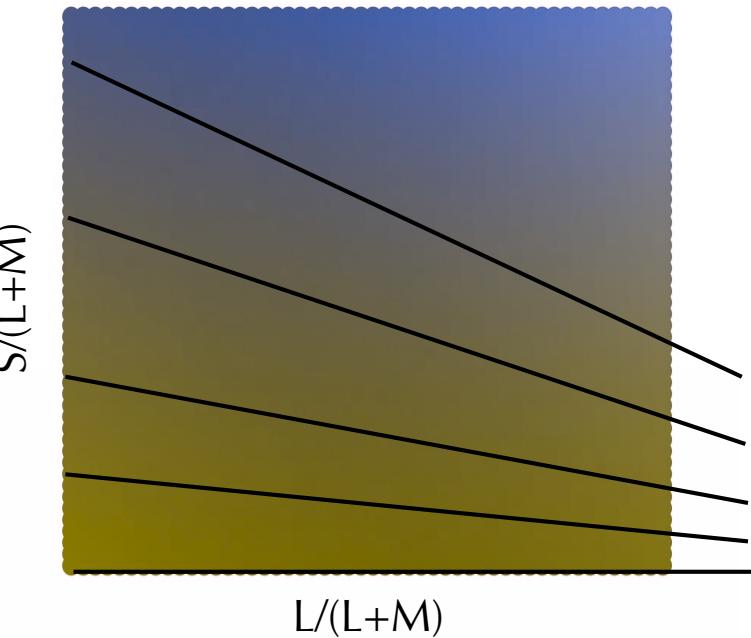
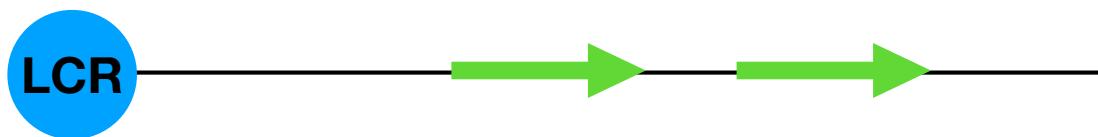
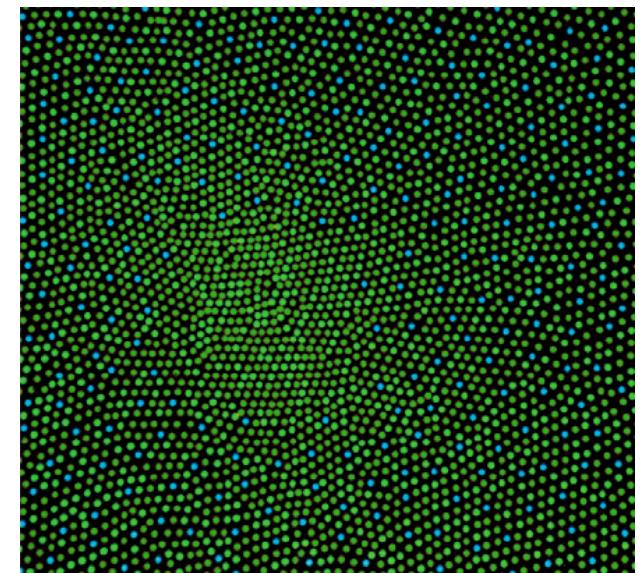
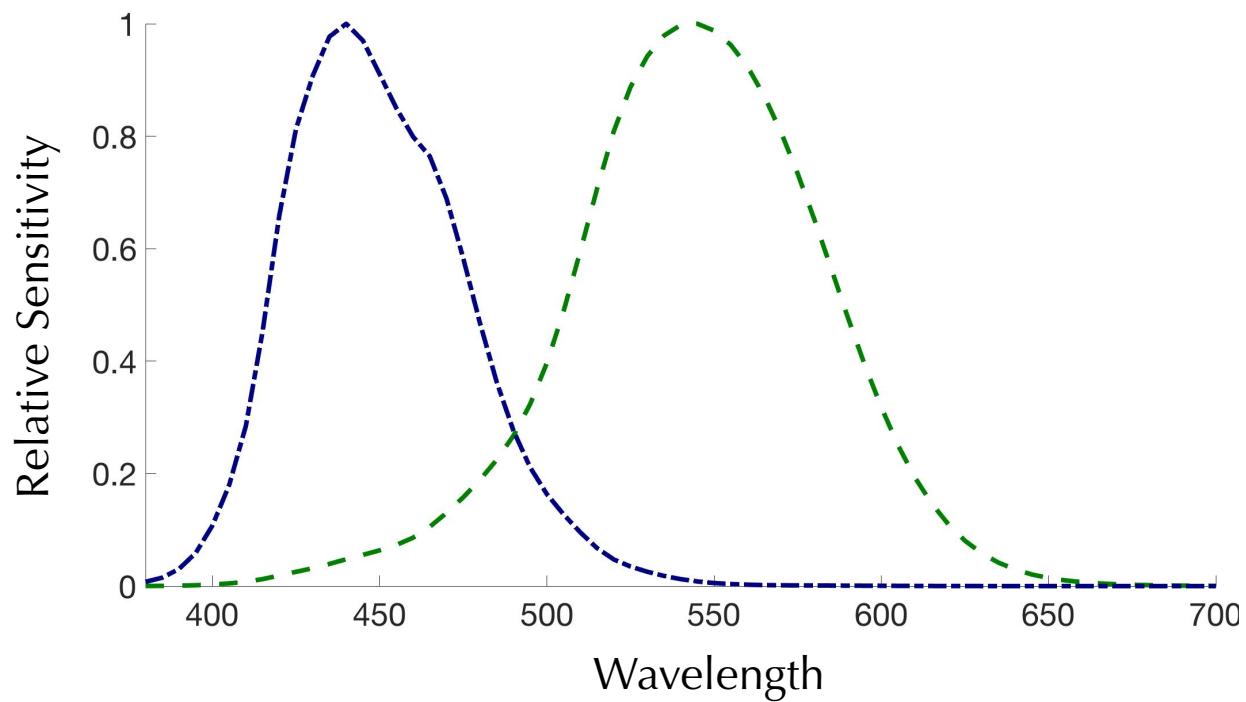
Normal trichromacy



Dichromacy: deutanopia

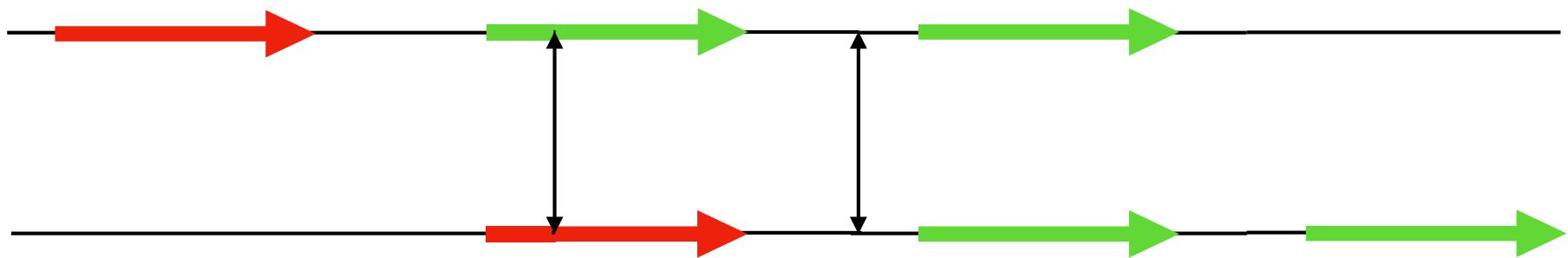


Dichromacy: protanopia



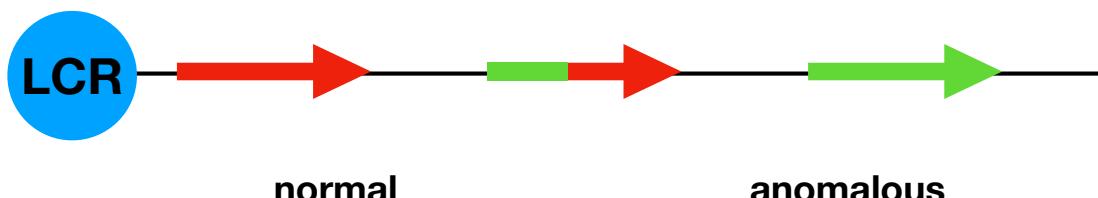
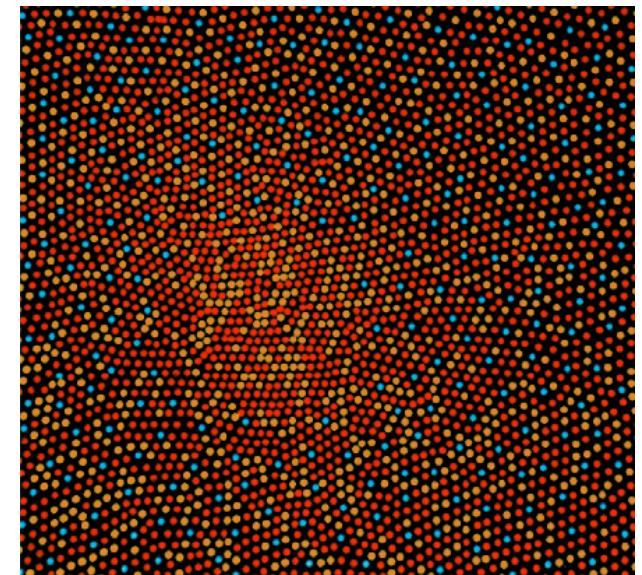
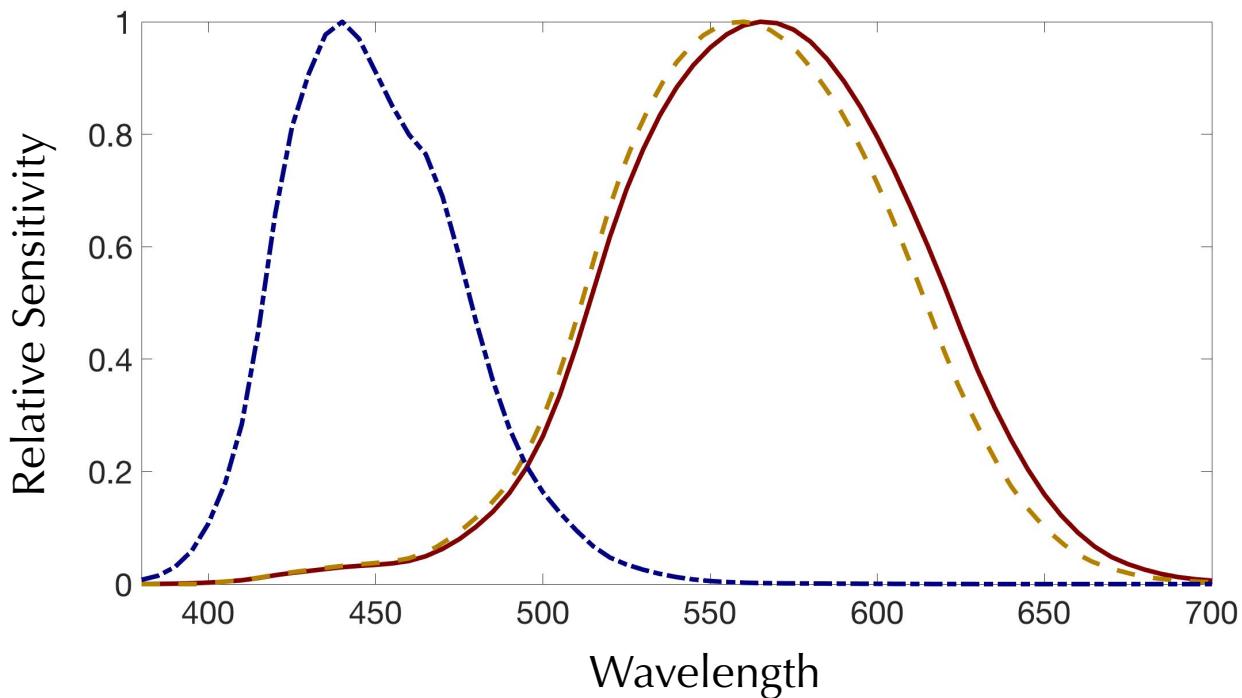
Genetic variation in the opsin genes

- OPN1LW or OPN1MW may be missing
- Parts of OPN1LW and OPN1MW may be combined in 1 (hybrid) gene

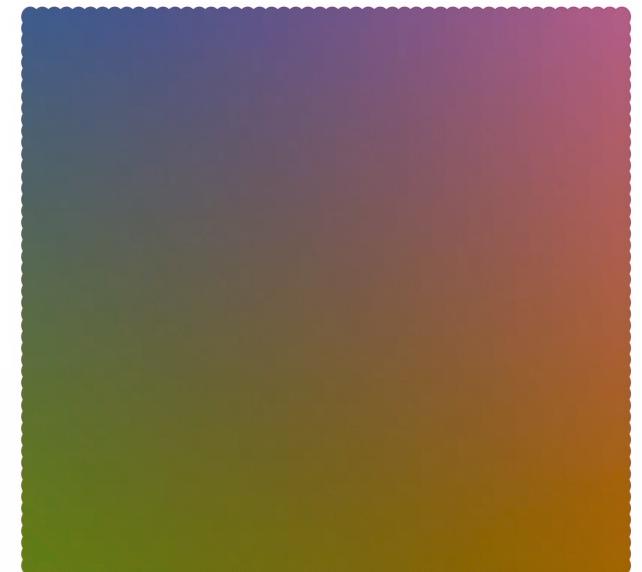


from Hunt and Carvalho (2016)

Anomalous trichromacy: deuteranomaly

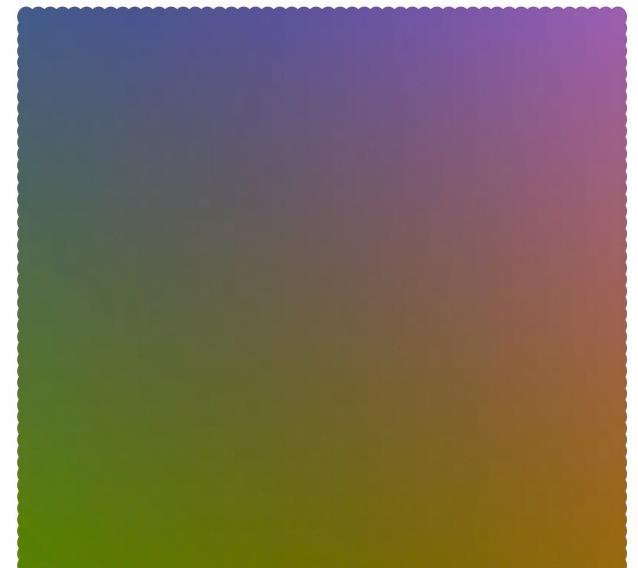
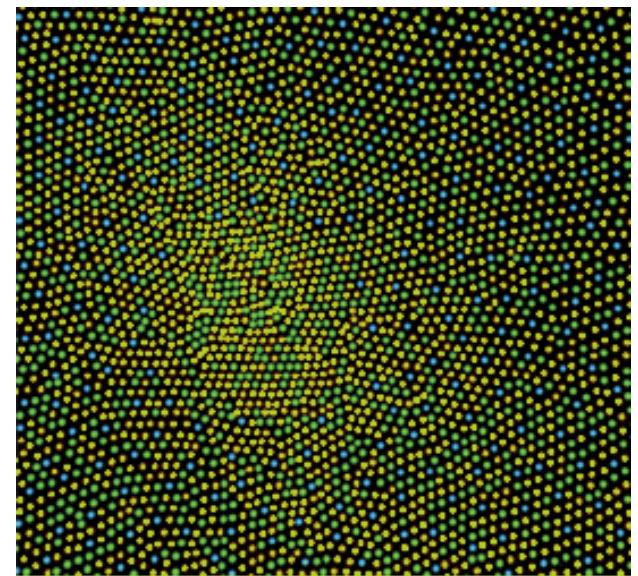
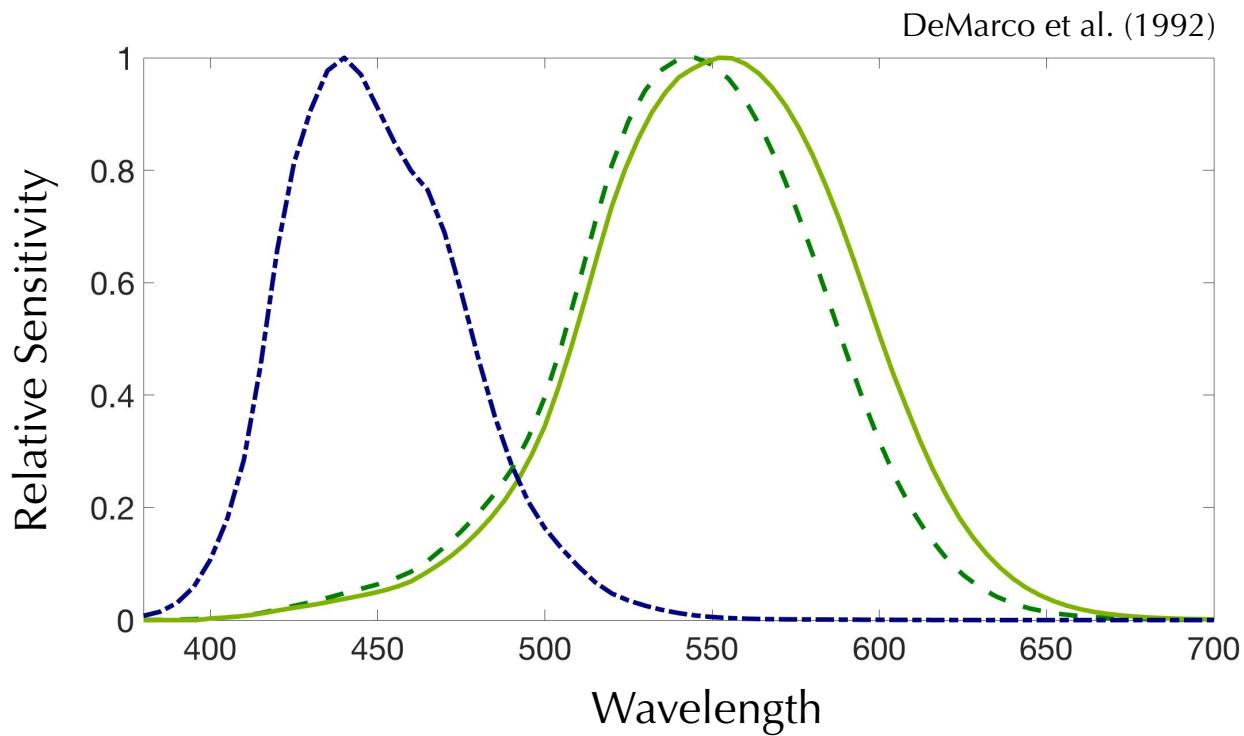


$S/(L+M)$



$L/(L+M)$

Anomalous trichromacy: protanomaly

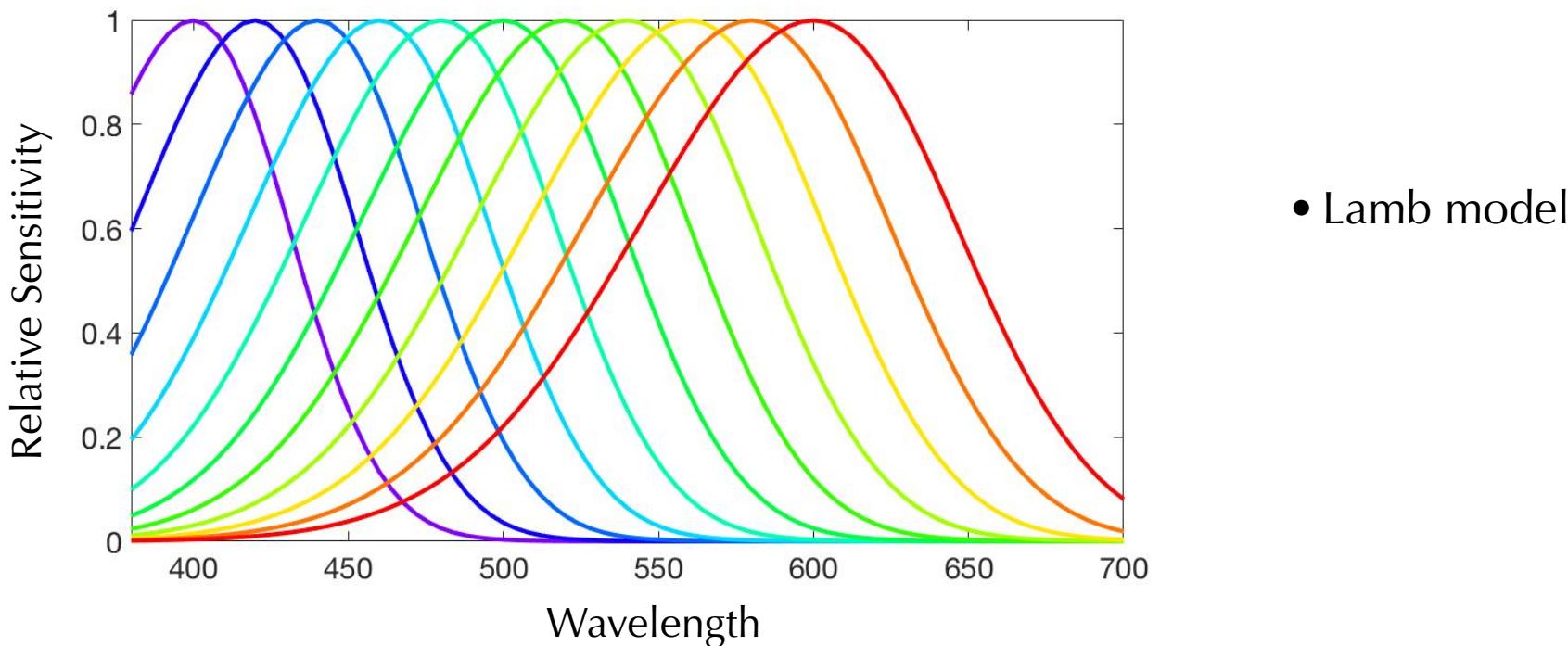


$S/(L+M)$

$L/(L+M)$



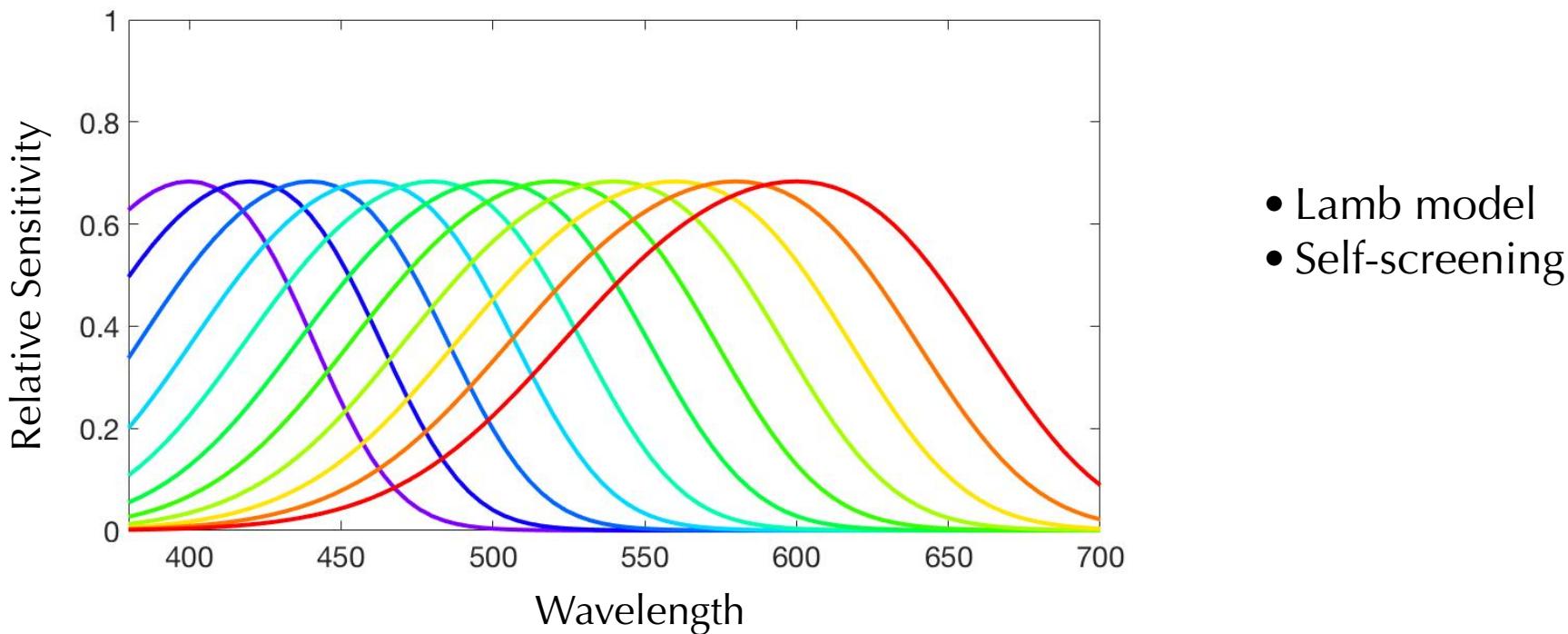
Anomalous cone fundamentals: model



Lamb's (1995) formula:

$$S(\lambda) = \frac{1}{\left\{ \exp a\left(A - \frac{\lambda_{\max}}{\lambda}\right) + \exp b\left(B - \frac{\lambda_{\max}}{\lambda}\right) + \exp c\left(C - \frac{\lambda_{\max}}{\lambda}\right) + D \right\}}$$

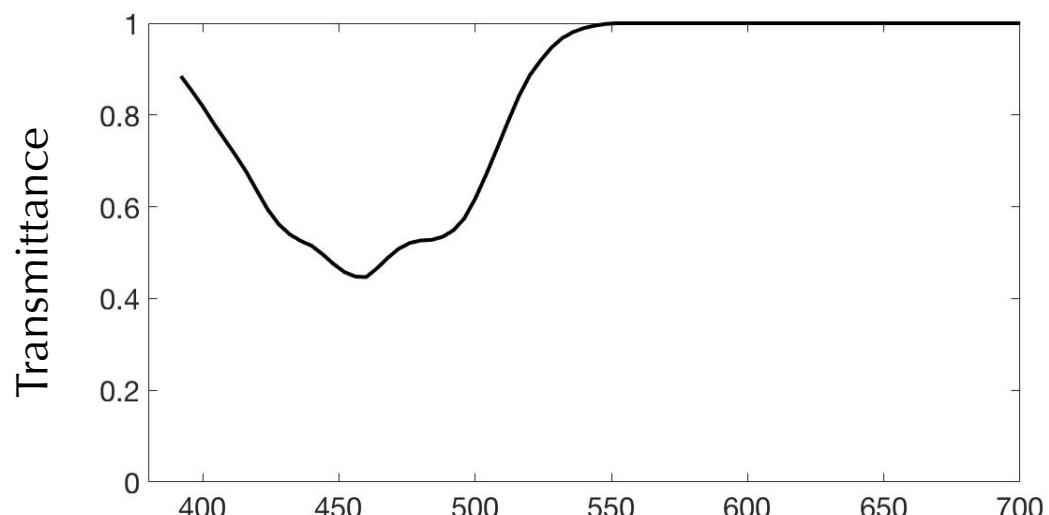
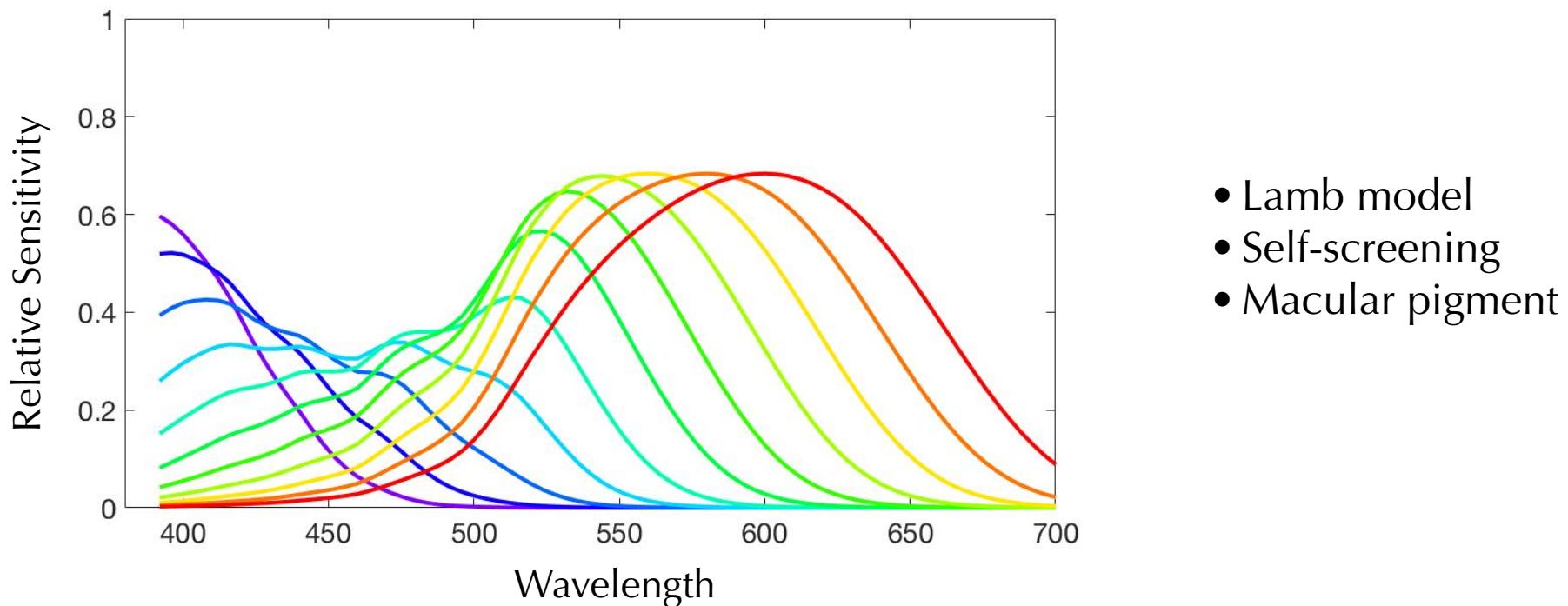
Anomalous cone fundamentals: model



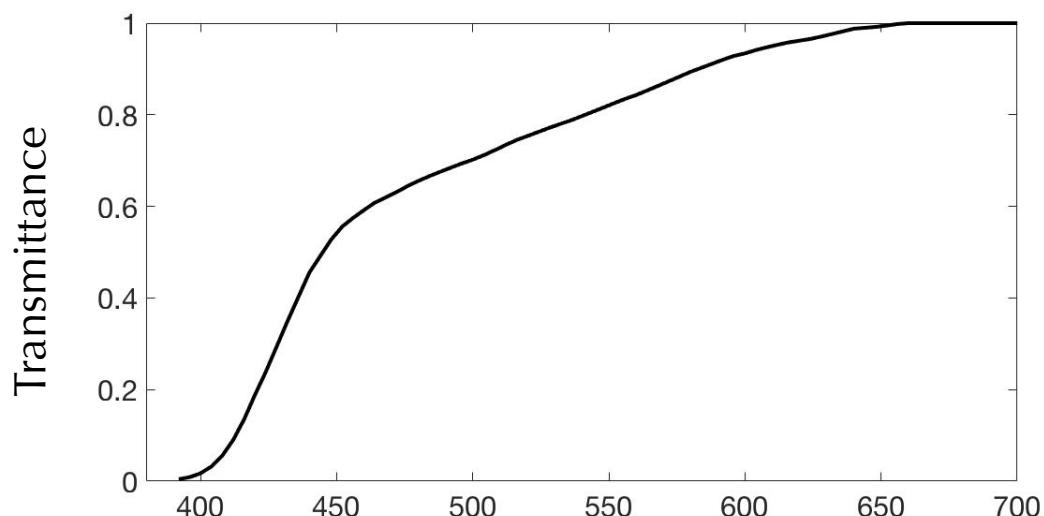
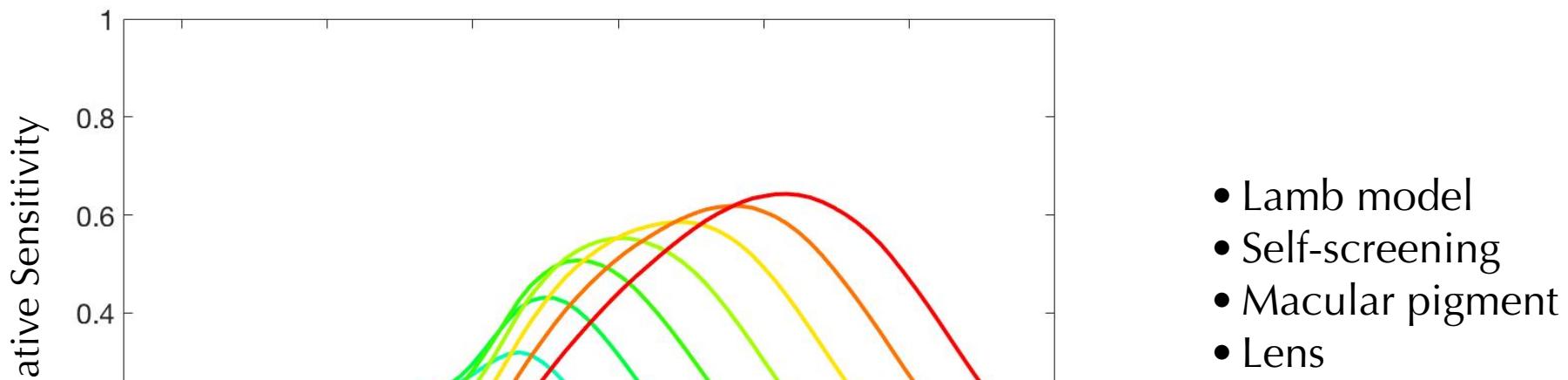
Self-screening:

$$S(\lambda) = 1 - 10(-0.5 * s(\lambda)).$$

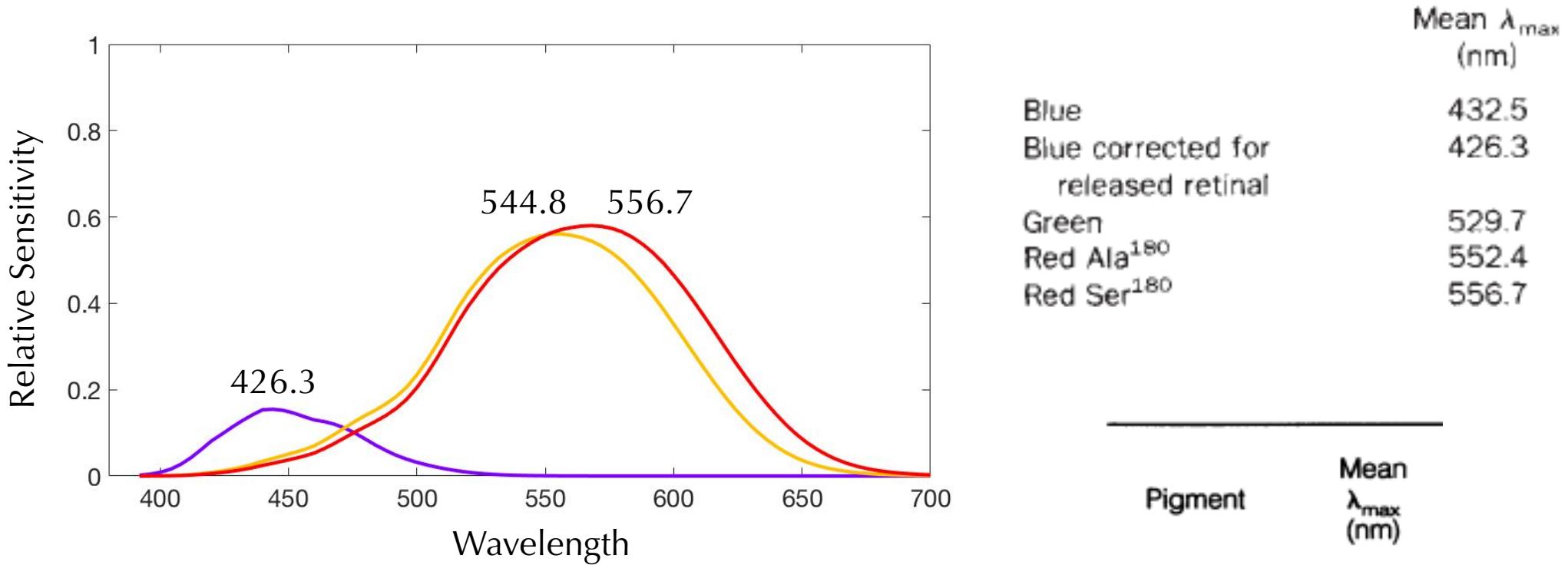
Anomalous cone fundamentals: model



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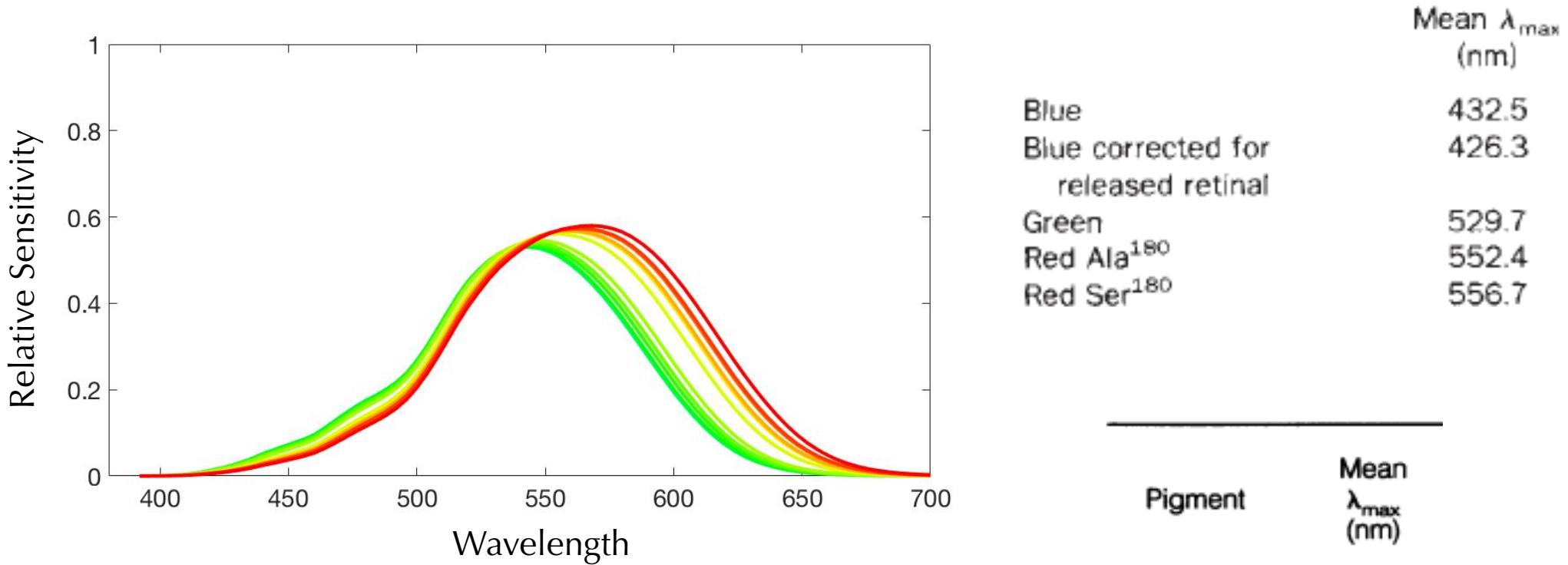
Absorption Spectra of the Hybrid Pigments Responsible for Anomalous Color Vision

Shannath L. Merbs and Jeremy Nathans

Absorption spectra of human cone pigments

Shannath L. Merbs* & Jeremy Nathans*†

Anomalous cone fundamentals: model



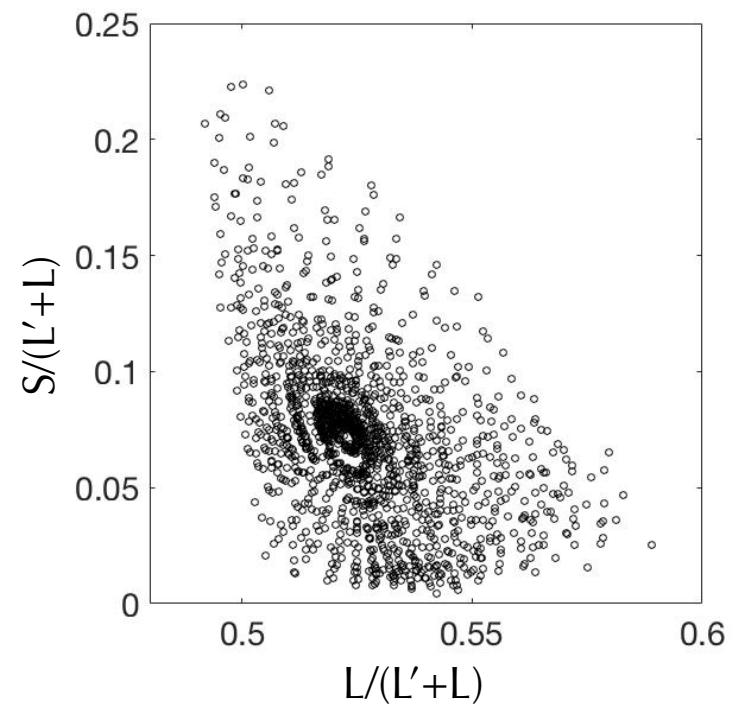
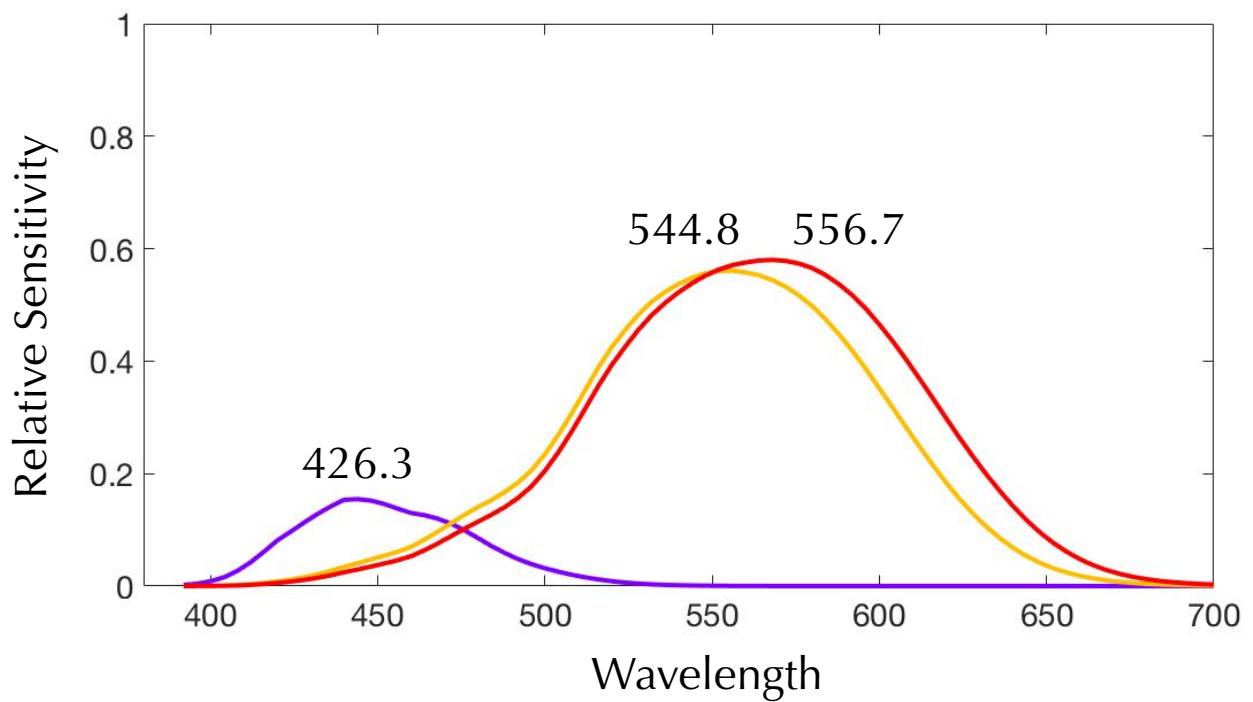
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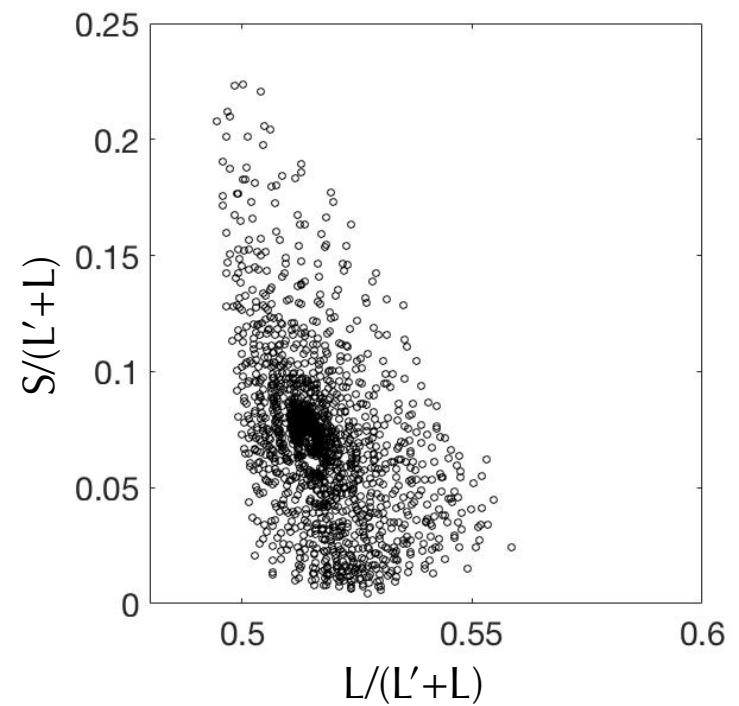
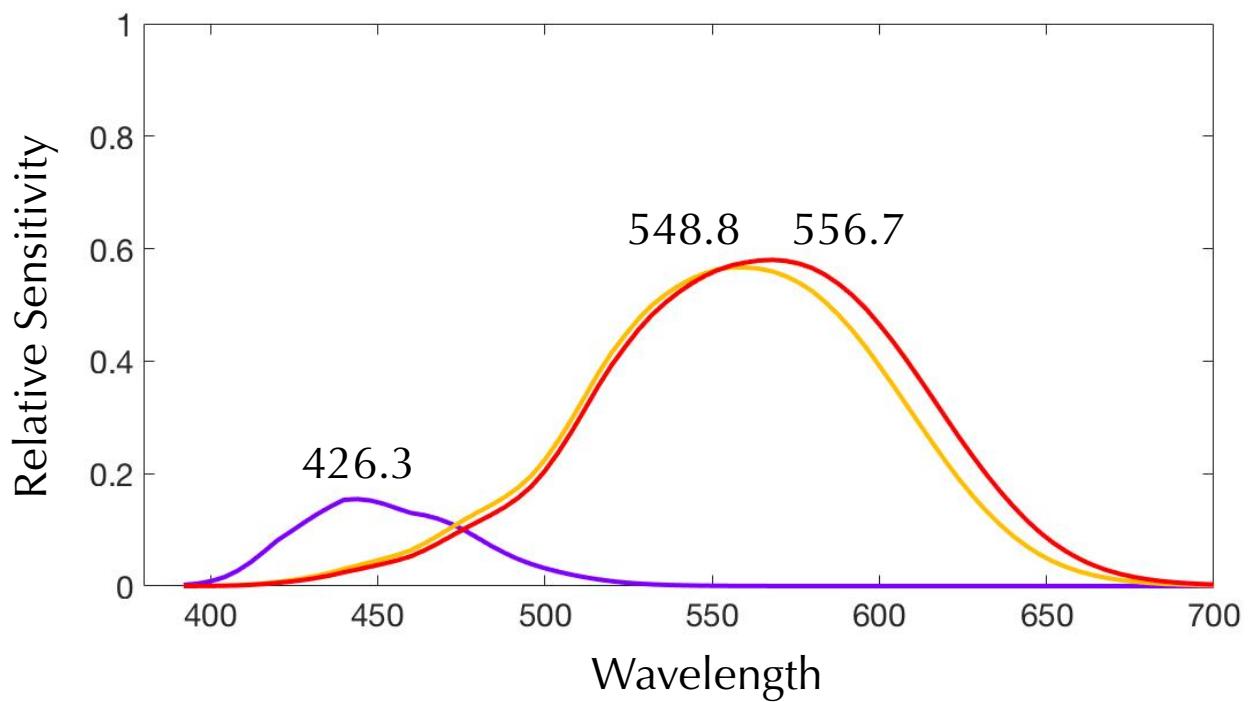
Absorption spectra of human cone pigments

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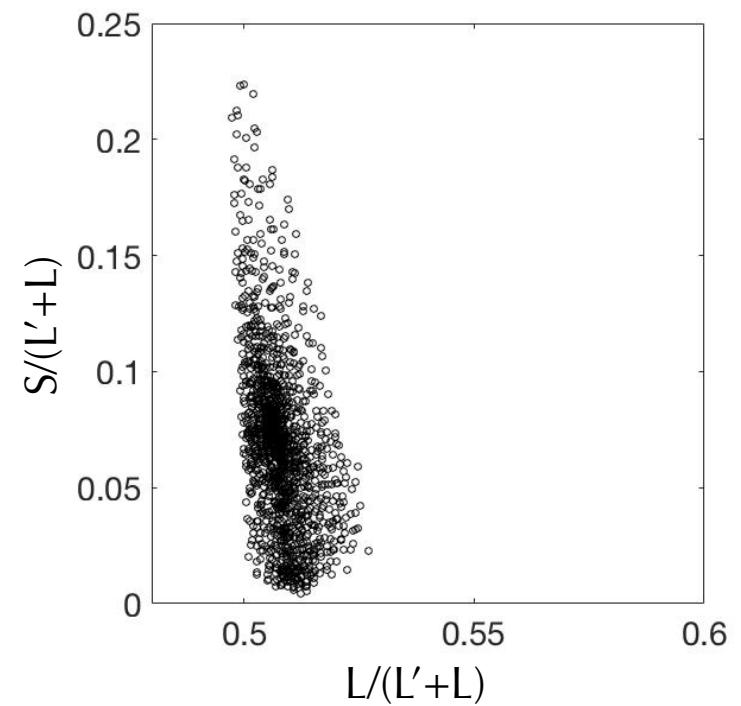
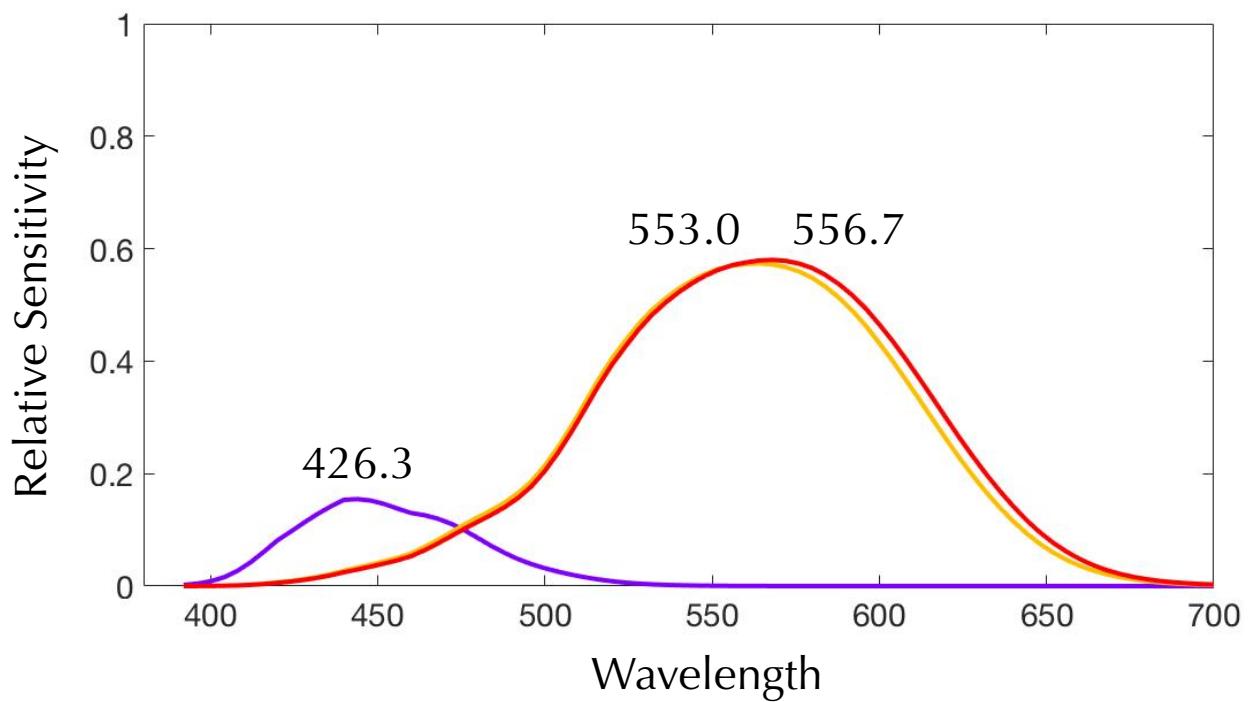
Anomalous cone fundamentals: model



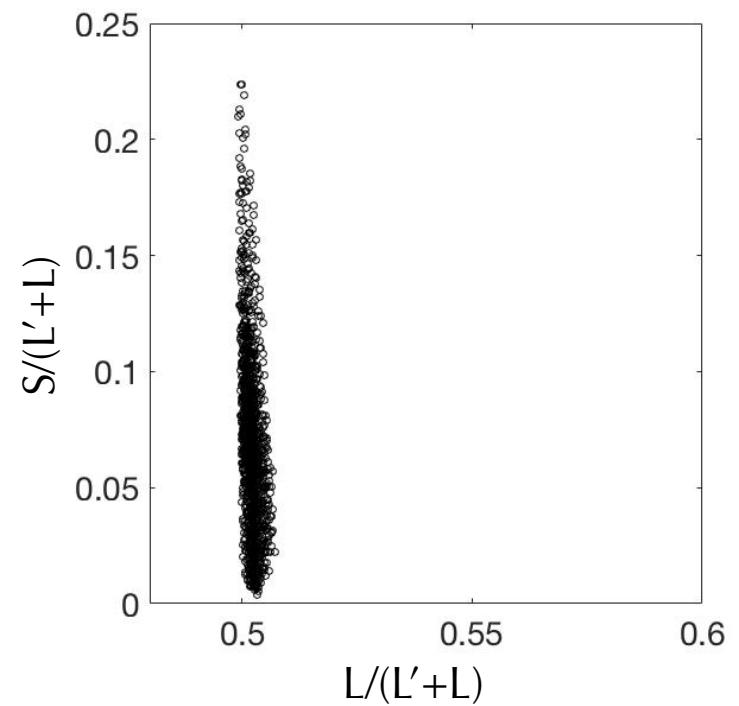
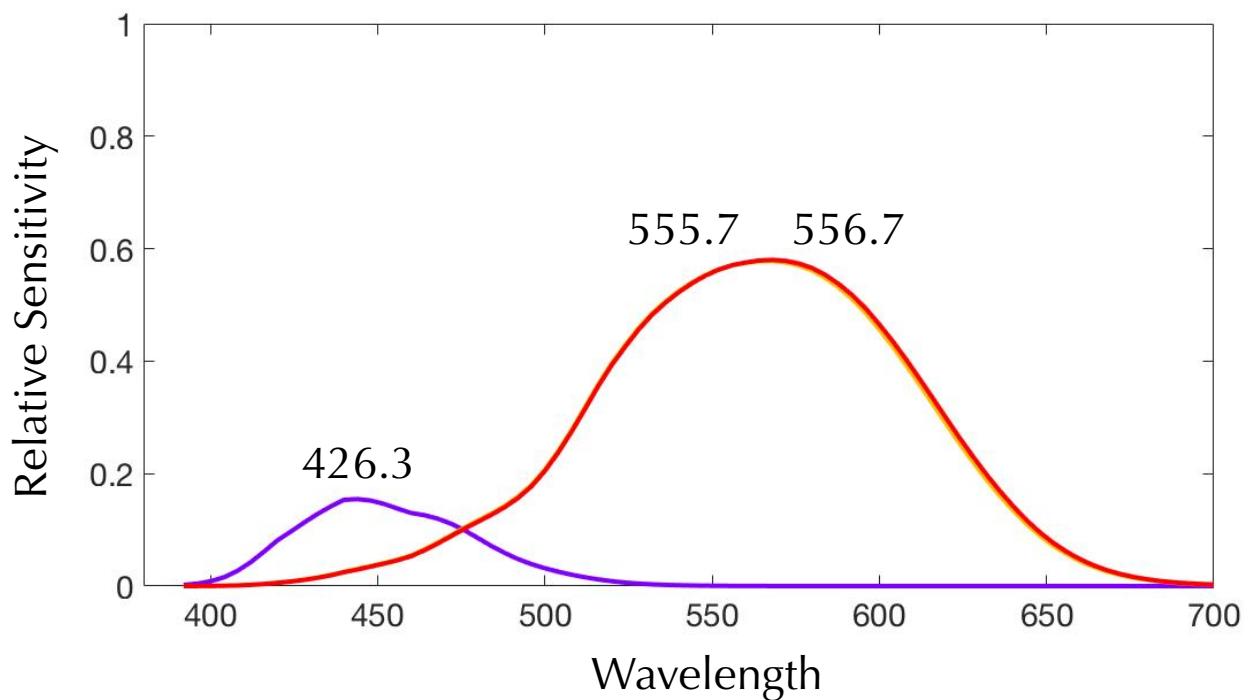
Anomalous cone fundamentals: model



Anomalous cone fundamentals: model

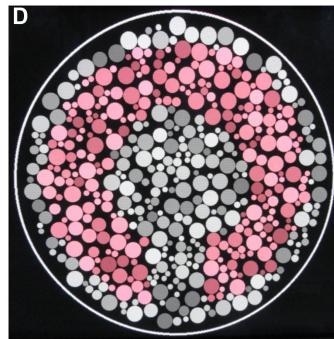


Anomalous cone fundamentals: model



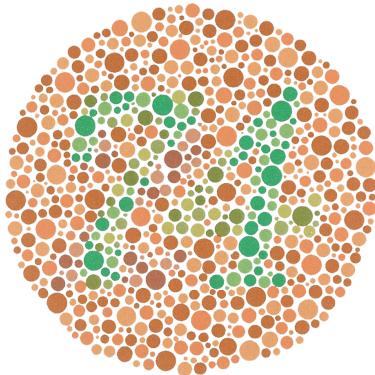
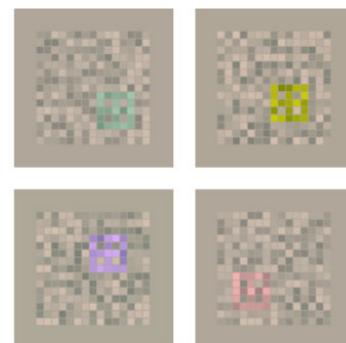
Diagnosing colour vision ‘deficiency’

Detection tests



Cambridge Colour Test
(Regan and Mollon, Sold by CRS)

CAD test
(Barbur et al.)



Ishihara test
(Ishihara, 1917)

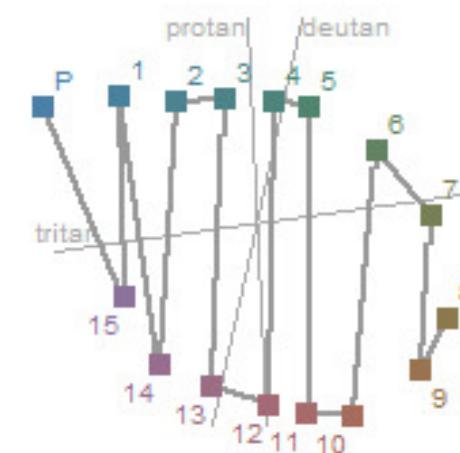
Arrangement tests



Farnsworth Munsell 100 hue test



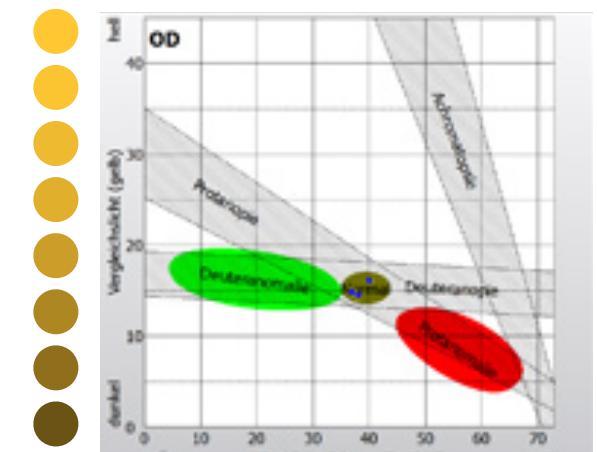
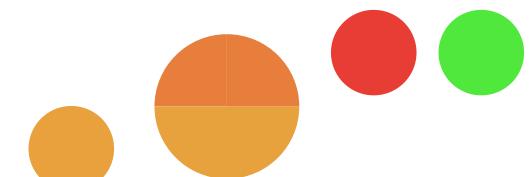
Farnsworth Munsell D15 test



Matching tests

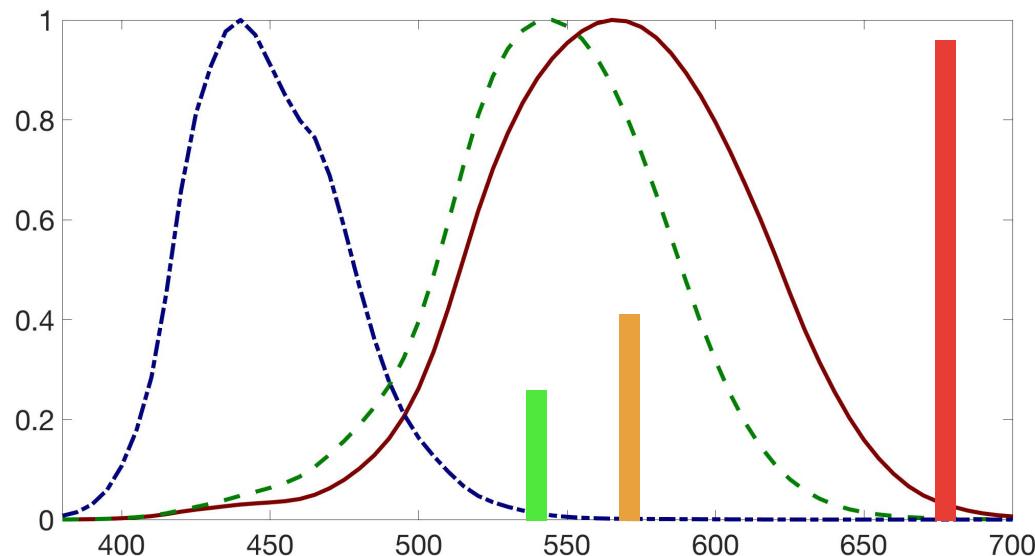


Anomaloscope (Nagel, 1907)



Diagnosing colour vision ‘deficiency’

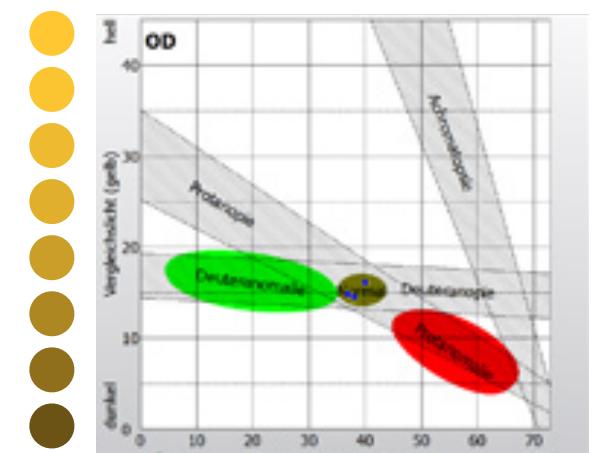
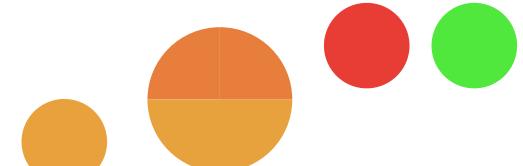
Normal trichromacy



Matching tests

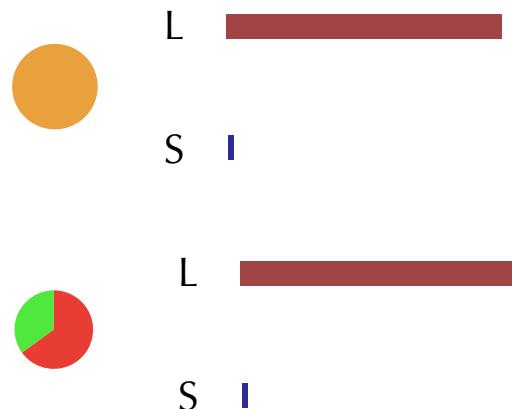
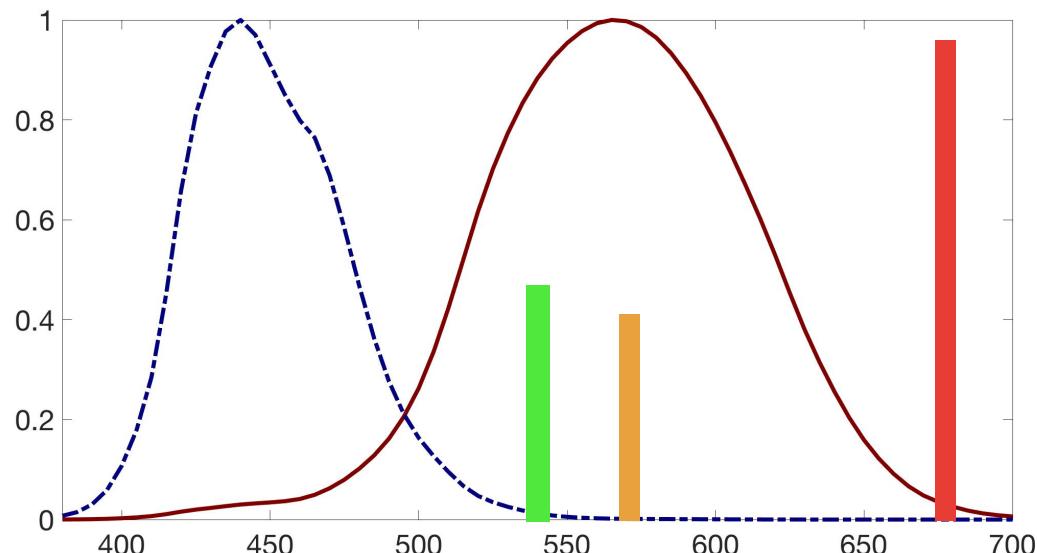


Anomaloscope (Nagel,
1907)



Diagnosing colour vision ‘deficiency’

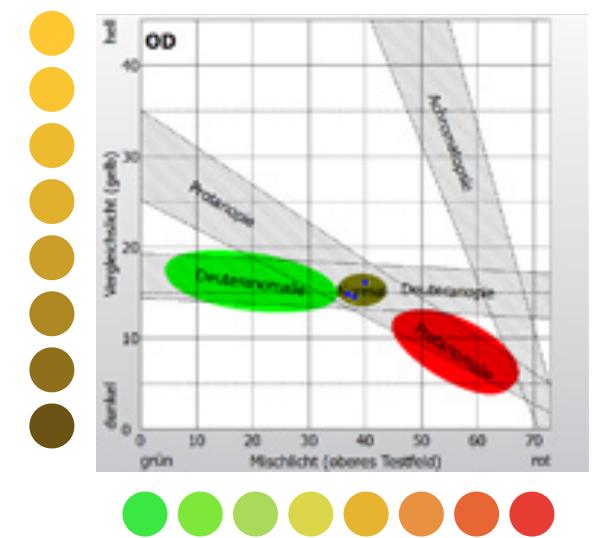
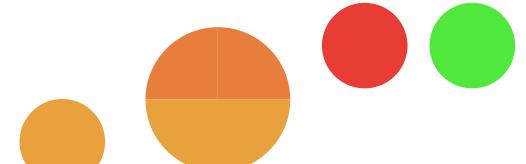
Deuteranopia



Matching tests

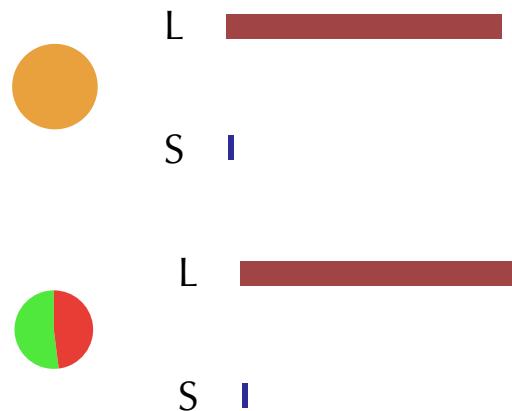
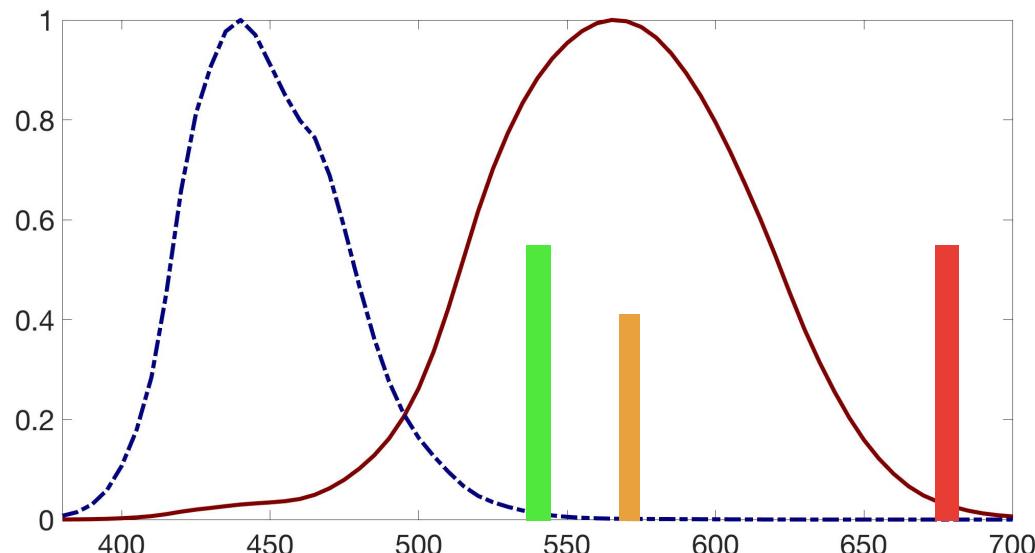


Anomaloscope (Nagel, 1907)



Diagnosing colour vision ‘deficiency’

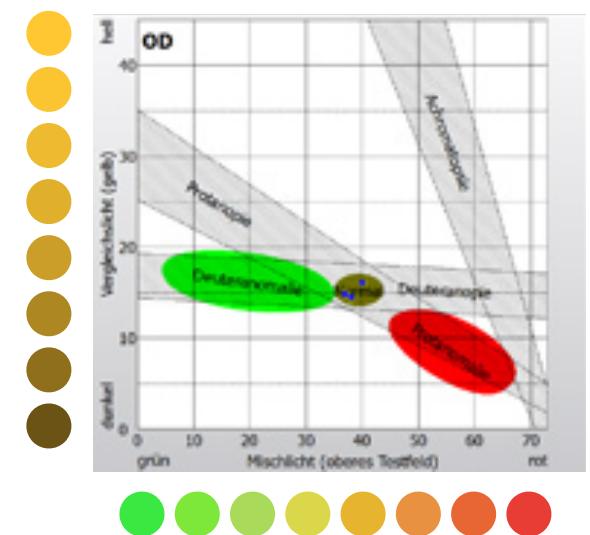
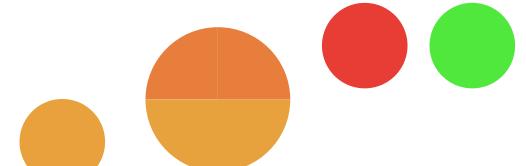
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Matching tests

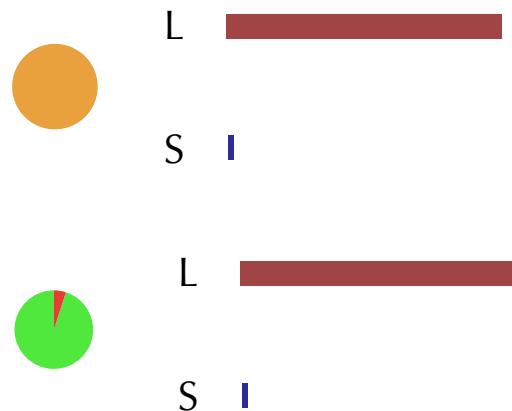
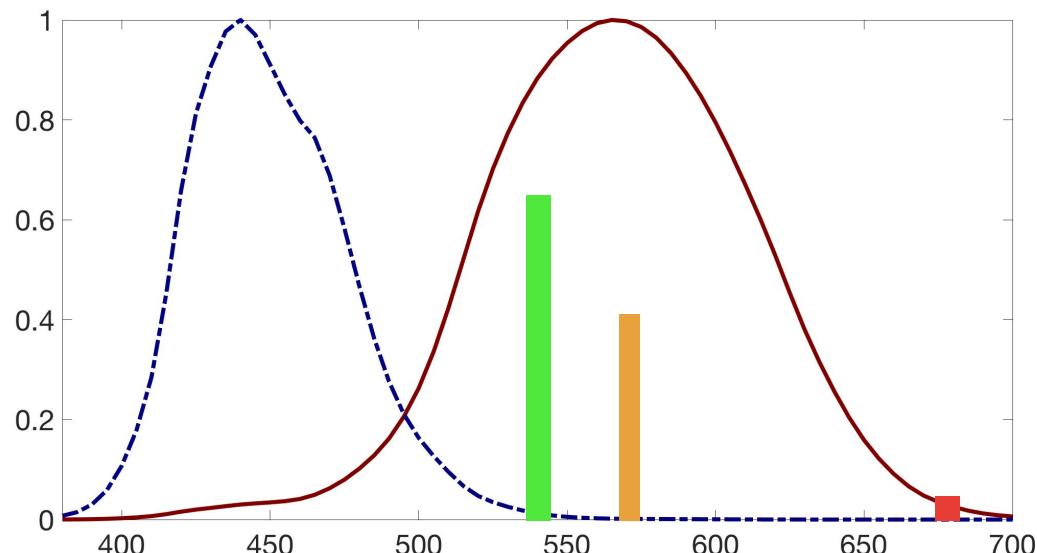


Anomaloscope (Nagel,
1907)



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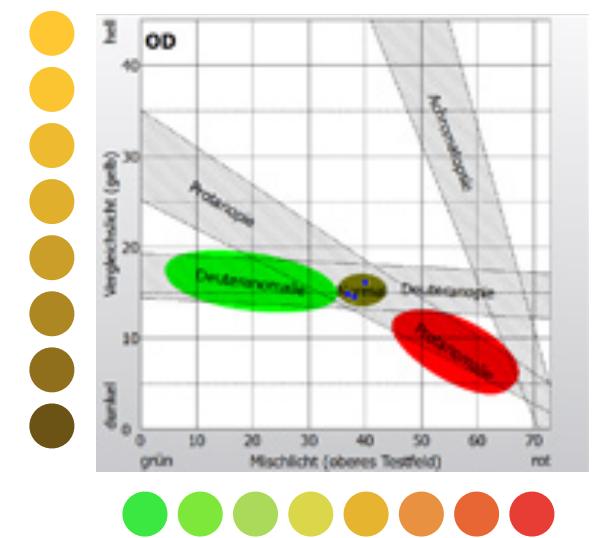
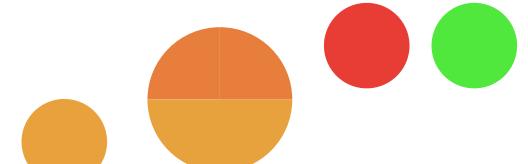
Deuteranopia



Matching tests

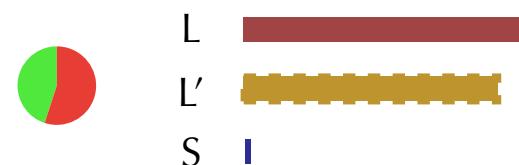
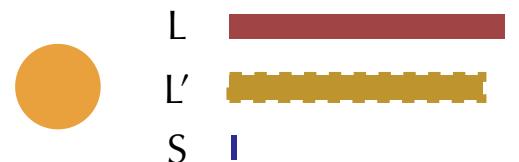
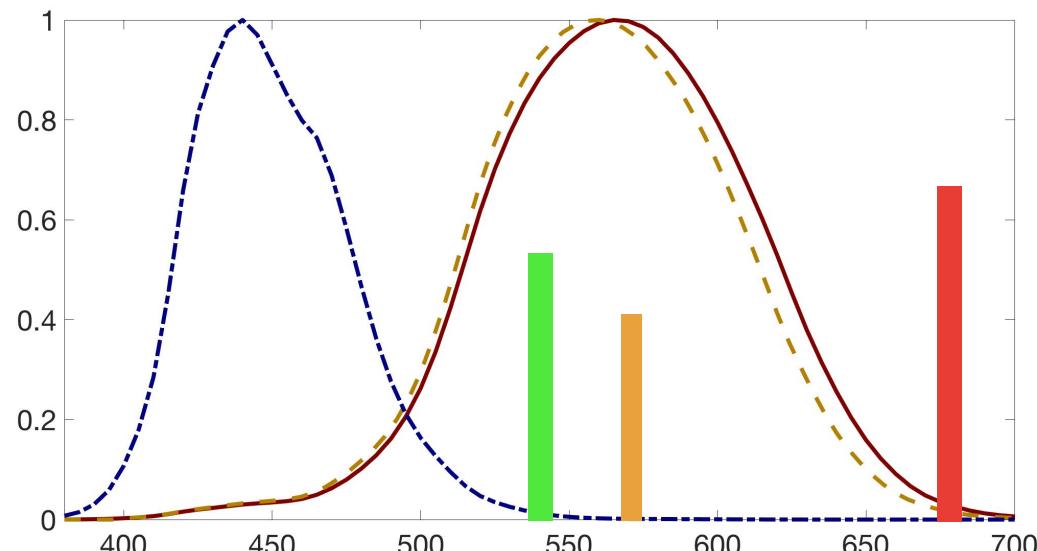


Anomaloscope (Nagel,
1907)



Diagnosing colour vision ‘deficiency’

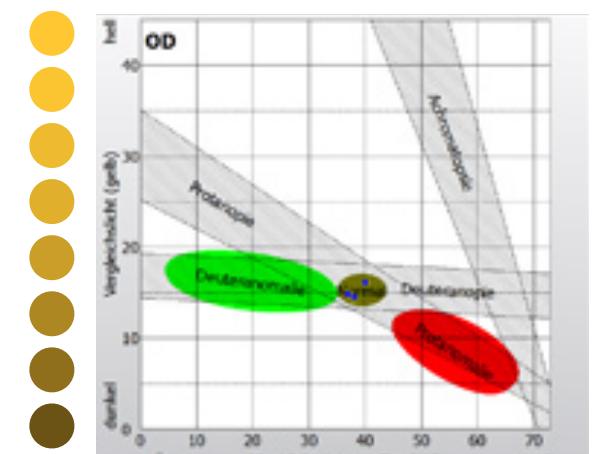
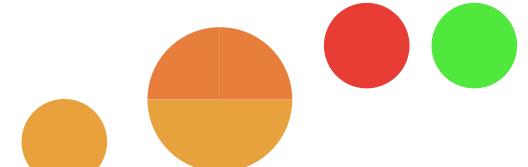
Deuteranomaly



Matching tests

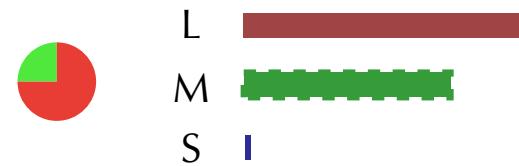
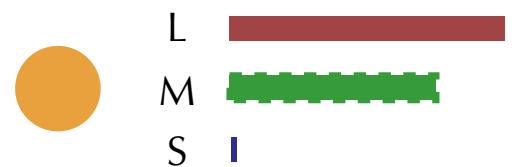
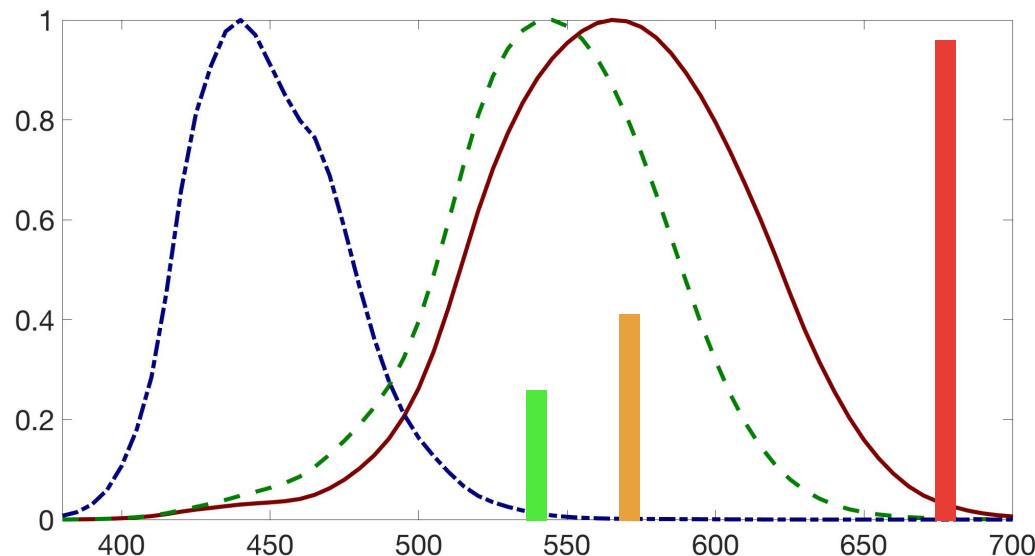


Anomaloscope (Nagel, 1907)



Diagnosing colour vision ‘deficiency’

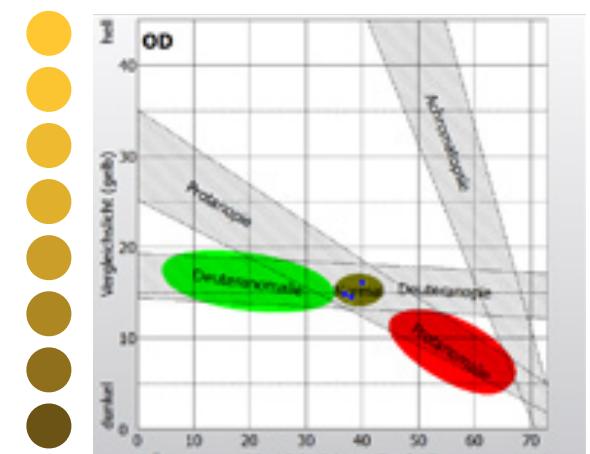
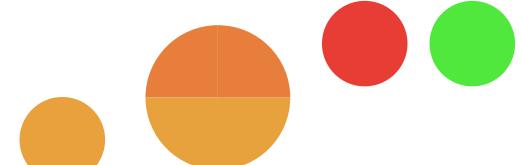
Normal trichromacy



Matching tests

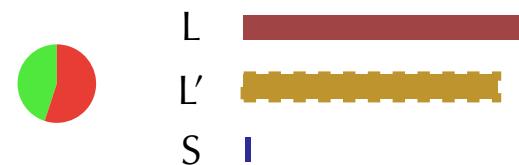
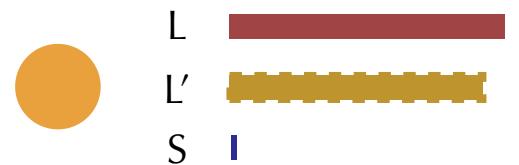
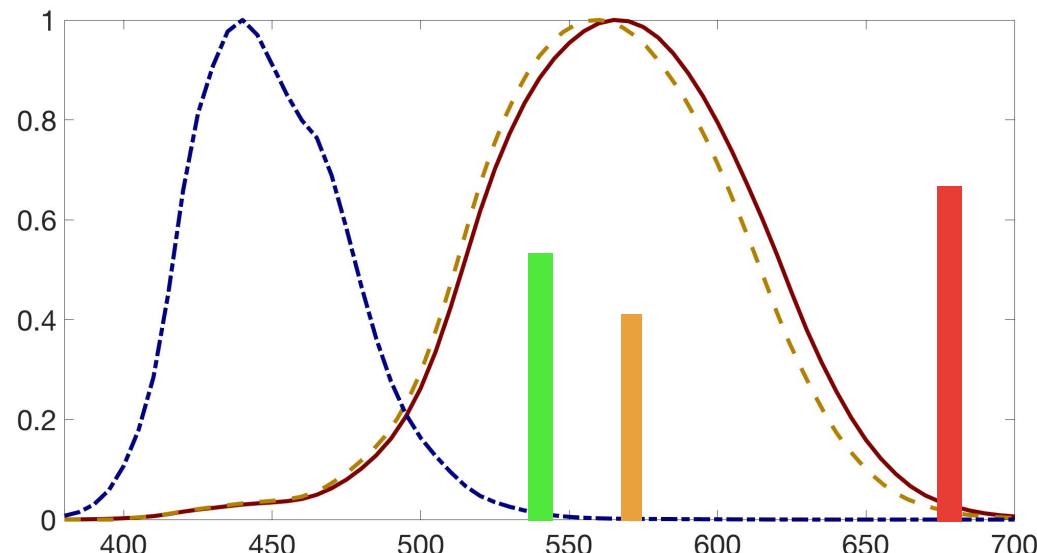


Anomaloscope (Nagel,
1907)



Diagnosing colour vision ‘deficiency’

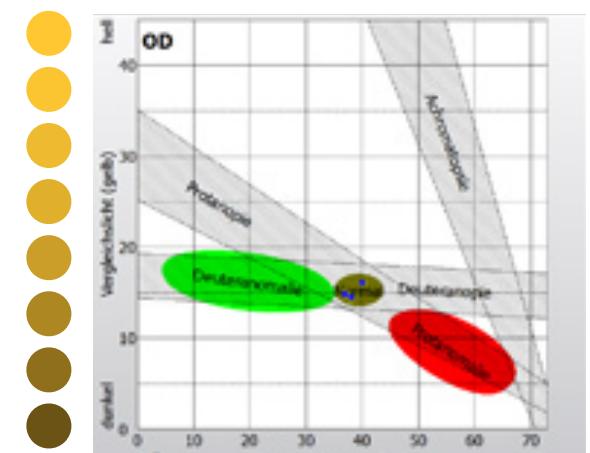
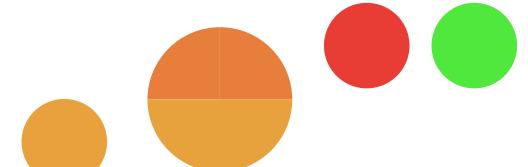
Deuteranomaly



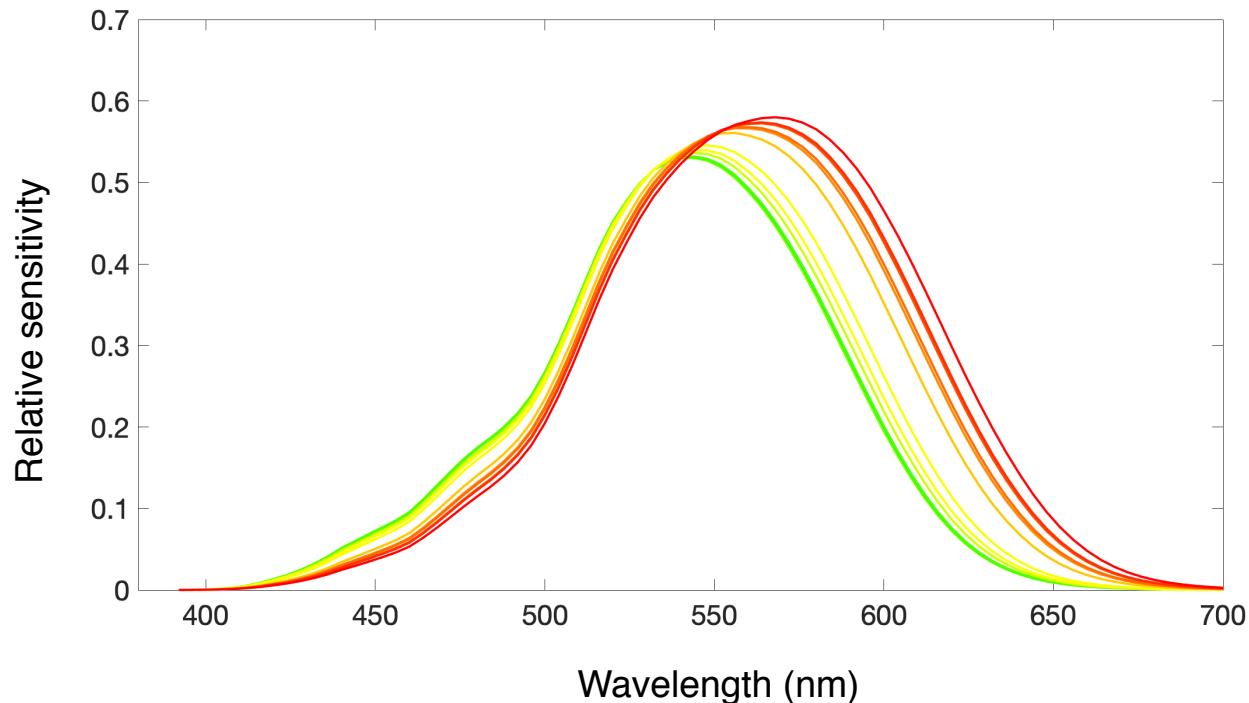
Matching tests



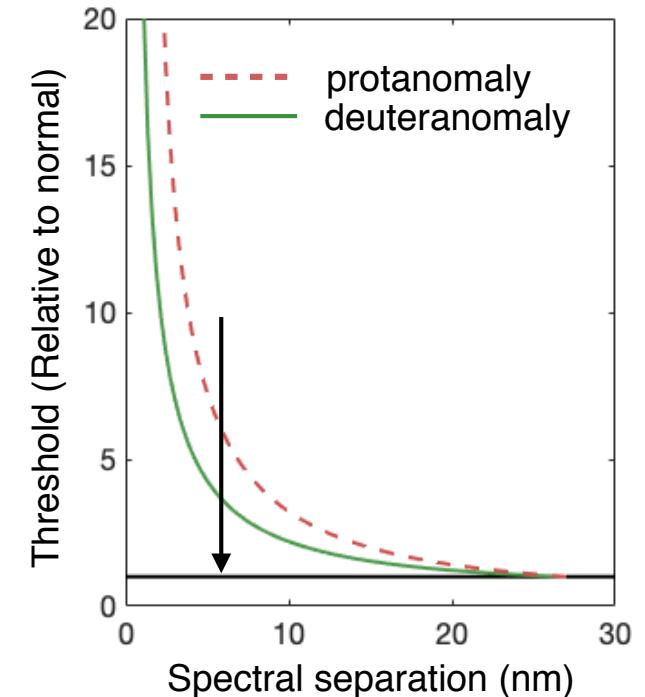
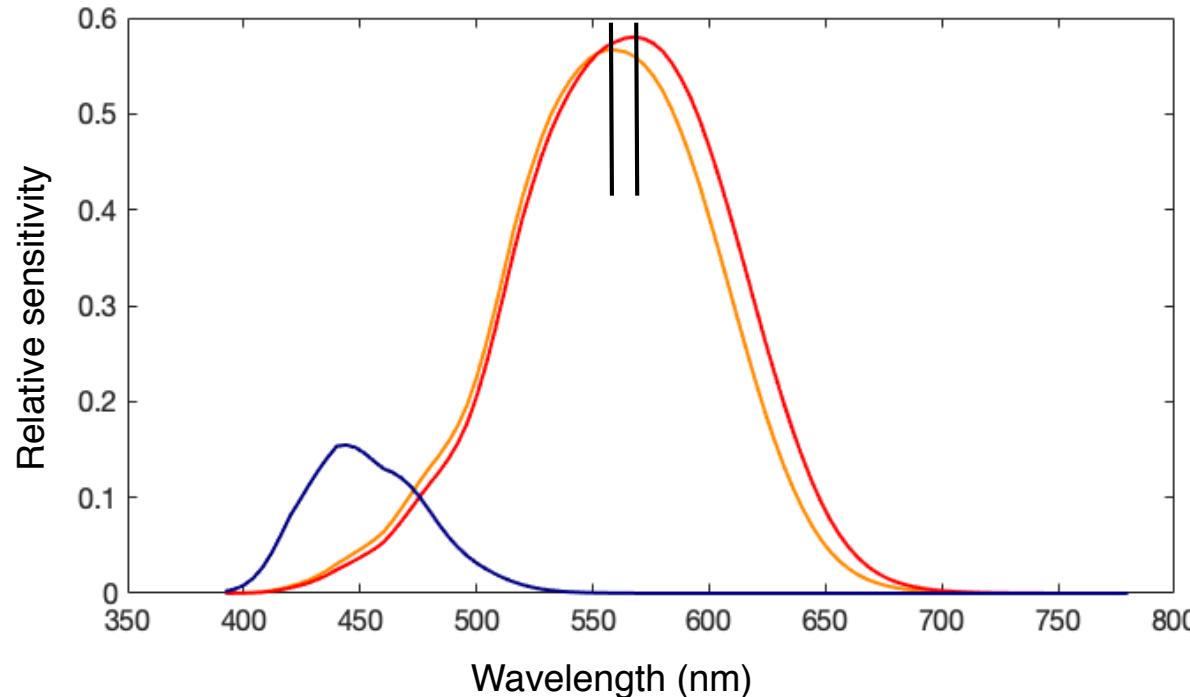
Anomaloscope (Nagel, 1907)



Anomalous cone fundamentals: model



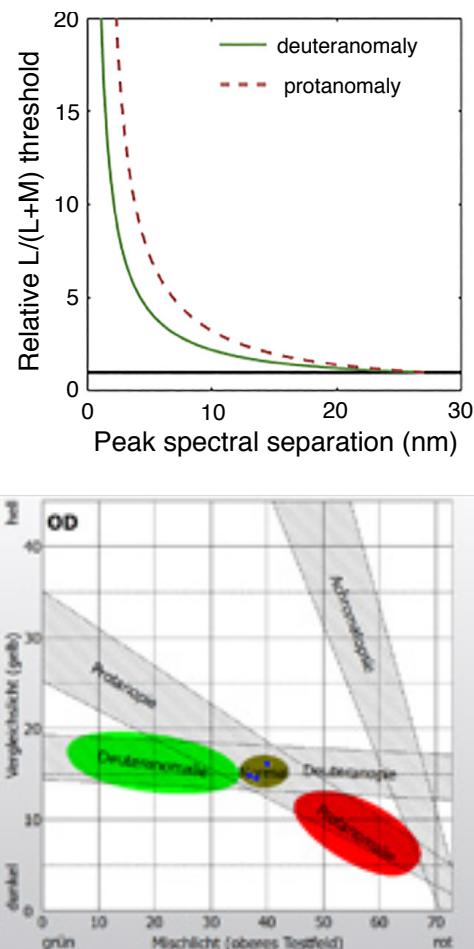
Anomalous cone fundamentals: model



Colour discrimination in anomalous trichromacy

Table 1

Correlation between spectral separation and sensitivity to red-green color differences [31*,32–35].

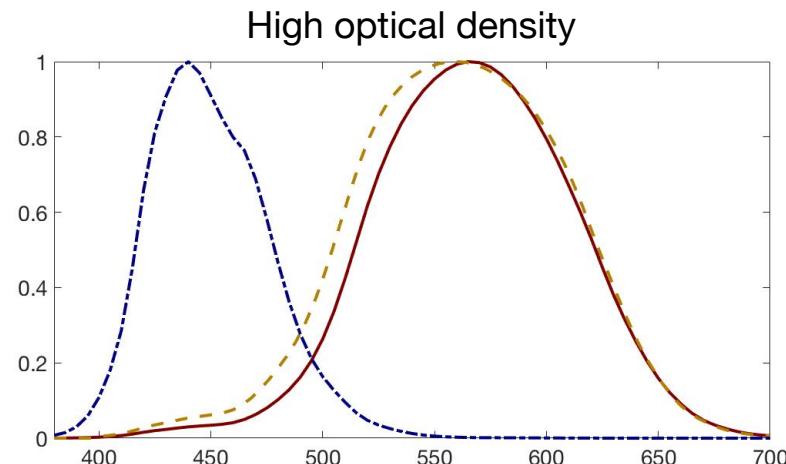
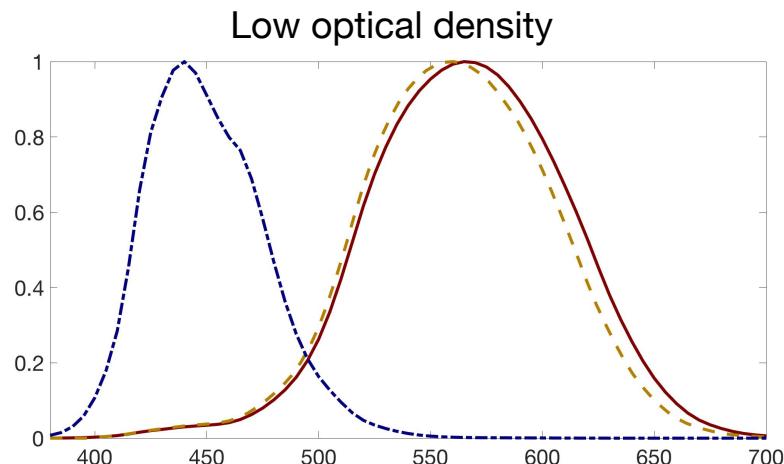


Reference and discrimination test	Deutan/ protan	Spearman's ρ	Pearson's r	n	Meta-analysis
Barbur et al. (2008) Nagel matching range [31]	D	-0.80	-0.75	11	
Barbur et al. (2008) Red-green CAD units [31]	D	-0.92	-0.85	11	
Crognale et al. (1998) small field matching range [32]	D	0.68	0.78	5	
Crognale et al. (1998) large field matching range [32]	D	-0.36	-0.3	7	
Jordan et al. (2010) matching range [23]	D	-0.35	-0.35	7	
Neitz et al. (1996) AO-HRR [10]	D	-0.93	-0.89	16	
Sanocki et al. (1997) small field matching range [33]	D	-0.37	-0.39	14	
Sanocki et al. (1997) large field matching range [33]	D	-0.45	-0.45	14	
Shevell et al. (1998) matching range [34]	D	-0.70	-0.76	8	
Deeb et al. (1993) matching range [35]	D	-0.54	-0.61	12	
Crognale et al. (1998) large field matching range [32]	P	-0.4	-0.35	7	
Crognale et al. (1998) small field matching range [32]	P	-0.51	-0.43	7	
Sanocki et al. (1997) small field matching range [33]	P	0.66	0.65	6	
Sanocki et al. (1997) large field matching range [33]	P	0.89	0.80	6	
Deeb et al. (1993) matching range [35]	P	-0.26	0.24	6	

N.B. Participants who are genetically or phenotypically dichromatic were excluded.

Relationship between cone fundamentals and colour discrimination

- Poor reliability and validity of matching range as a measure of colour discrimination
- Other individual differences: **Optical density variation by cone type** (Thomas et al. 2011; Humanski and Shevell, 1991), rods, IPRGCs, chromatic aberration, dynamic signals (Broakes, 2010).
- Postreceptoral compensation



Variation in Color Matching and
Discrimination Among Deutanomalous
Trichromats: Theoretical Implications of
Small Differences in Photopigments

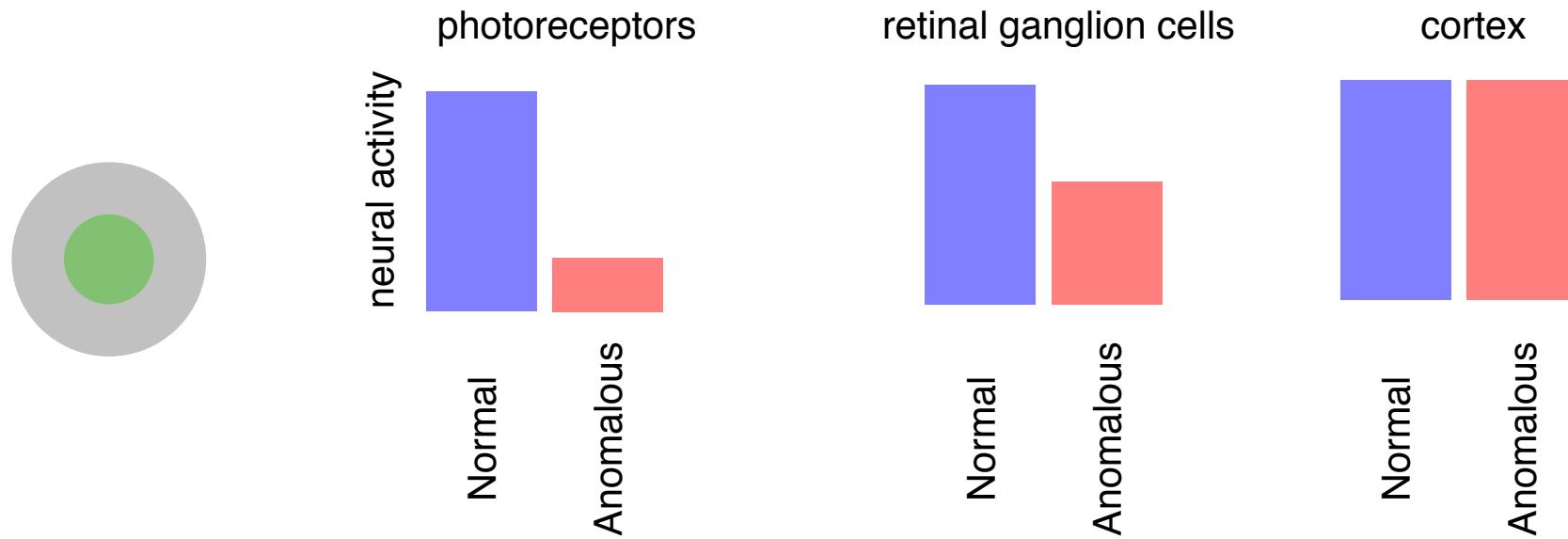
JI CHANG HE,* STEVEN K. SHEVELL†

The effect of photopigment optical density on the color vision
of the anomalous trichromat

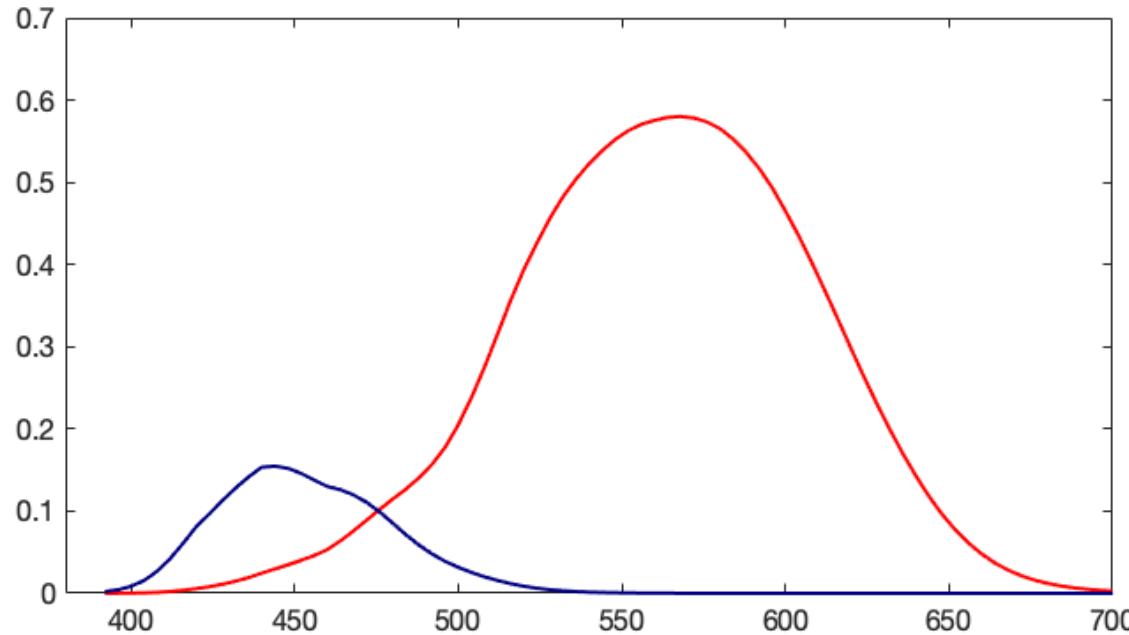
P.B.M. Thomas ^{a,b,*}, M.A. Formankiewicz ^{b,c}, J.D. Mollon ^b

Relationship between cone fundamentals and colour discrimination

- Poor reliability and validity of matching range as a measure of colour discrimination
- Other individual differences: Optical density variation by cone type (Thomas et al. 2011; Humanski and Shevell, 1991), rods, IPRGCs, chromatic aberration, dynamic signals (Broakes, 2010).
- **Postreceptoral compensation**



Anomalous cone fundamentals: model



529.7nm; 556.7 nm:
normal trichromacy



544.8nm; 556.7 nm:
deuteranomaly



548.8nm; 556.7 nm:
deuteranomaly



552.4; 556.7 nm:
deuteranomaly



555nm; 556.7 nm:
deuteranomaly



556.7 nm:
deutanopia



Suprathreshold compensation

80% postceptroal compensation

529.7nm; 556.7 nm:
normal trichromacy



544.8nm; 556.7 nm:
deuteranomaly



548.8nm; 556.7 nm:
deuteranomaly



552.4; 556.7 nm:
deuteranomaly



555nm; 556.7 nm:
deuteranomaly



556.7 nm:
deuteranopia



No compensation

529.7nm; 556.7 nm:
normal trichromacy



544.8nm; 556.7 nm:
deuteranomaly



548.8nm; 556.7 nm:
deuteranomaly



552.4; 556.7 nm:
deuteranomaly



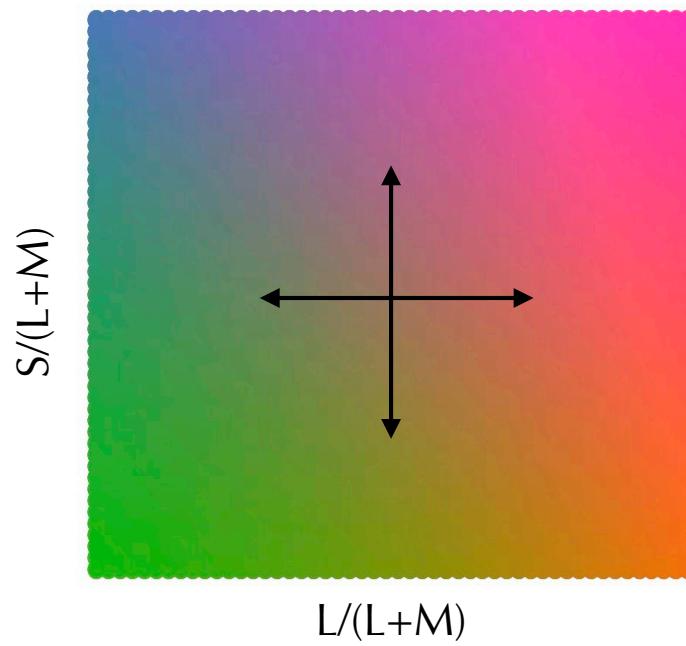
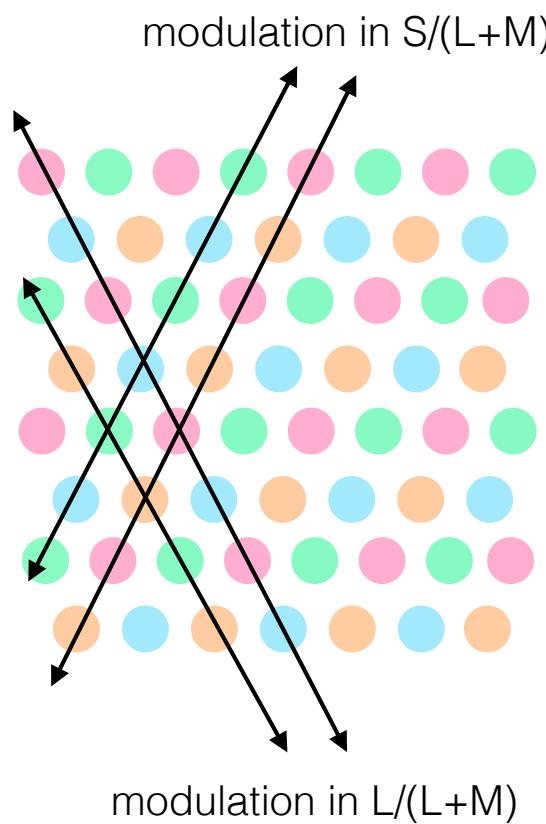
555nm; 556.7 nm:
deuteranomaly



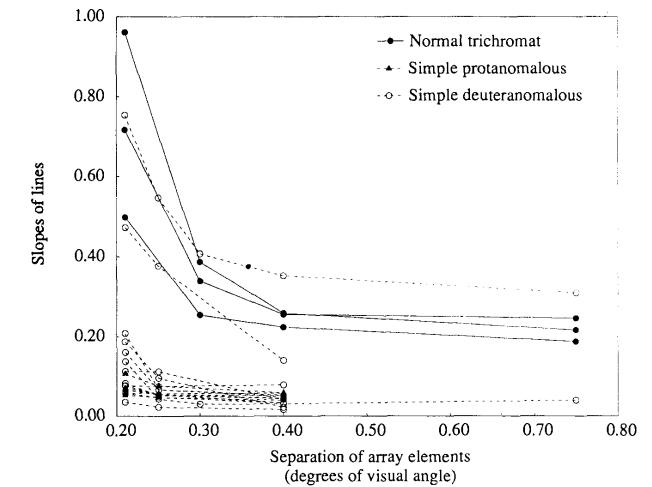
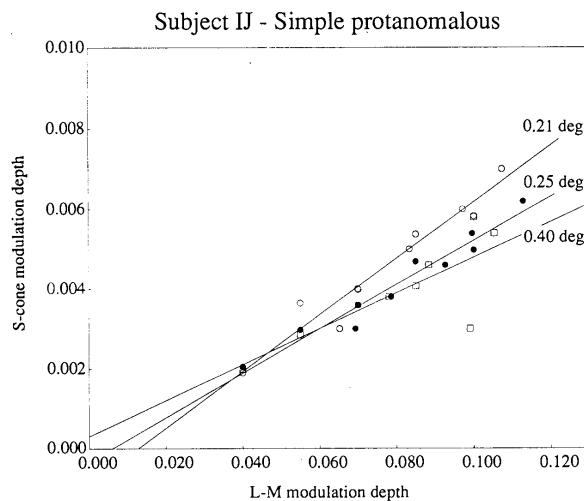
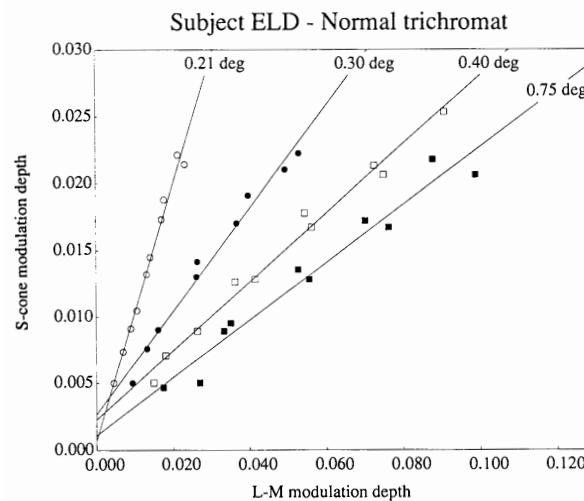
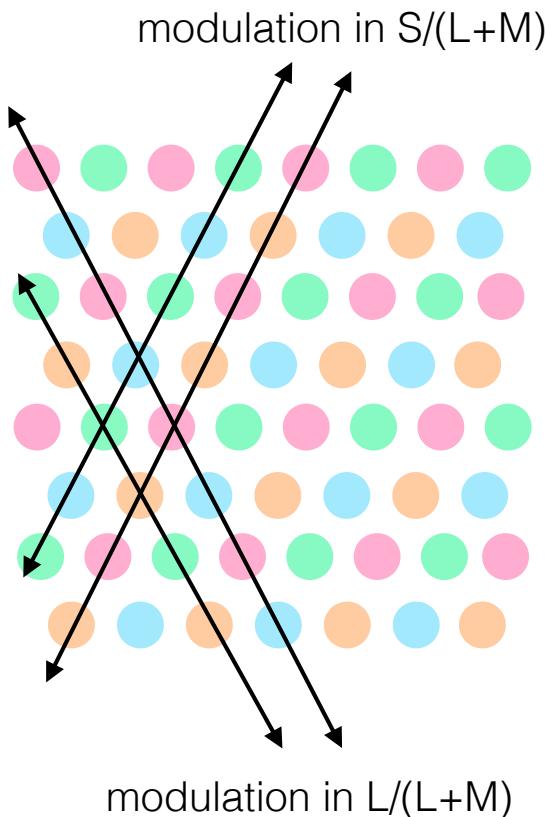
556.7 nm:
deuteranopia



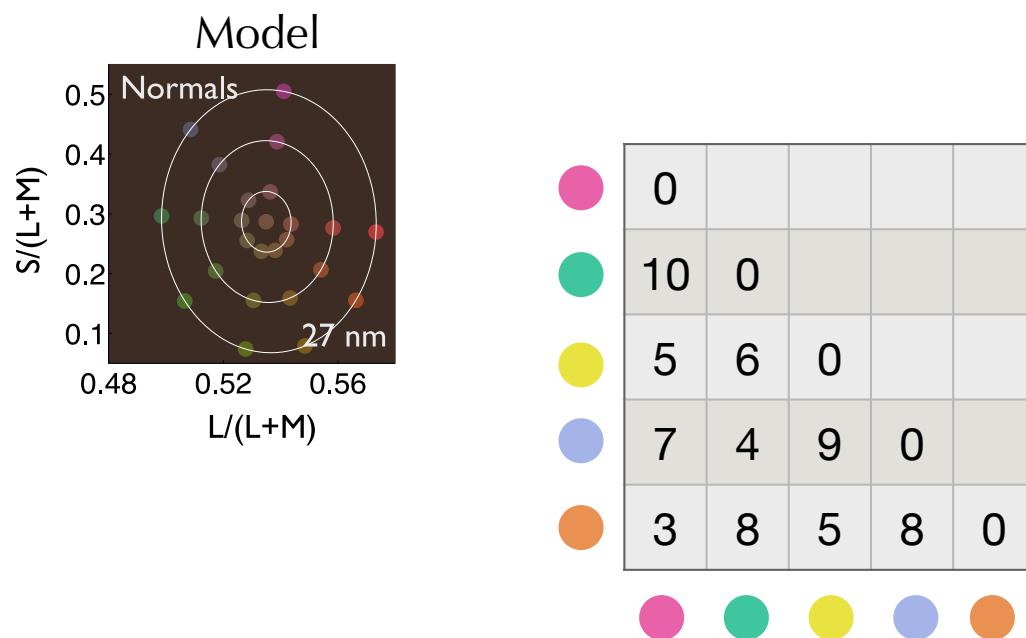
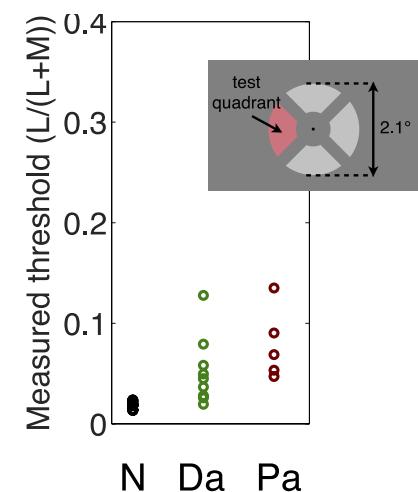
Postreceptoral compensation



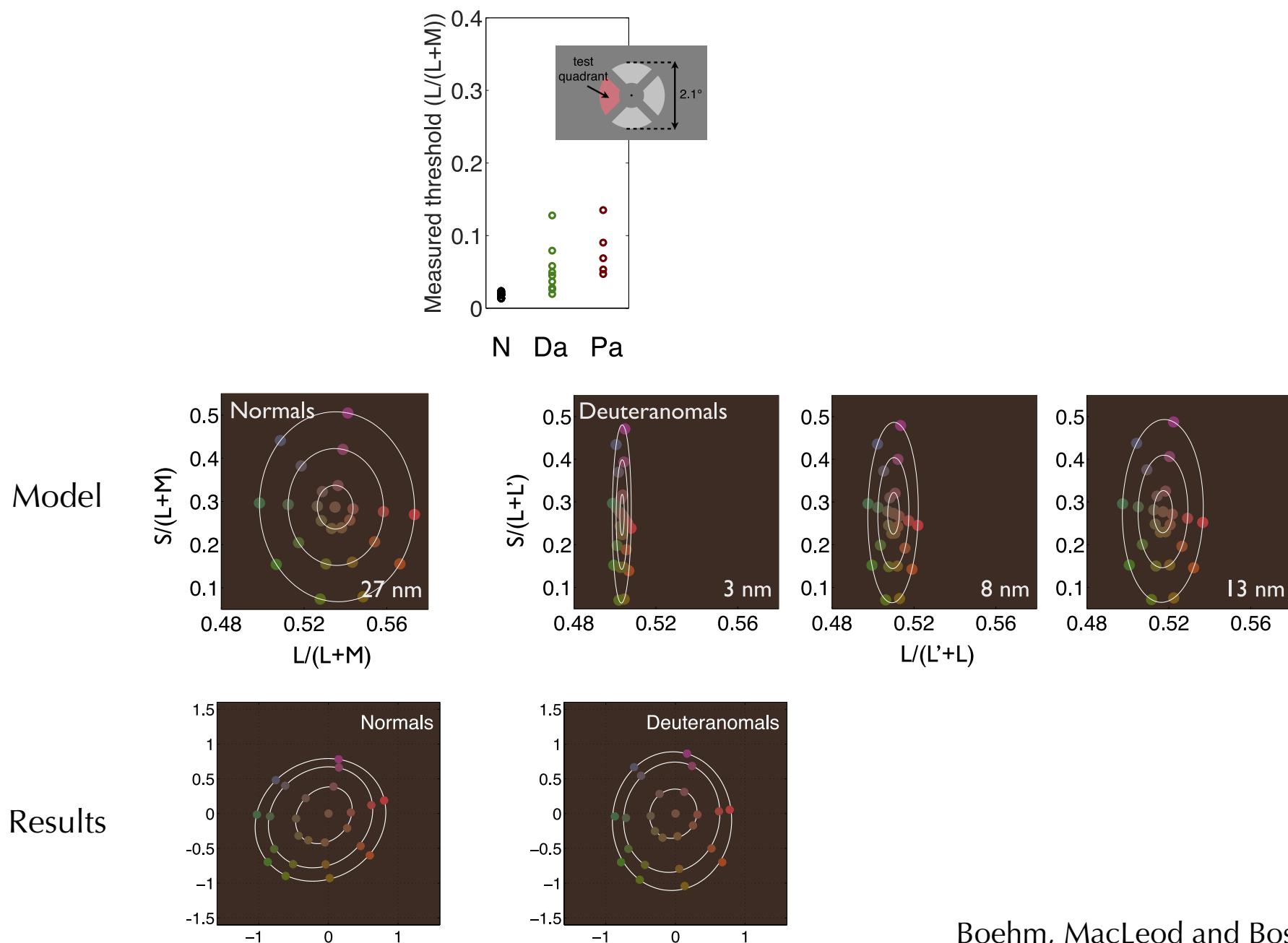
Postreceptoral compensation



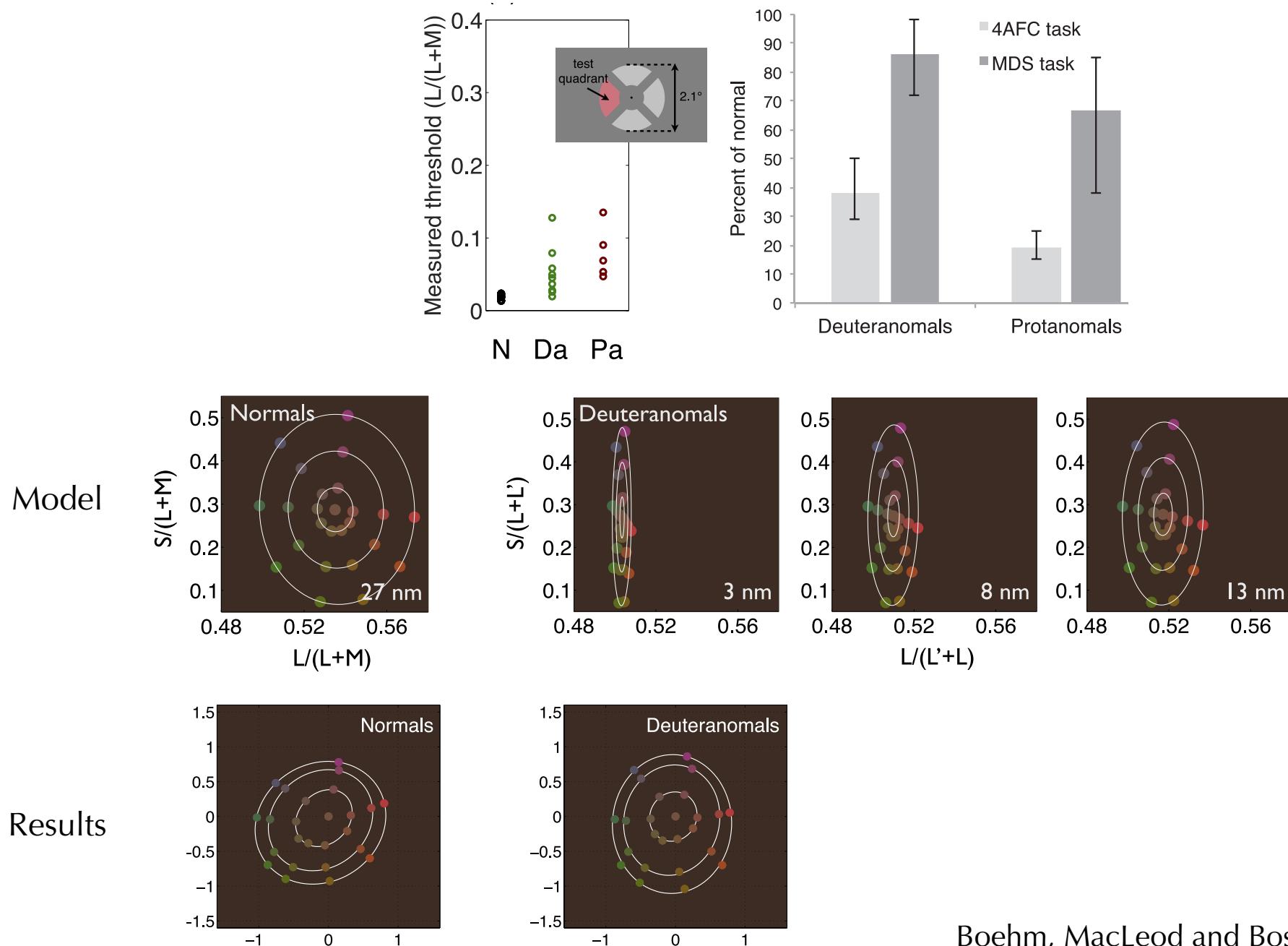
Postreceptoral compensation



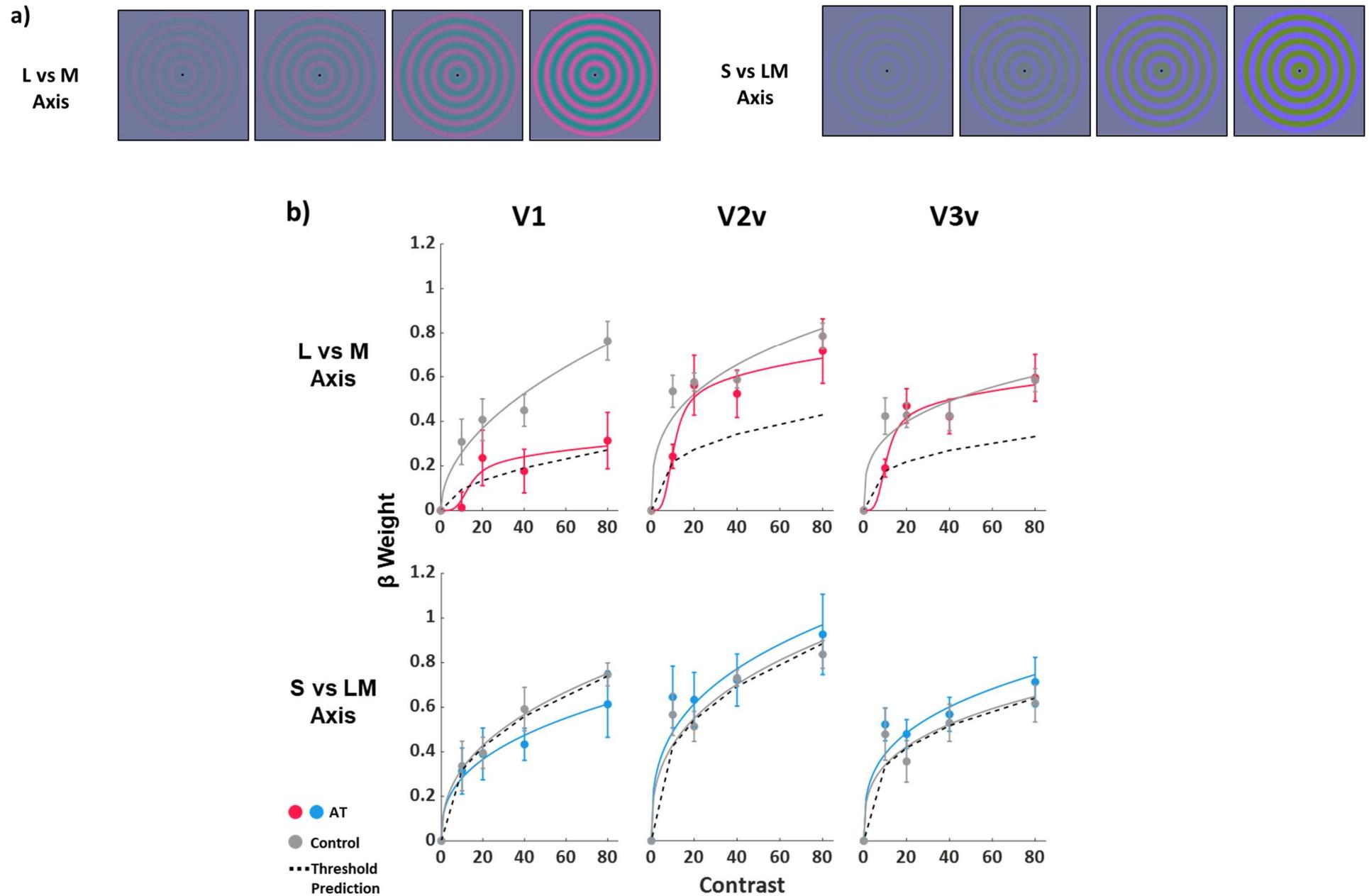
Postreceptoral compensation



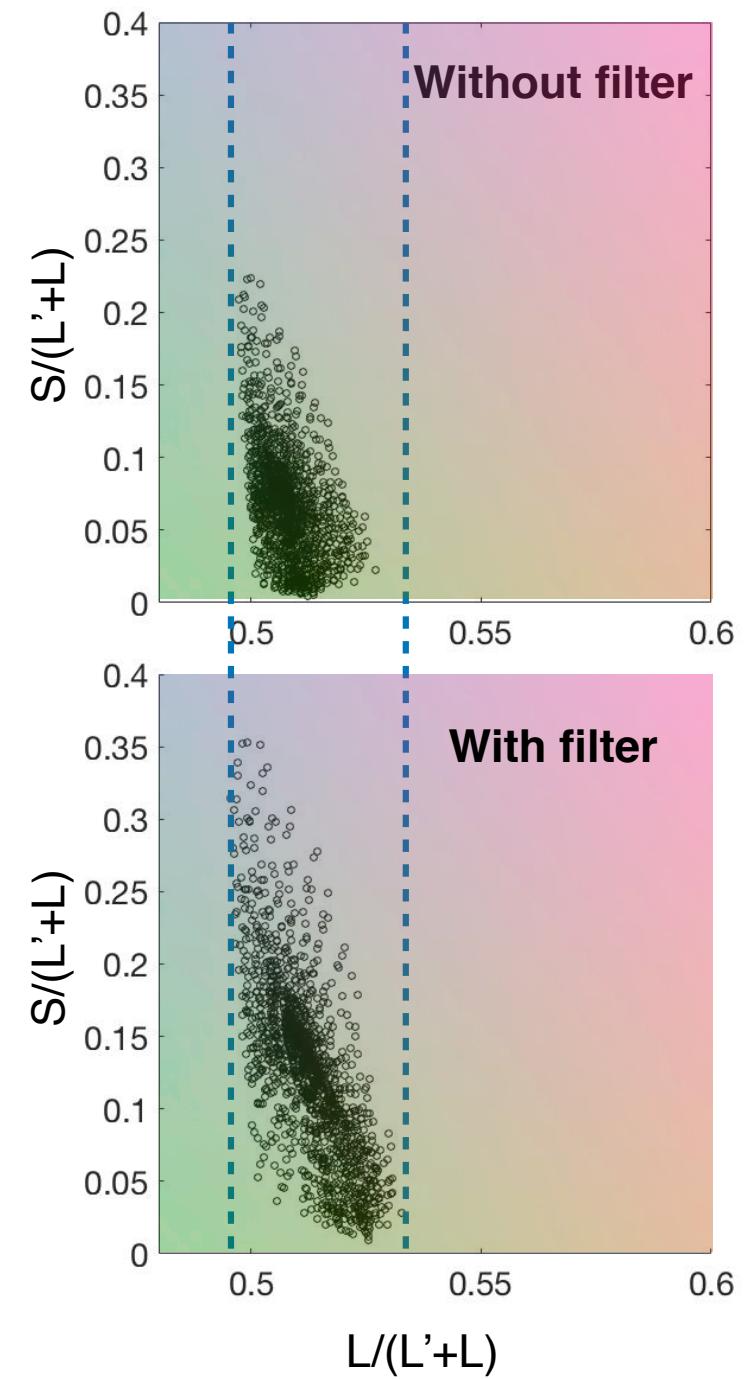
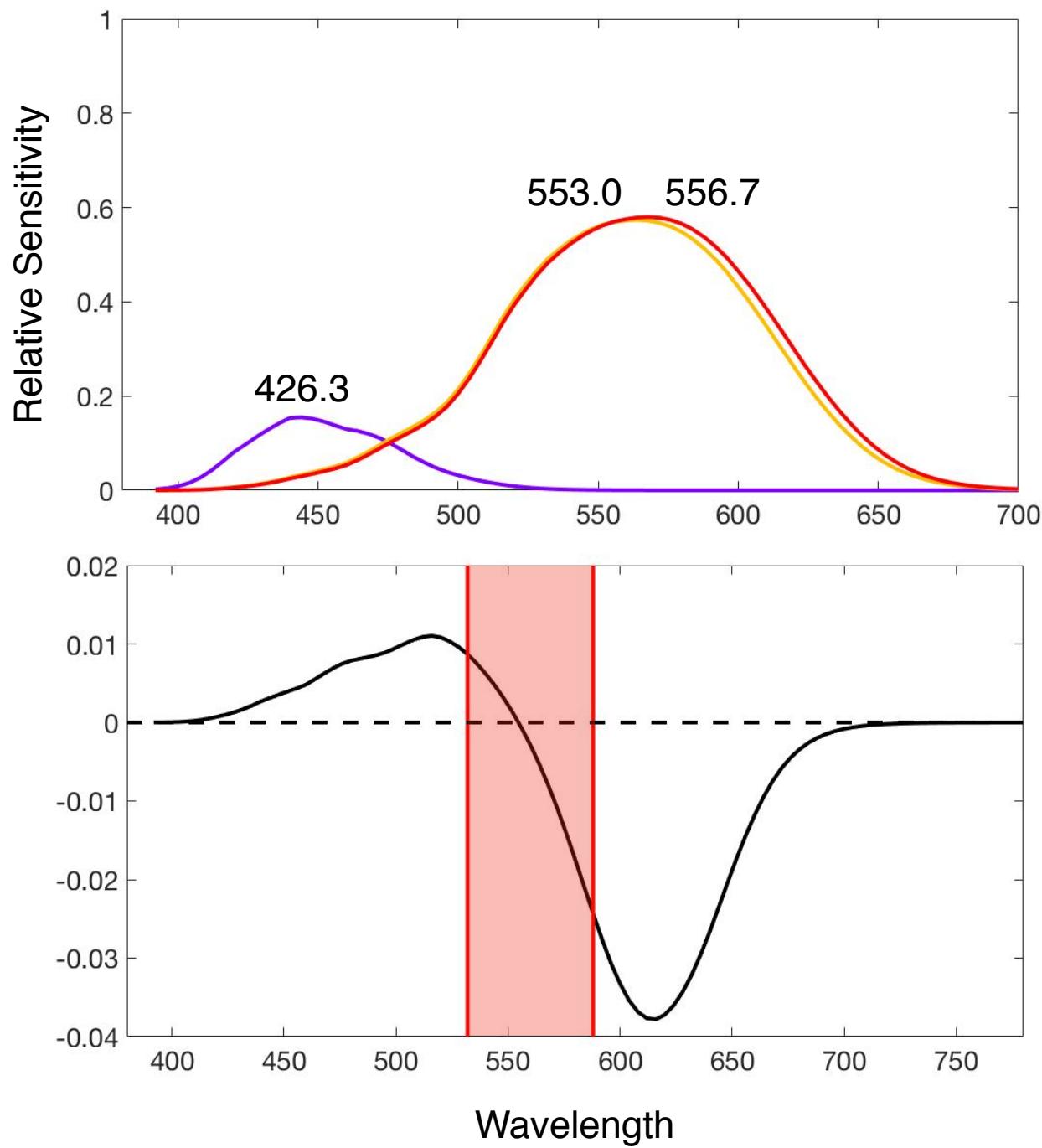
Postreceptoral compensation



Postreceptoral compensation



Notch filters





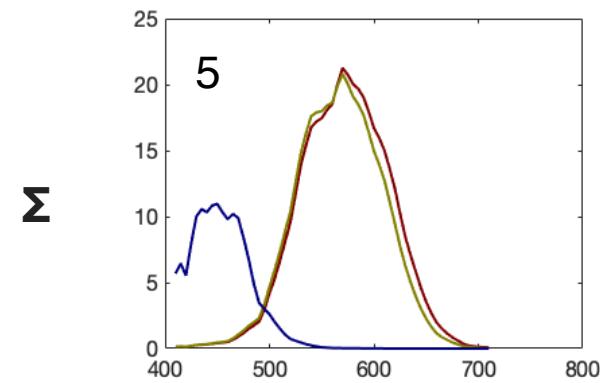
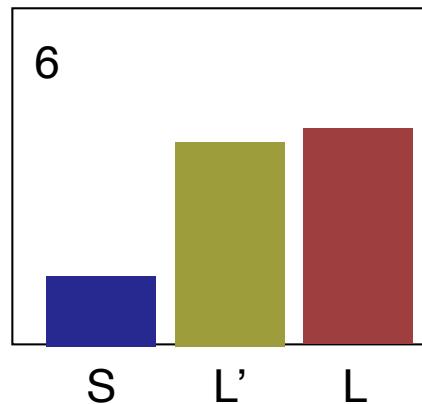
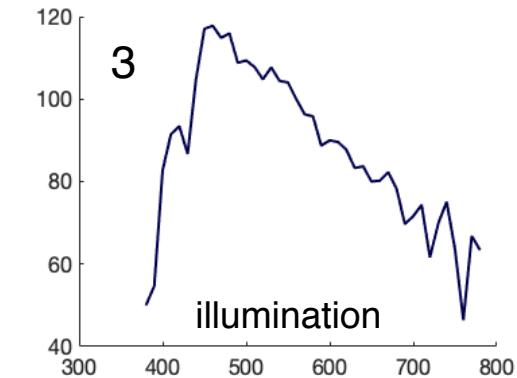
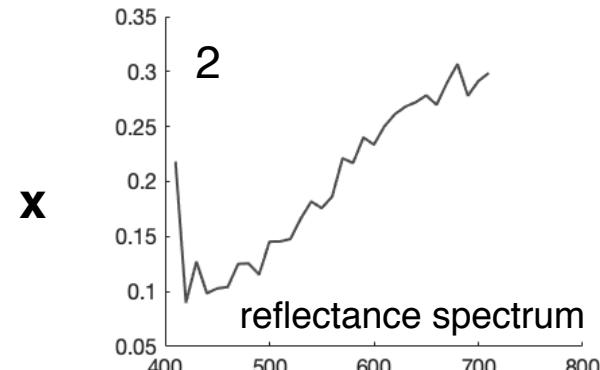
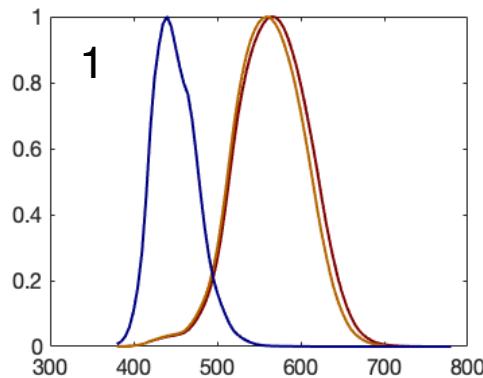
EnChroma

**“Bring greater vibrancy and colour to
your world with EnChroma”**

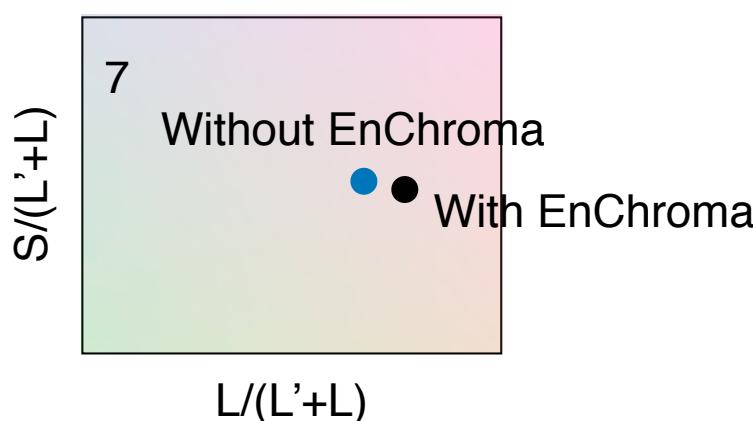




EnChroma: The model

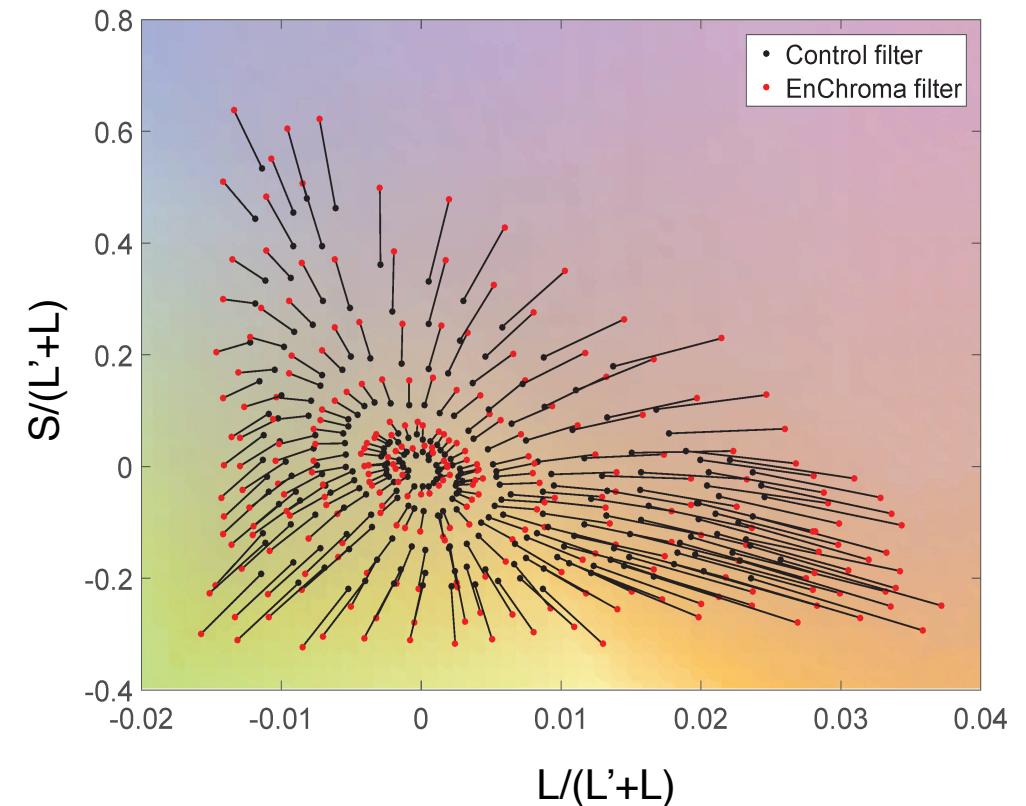
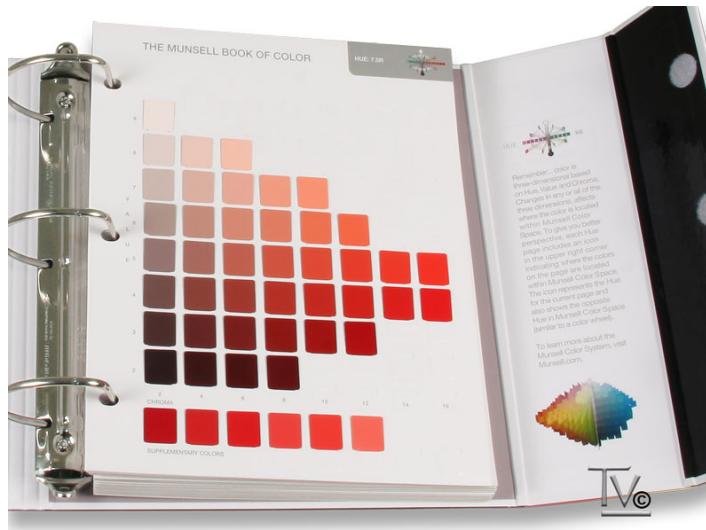
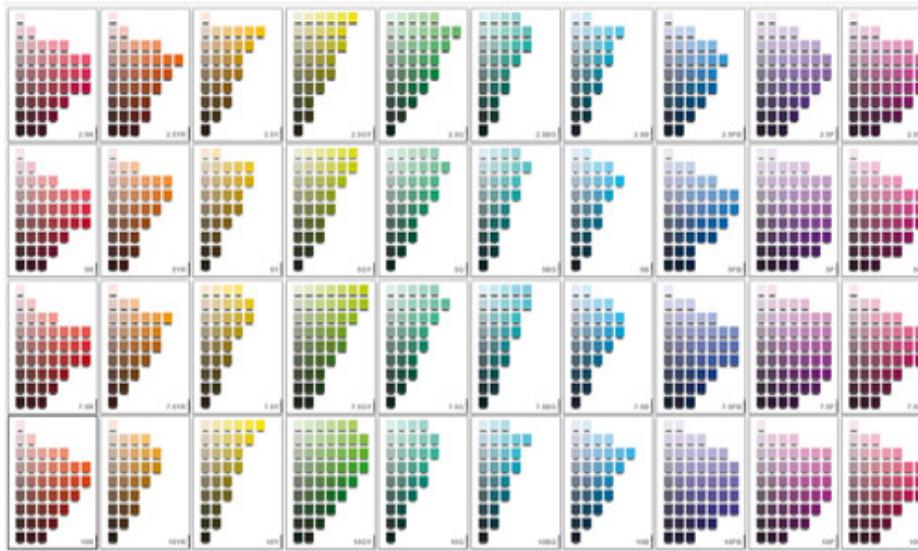


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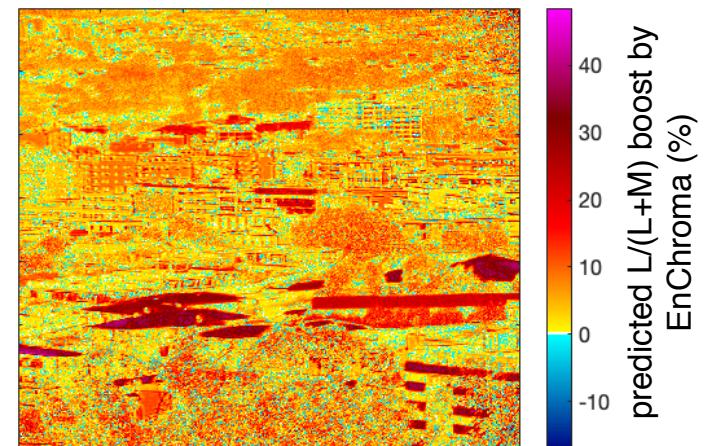
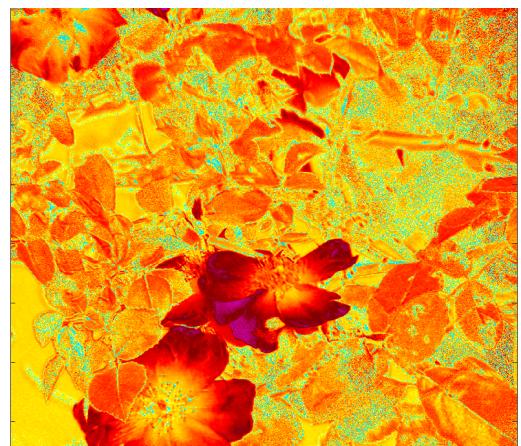
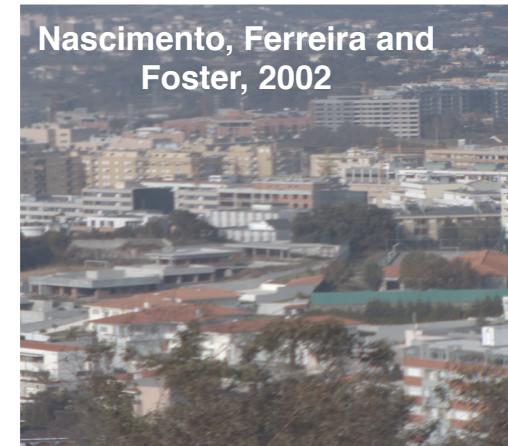
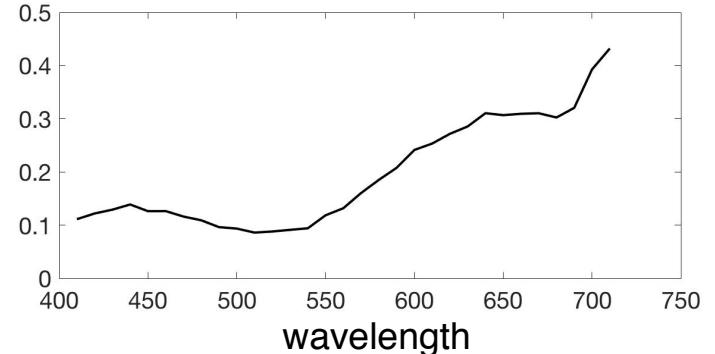
EnChroma: The model

Munsell colours



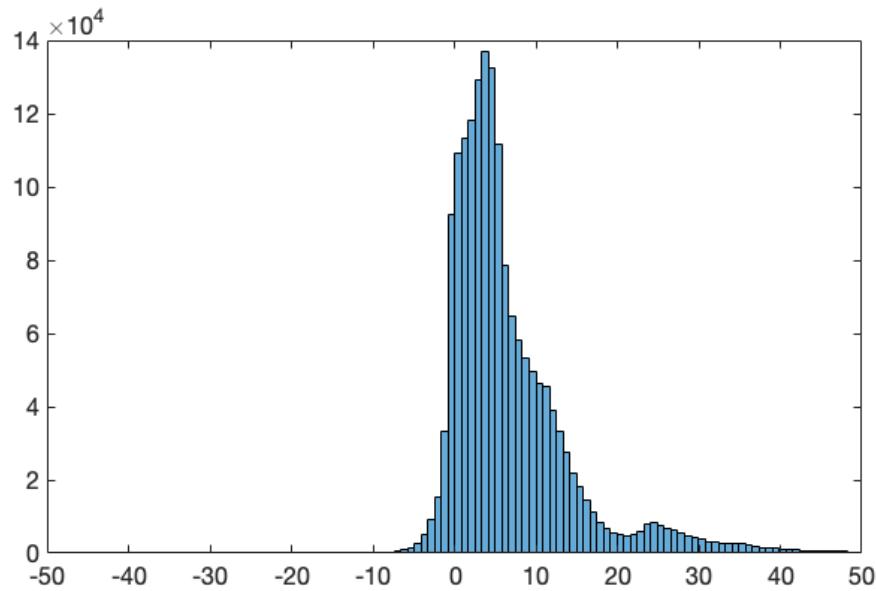


EnChroma: The model

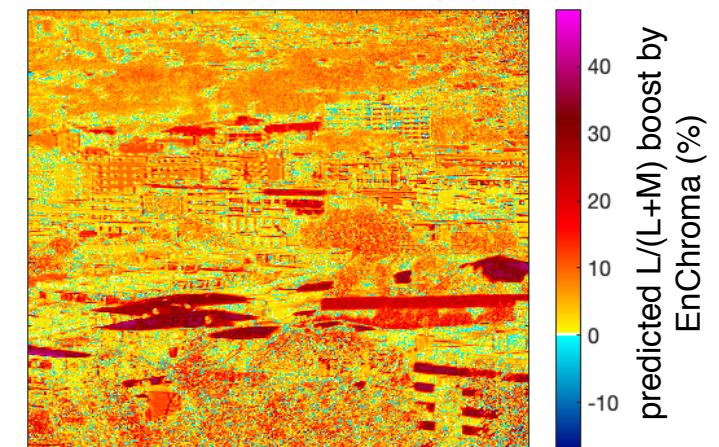
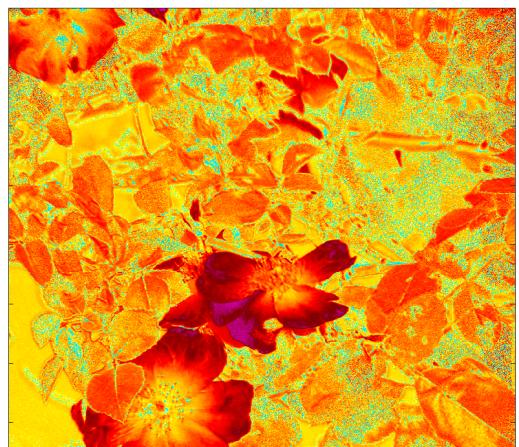




EnChroma: The model



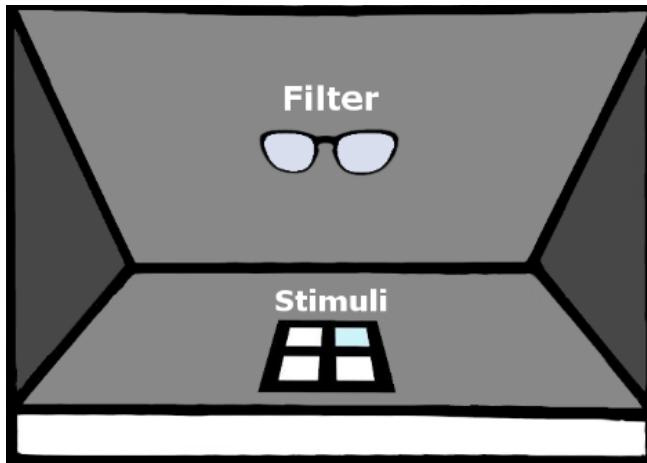
predicted $L/(L+M)$ boost by EnChroma
(as a % of $L/(L+M)$ gamut)



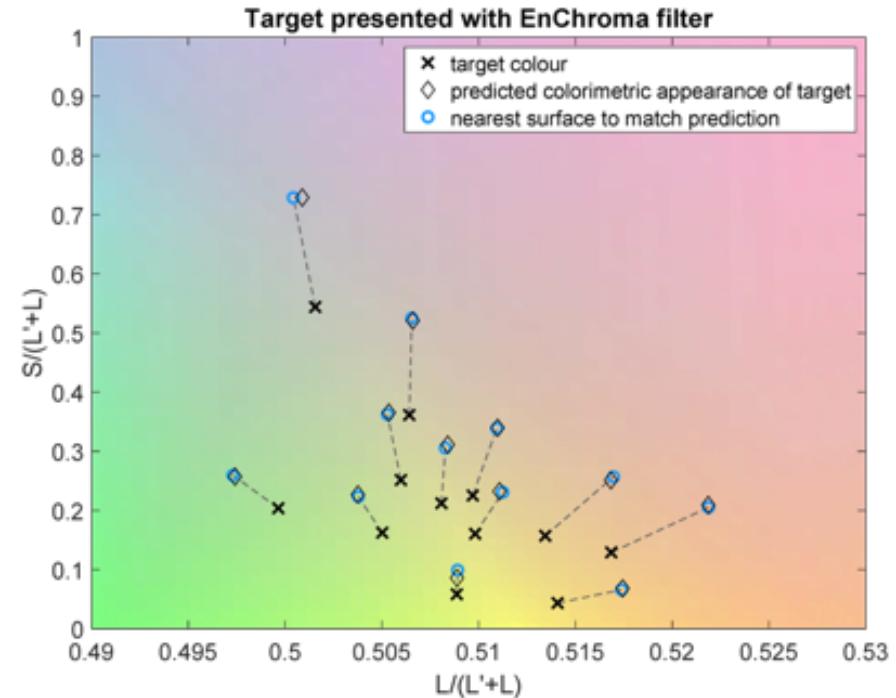
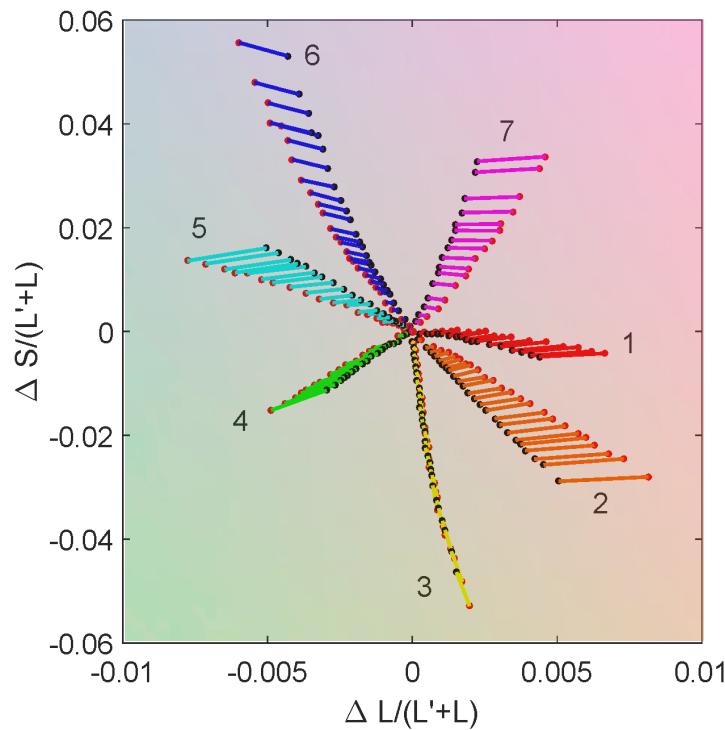
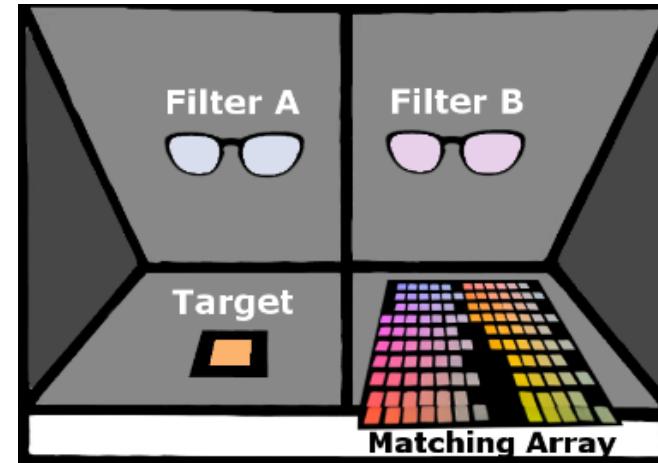
Nascimento, Ferreira and Foster, 2002

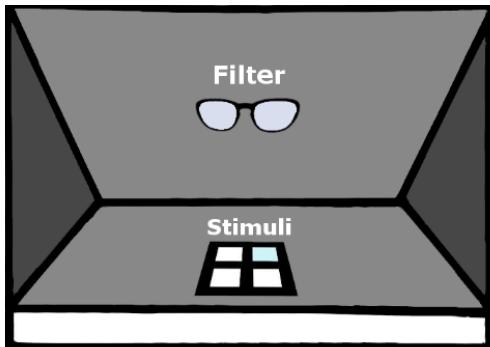
EnChroma: The test

Task 1: 4AFC

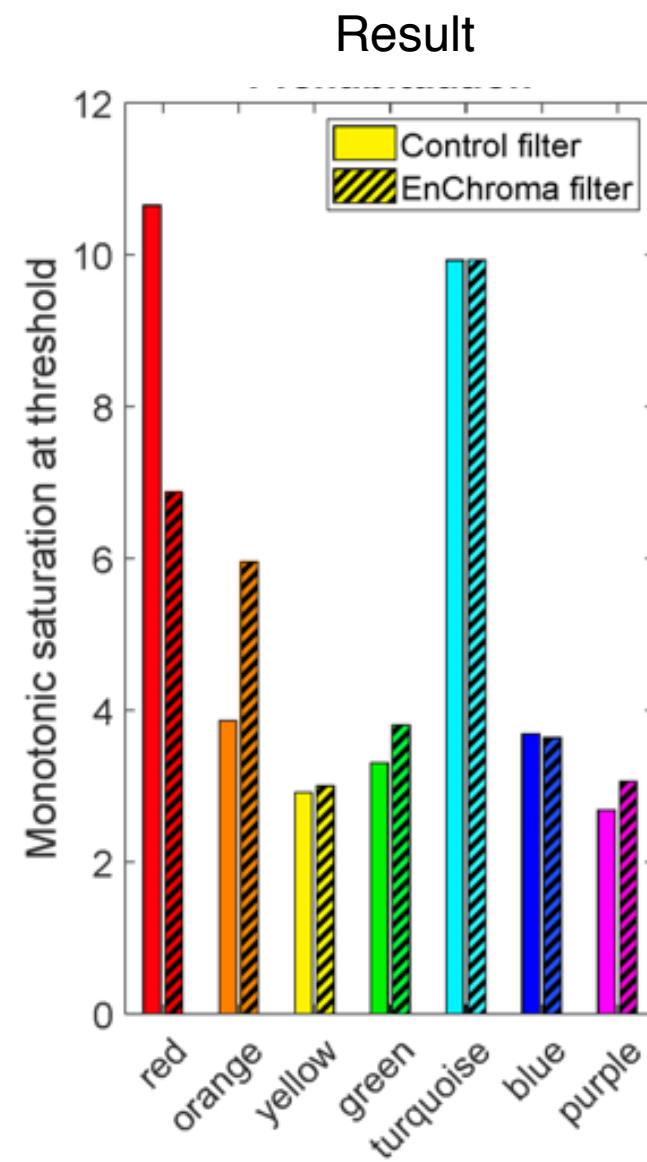
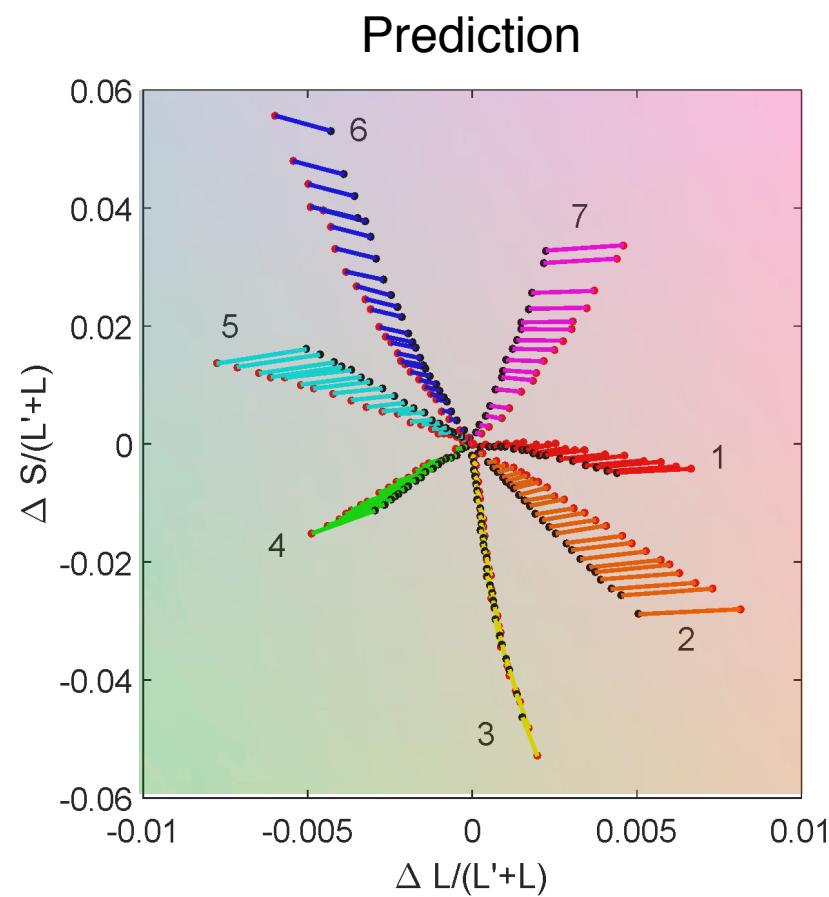


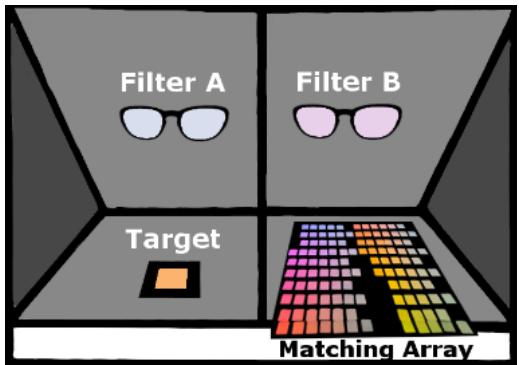
Task 2: Metameric matching





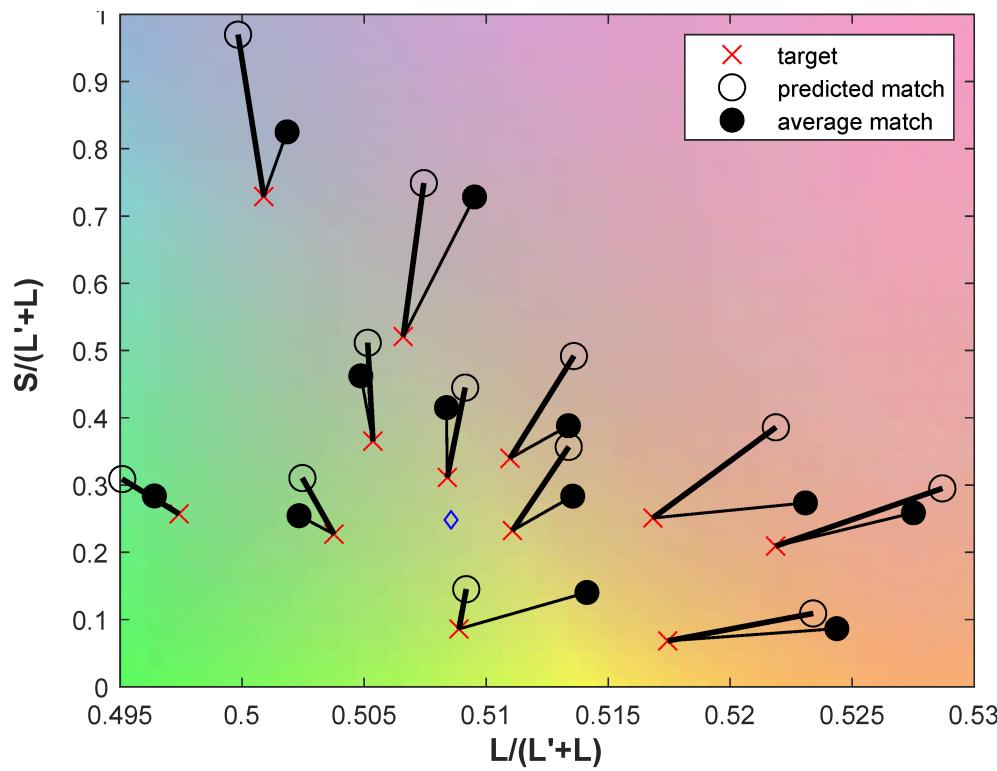
EnChroma: Results task 1



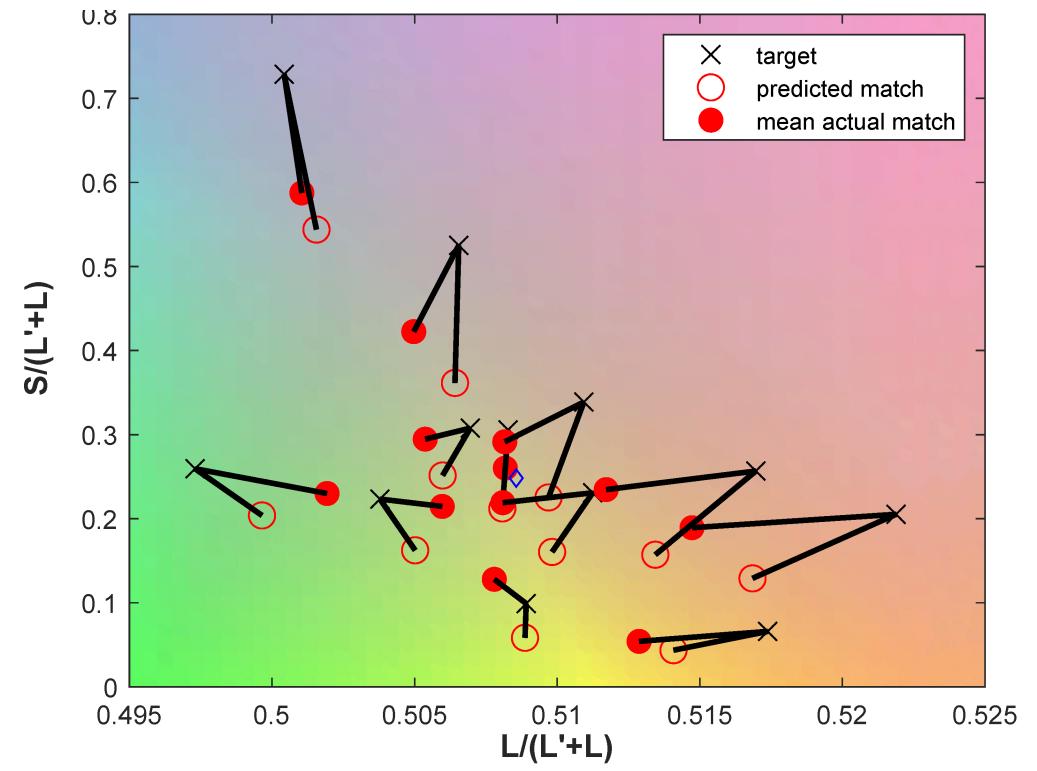


EnChroma: Results task 2

Target viewed under EnChroma

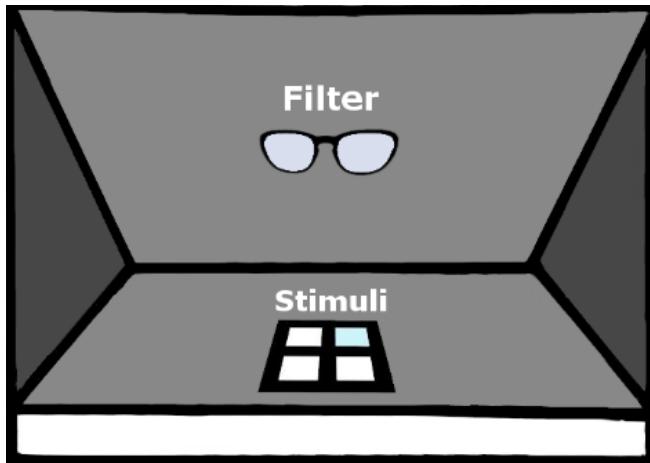


Target viewed under control

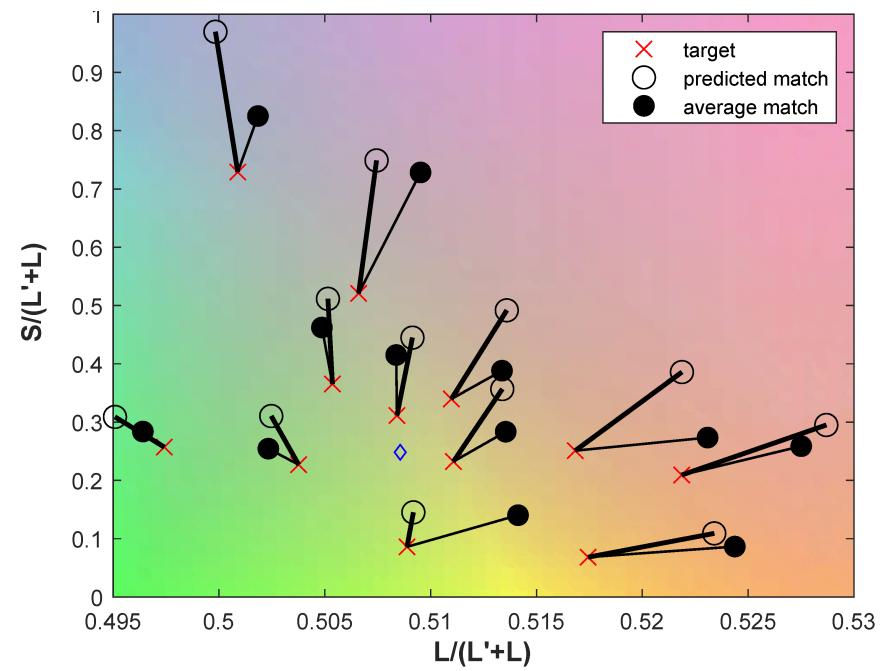
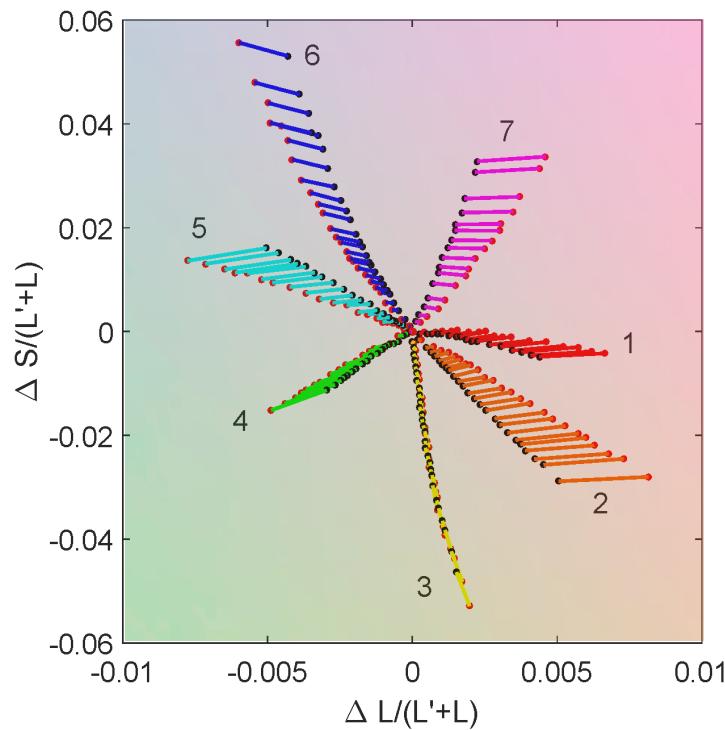
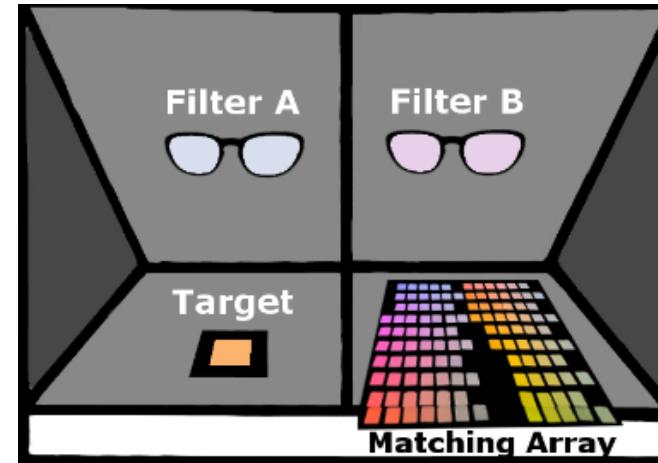


EnChroma: Conclusions

Task 1: 4AFC



Task 2: Metameric matching



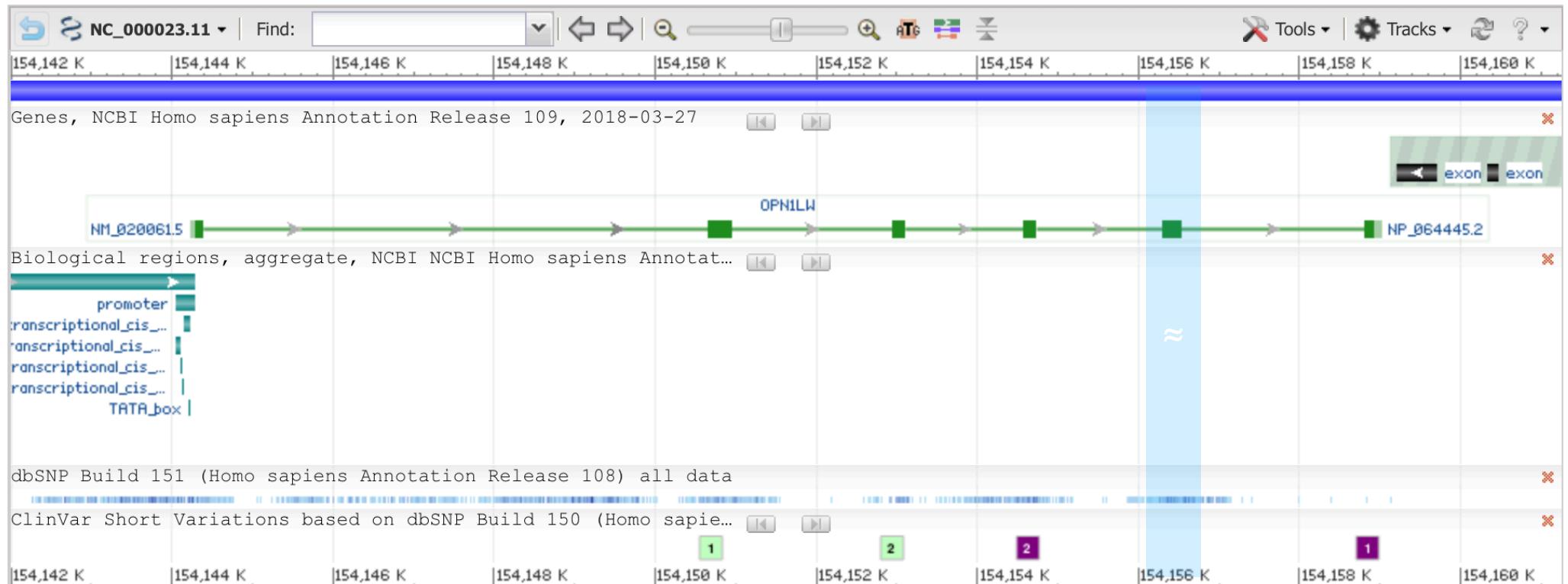
Differences between M and L opsins genes



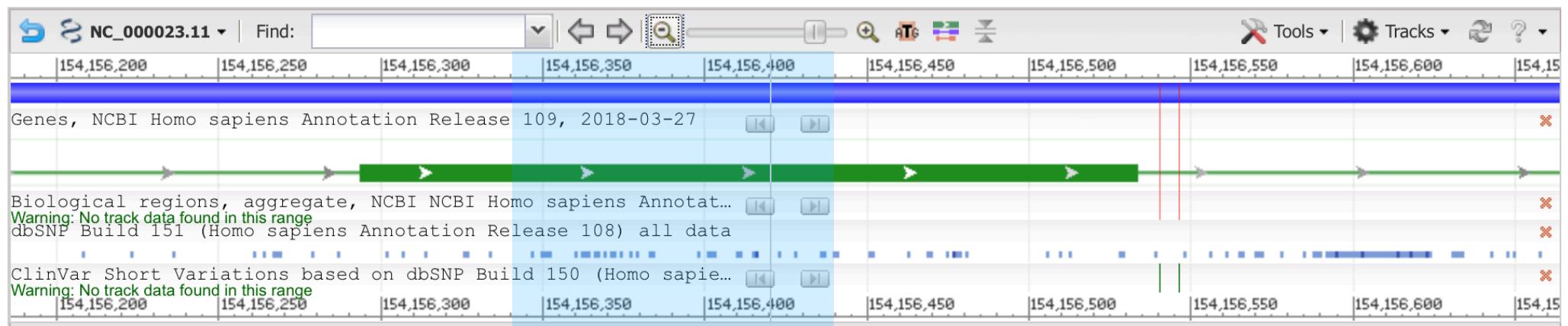
Differences between M and L opsins genes



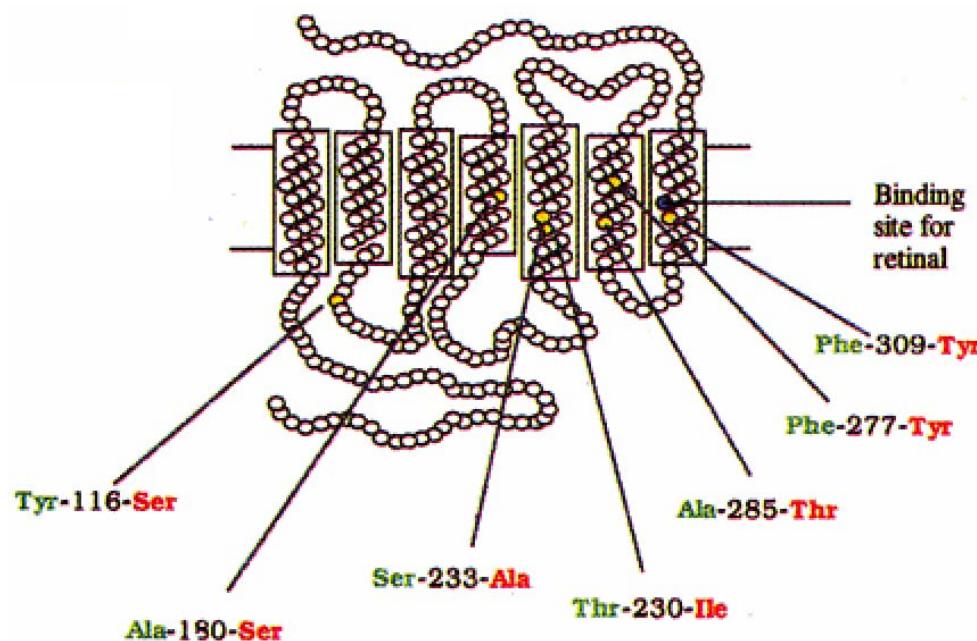
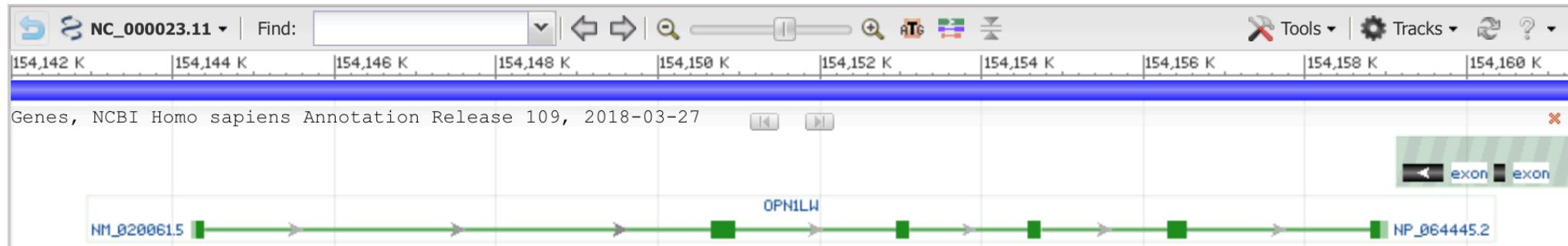
Differences between M and L opsins genes



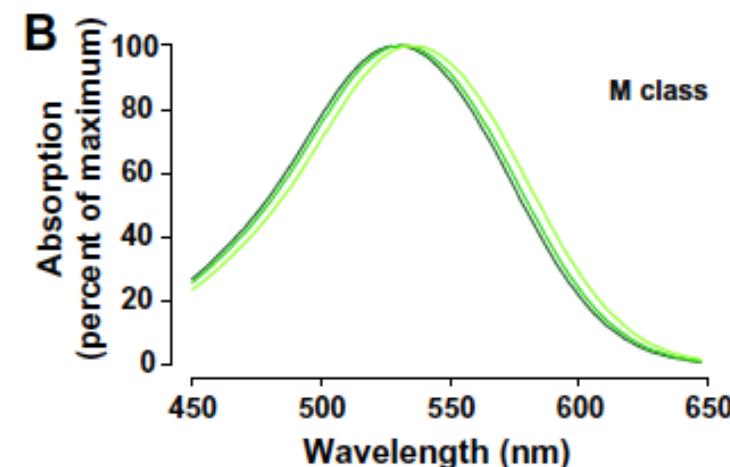
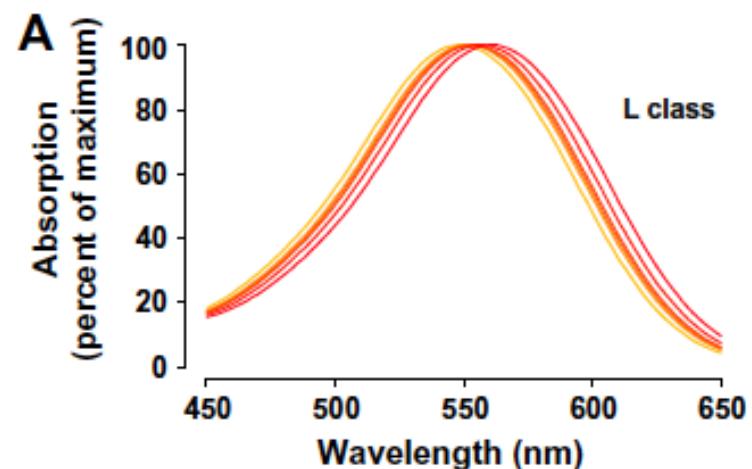
Differences between M and L opsin genes



Differences between M and L opsins genes

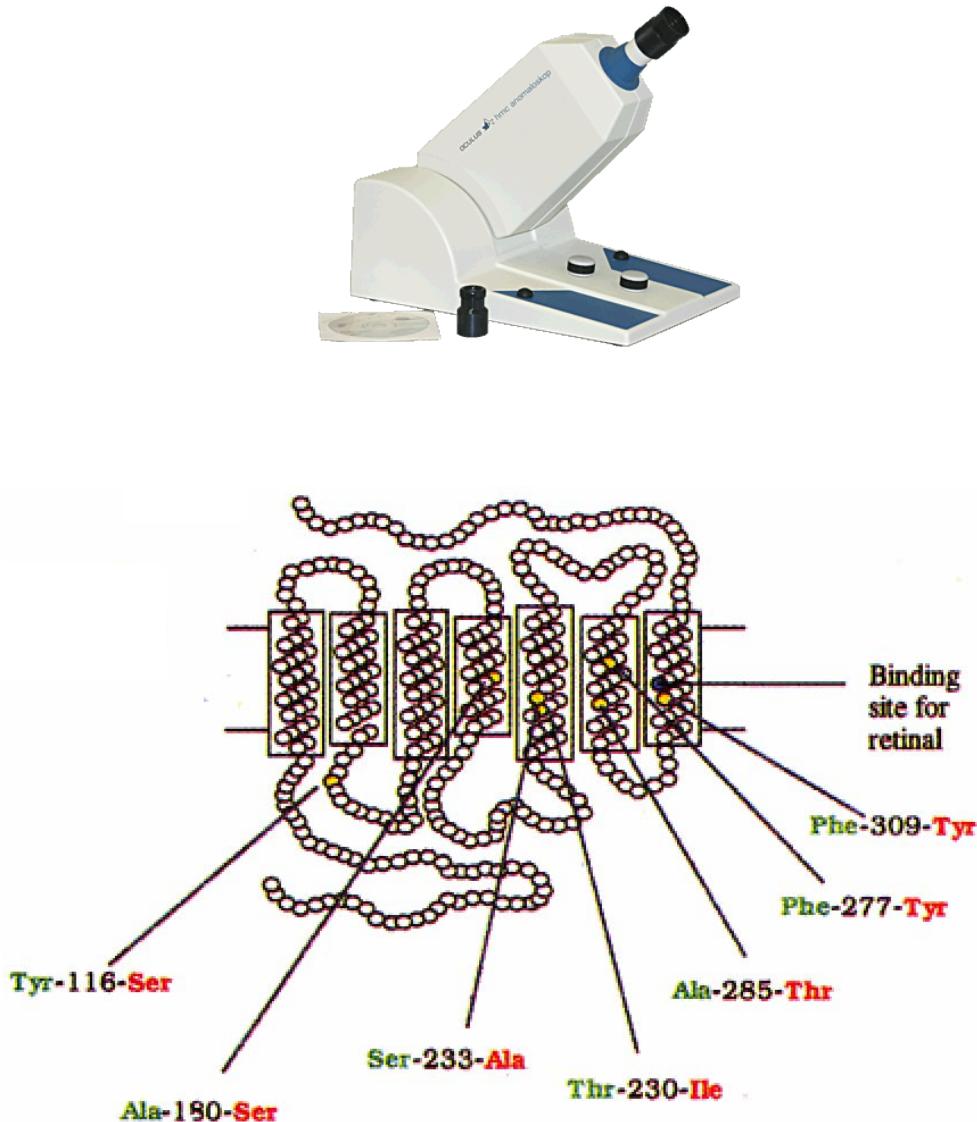


Asemjo et al. (1994)

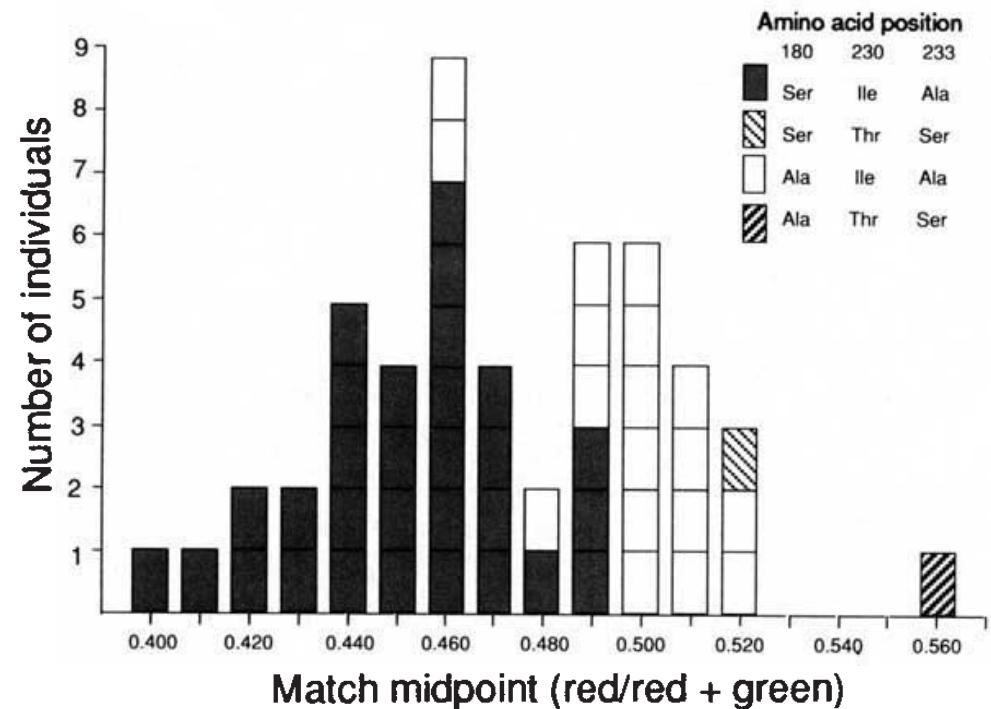


Neitz and Neitz (2011)

Polymorphisms of 'normal' opsins

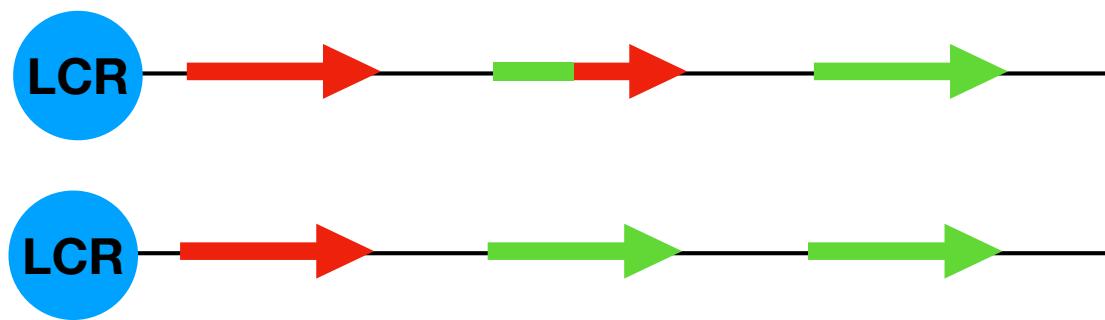
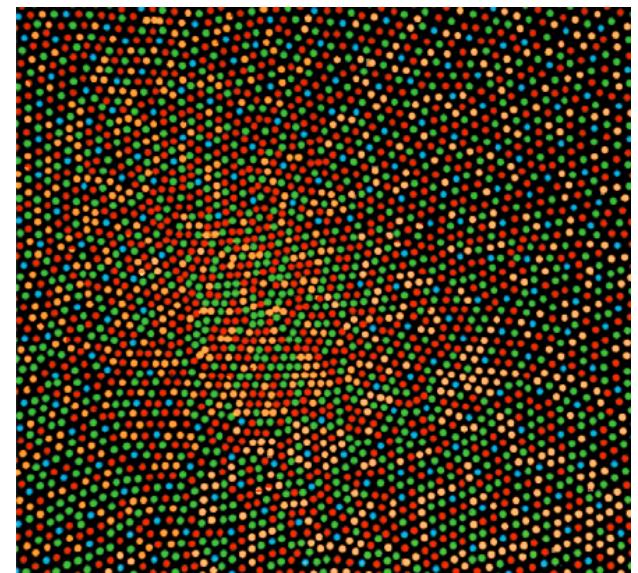
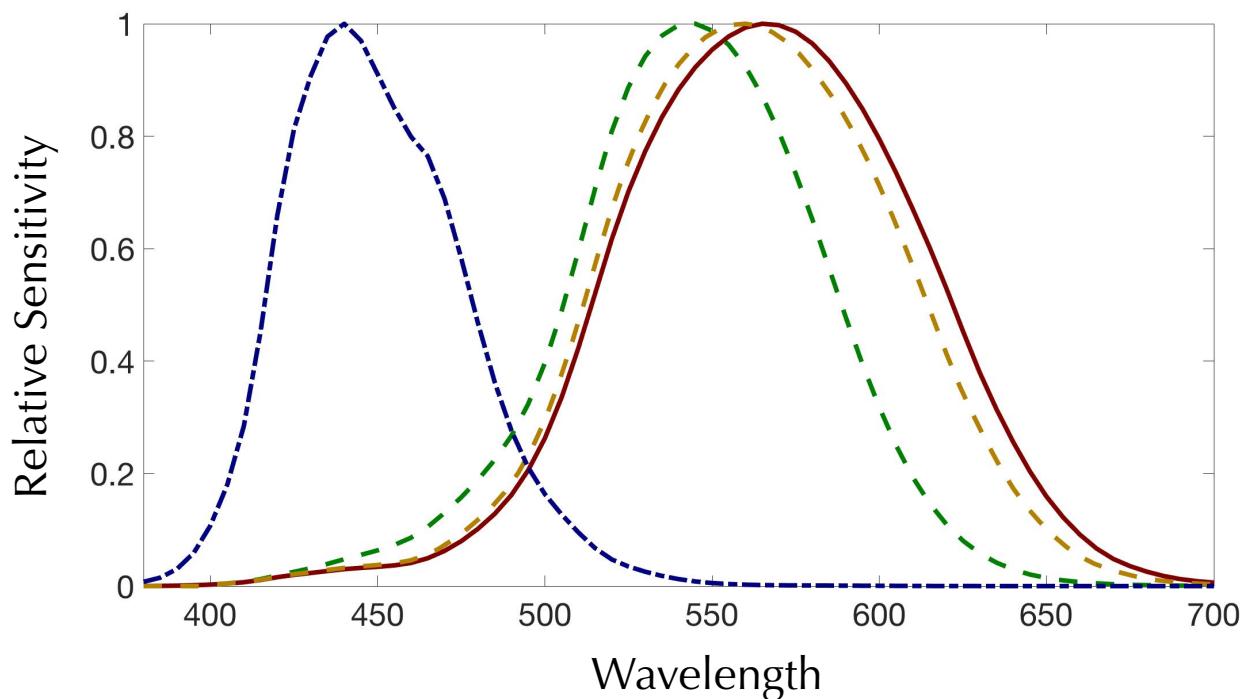


Asemjo et al. (1994)

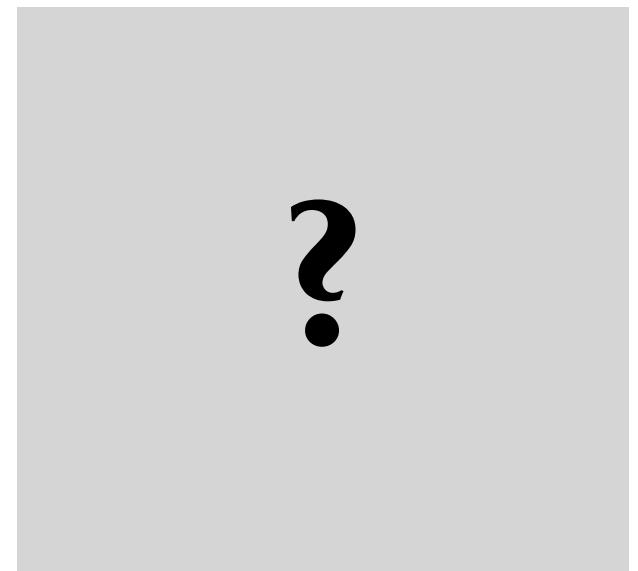


Windericks et al. (1992)

Tetrachromacy



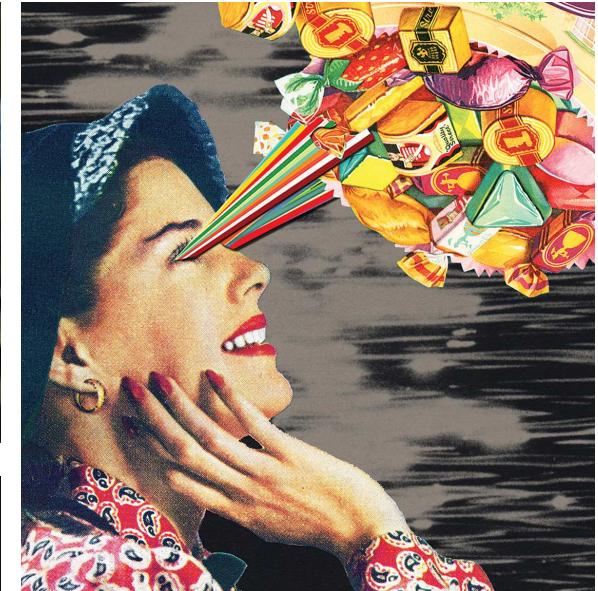
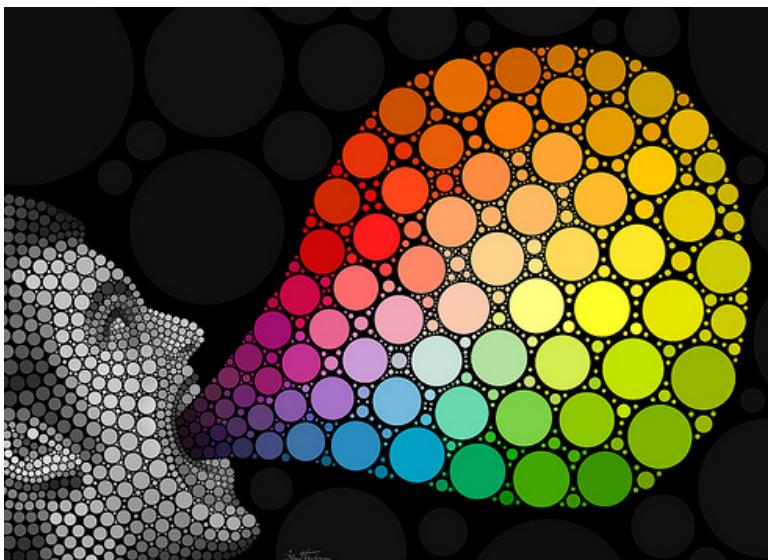
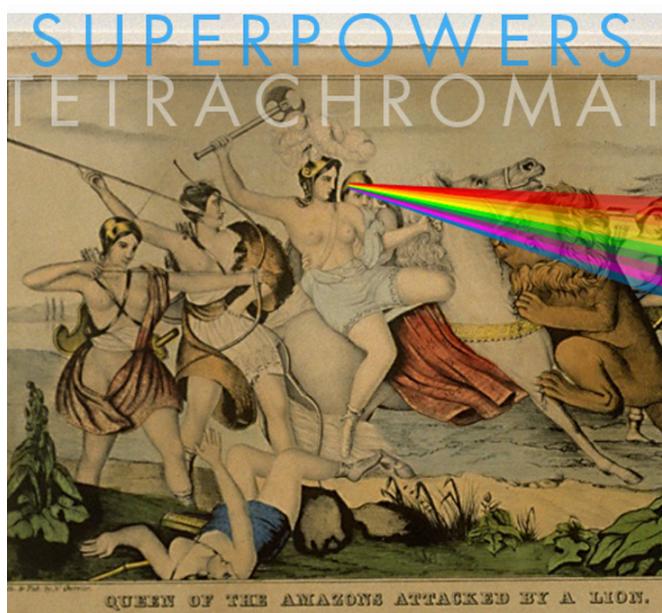
$S/(L+M)$



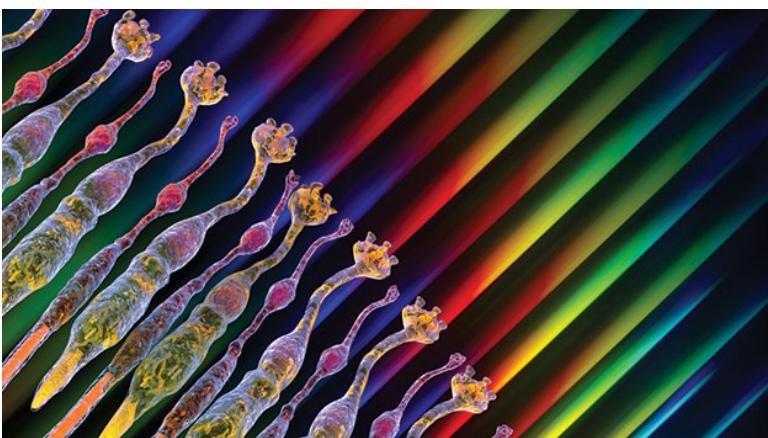
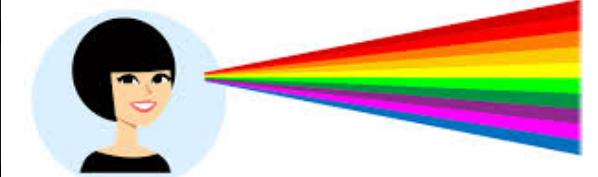
$L/(L+M)$



Superhuman color: **Tetrachromats**

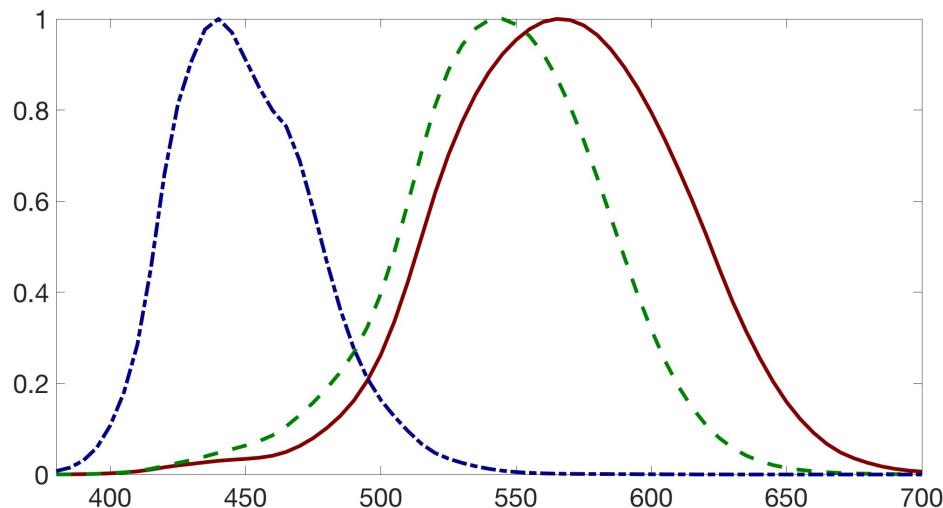


TETRACHROMATS
SEE 10 COLORS IN THE RAINBOW
can differentiate one HUNDRED million colors

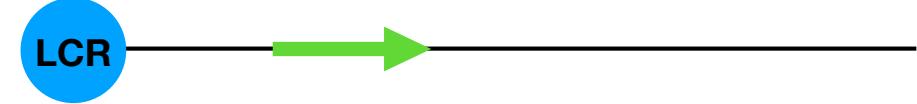
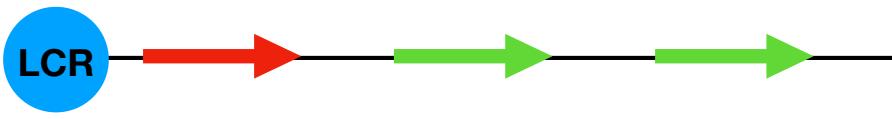
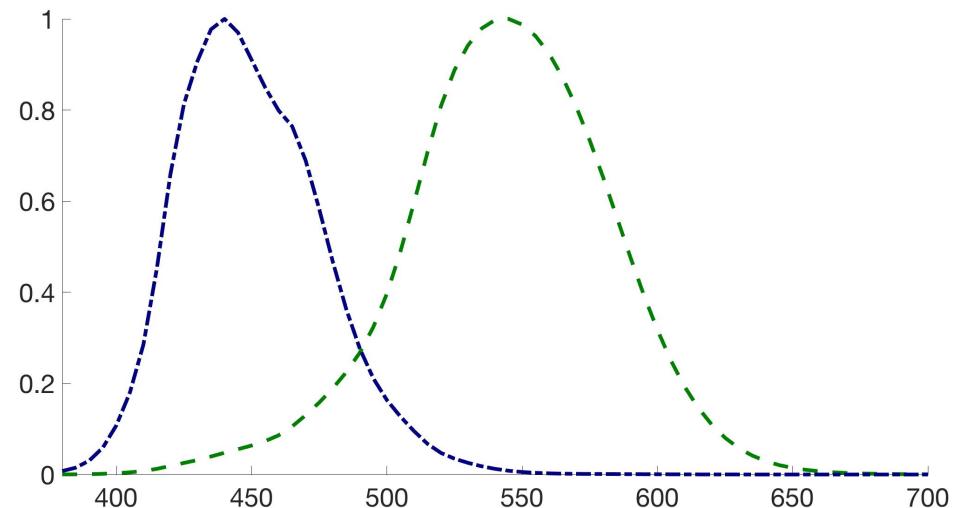


Genetic knock-in makes mice trichromatic

humans

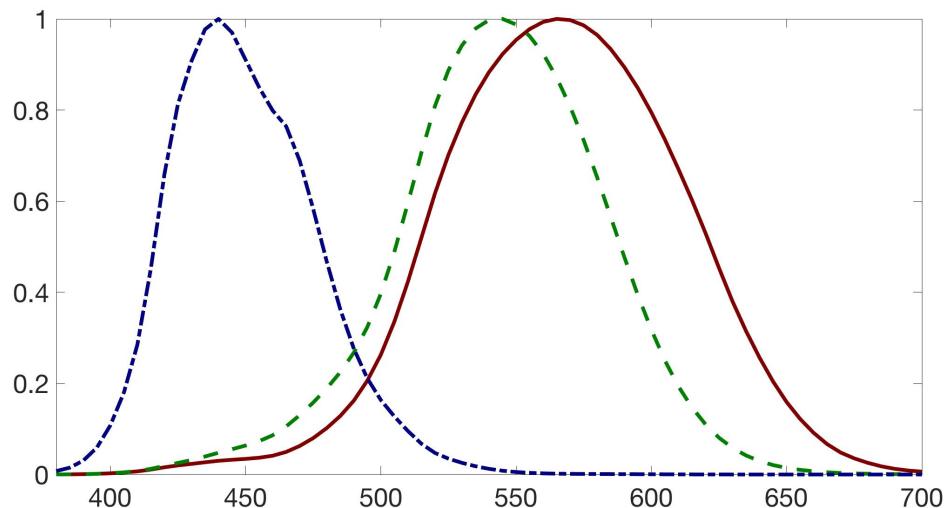


mice

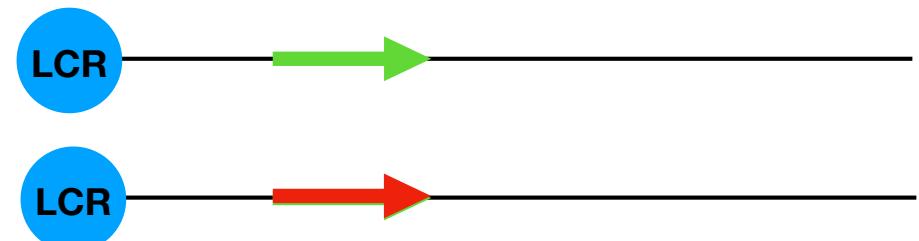
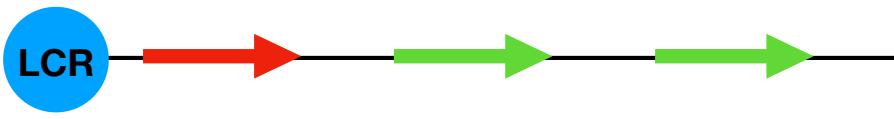
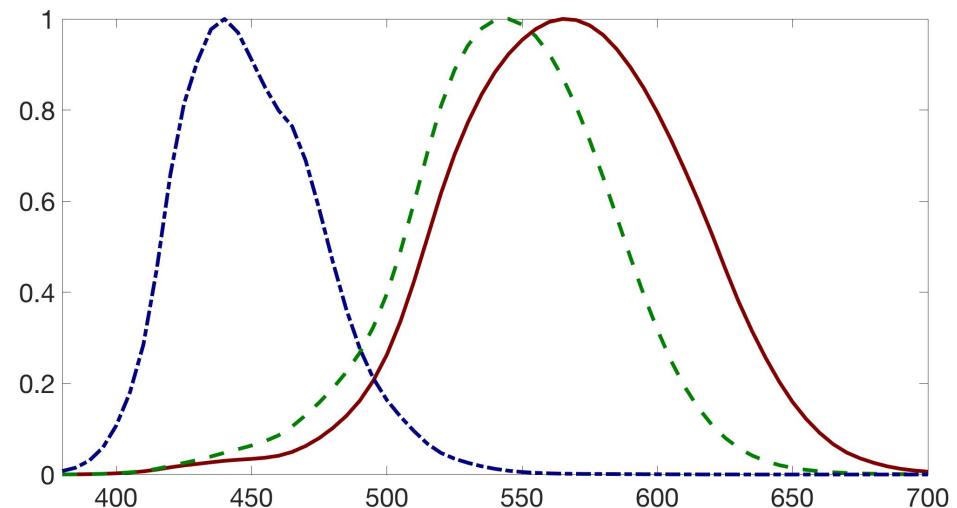


Genetic knock-in makes mice trichromatic

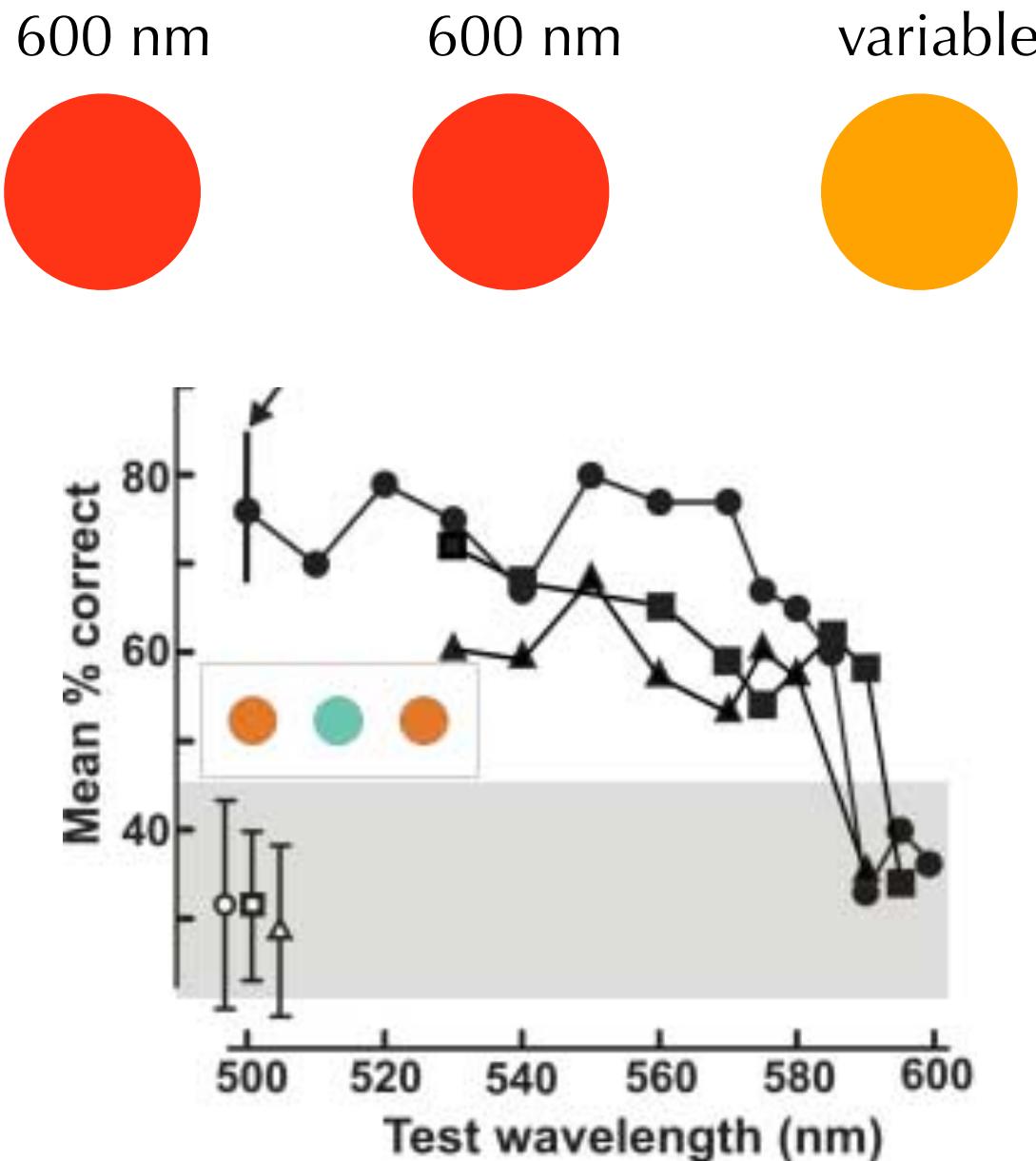
humans



mice



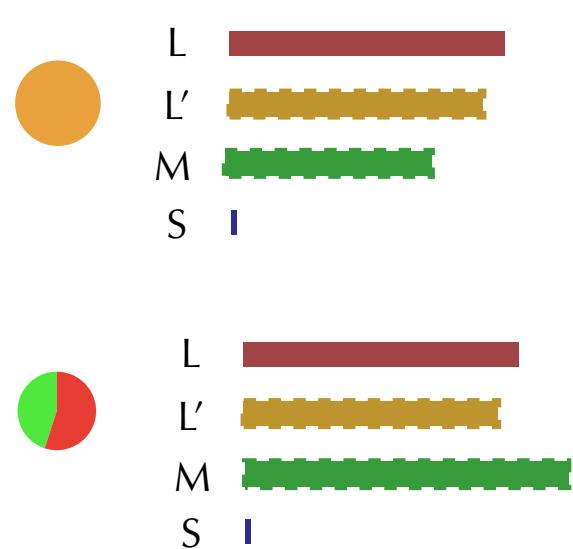
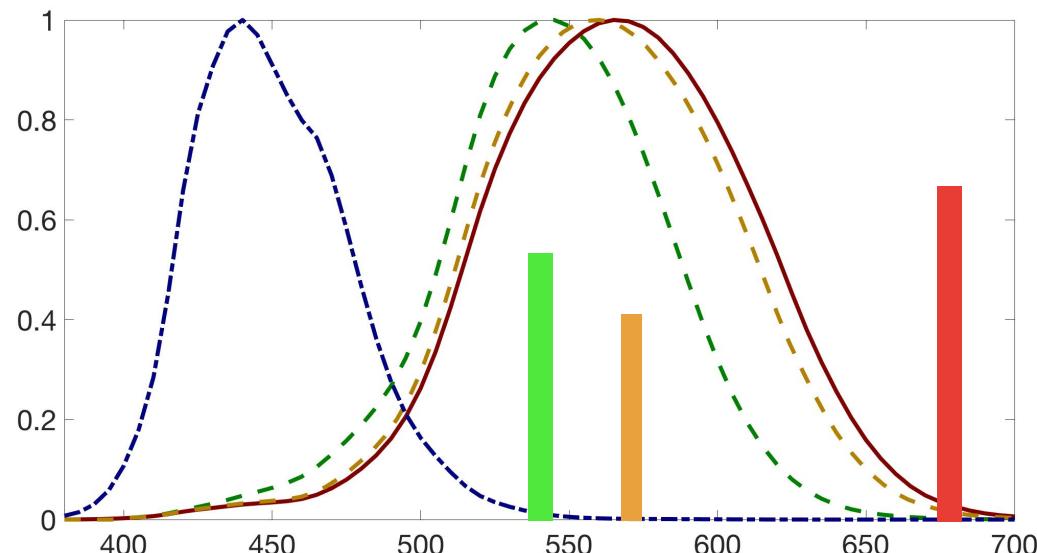
Genetic knock-in makes mice trichromatic



Jacobs et al. (2007). Science.

Tetrachromacy: Rayleigh match rejection

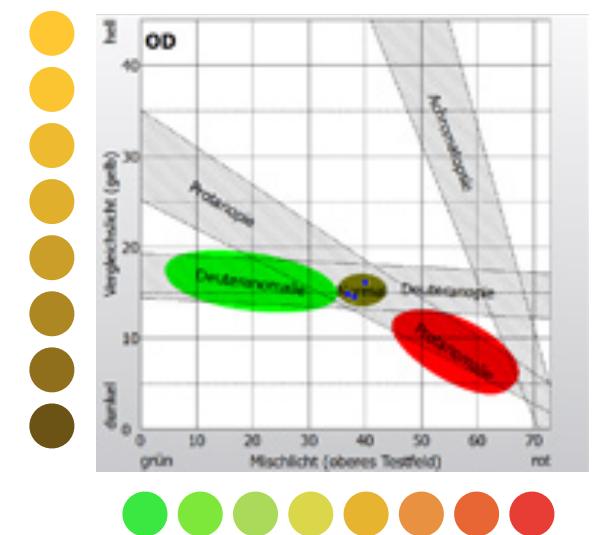
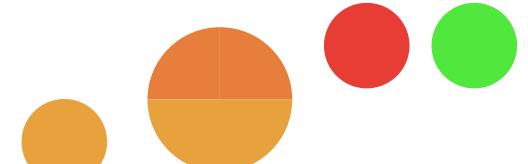
Tetrachromacy



Matching tests

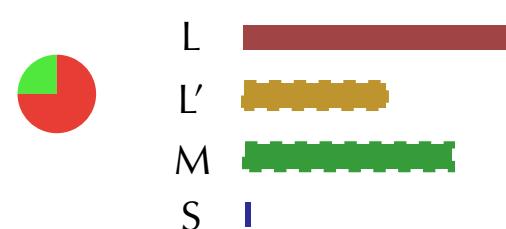
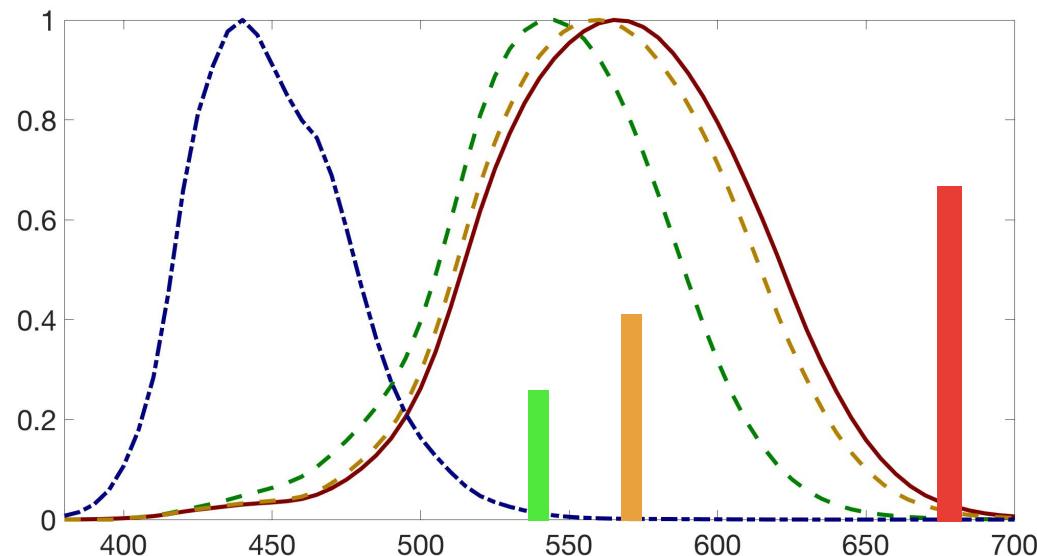


Anomaloscope (Nagel, 1907)



Tetrachromacy: Rayleigh match rejection

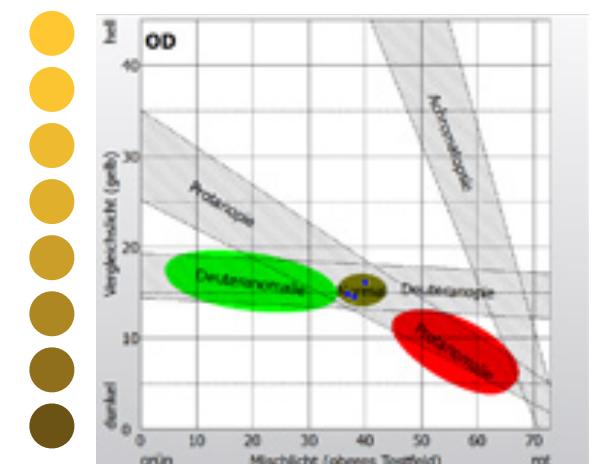
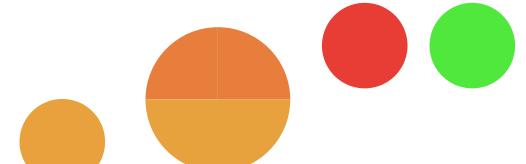
Tetrachromacy



Matching tests



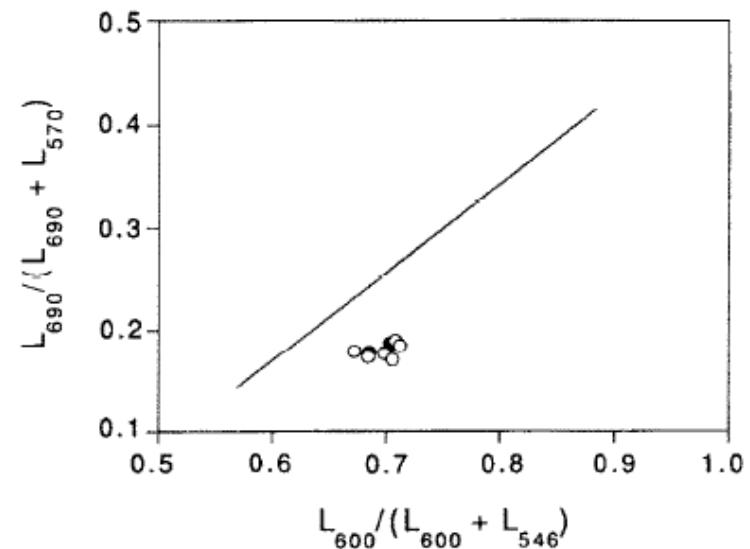
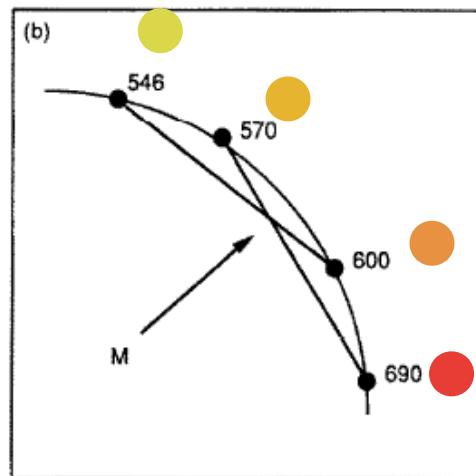
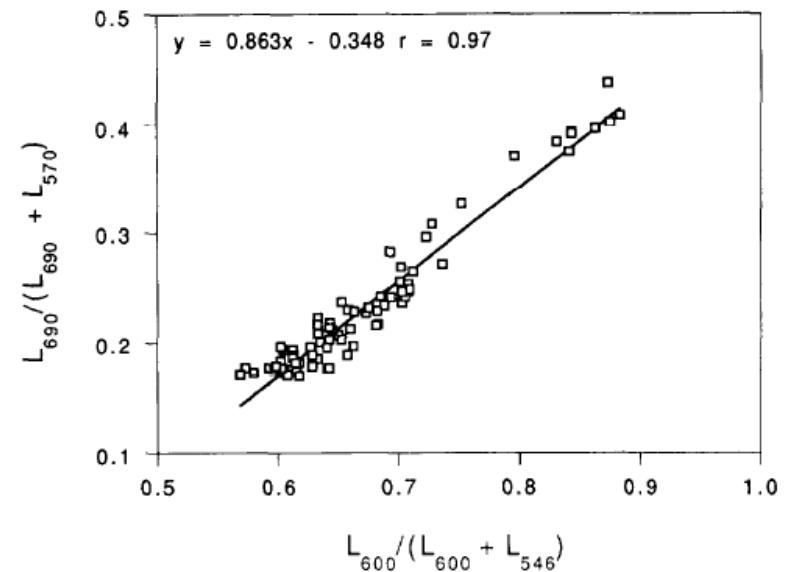
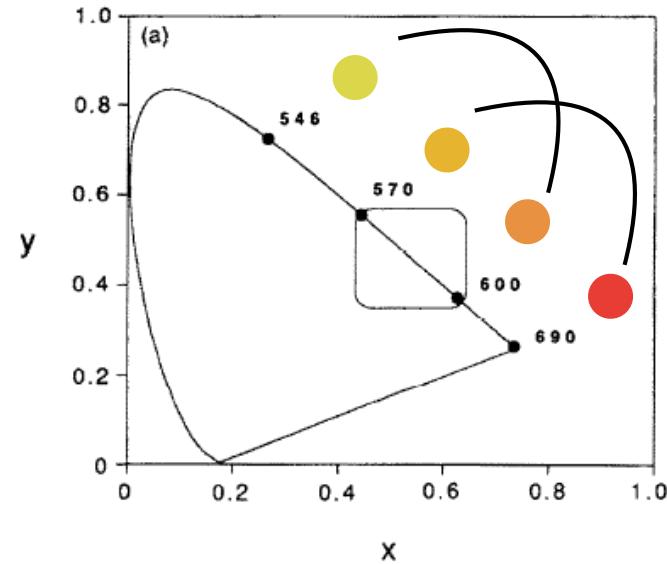
Anomaloscope (Nagel, 1907)



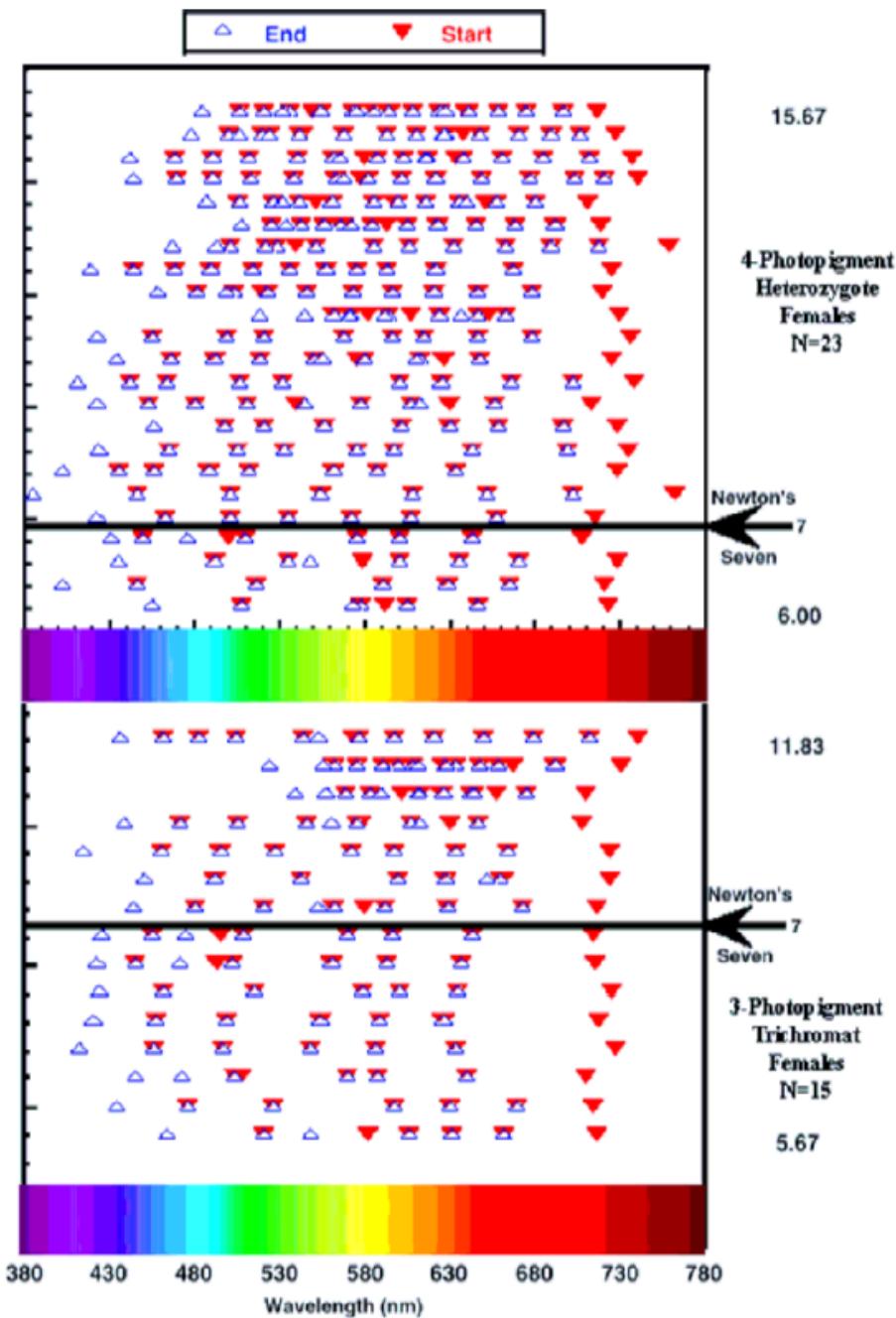
Tetrachromacy

A Study of Women Heterozygous for Colour Deficiencies

G. JORDAN,* J. D. MOLLON*



Tetrachromacy



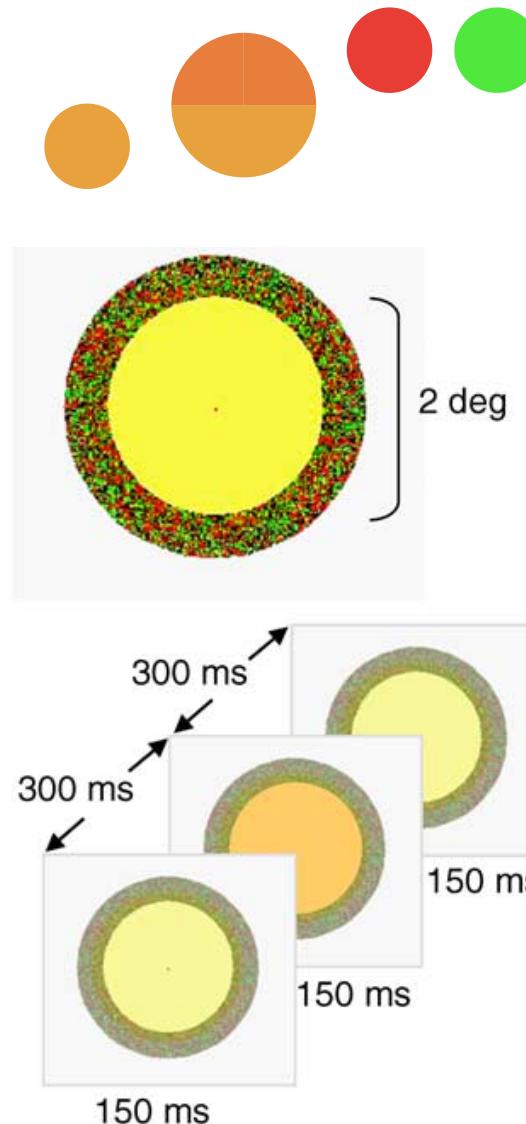
Richer color experience in observers with multiple photopigment opsin genes

KIMBERLY A. JAMESON and SUSAN M. HIGHNOTE
University of California at San Diego, La Jolla, California

and
LINDA M. WASSERMAN
University of California at San Diego School of Medicine, La Jolla, California

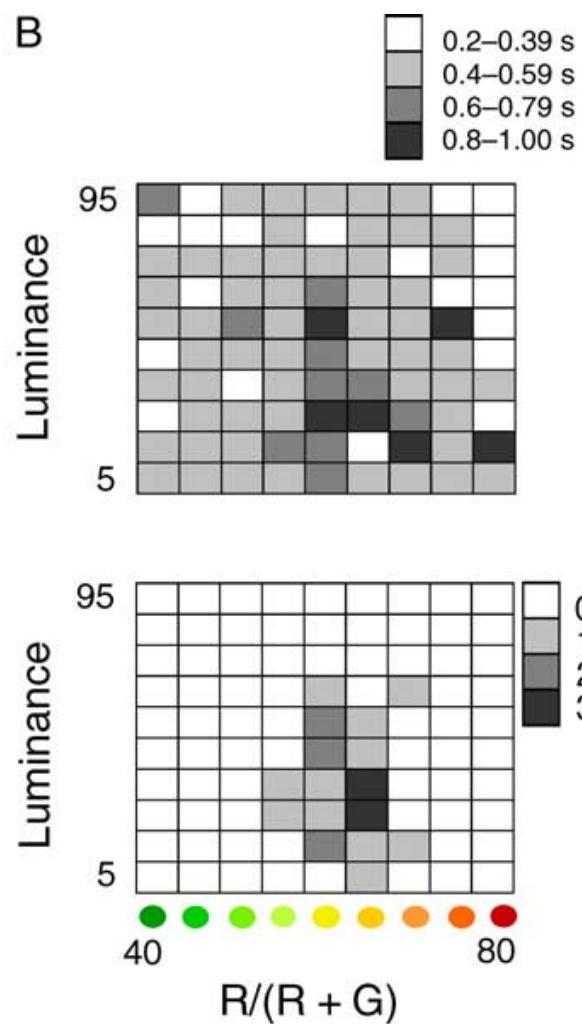
Mean Number of Perceived Spectral Delineations for Four Subject Partitions

Subject Partitions	M	SD	n
(1) Four-pigment females (of heterozygote genotype)	10	2.96	23
(2) Trichromat females	7.6	1.80	15
(3) Trichromats (females and males)	7.3	1.93	37
(4) Dichromat males (protanopes)	5.3	1.53	4

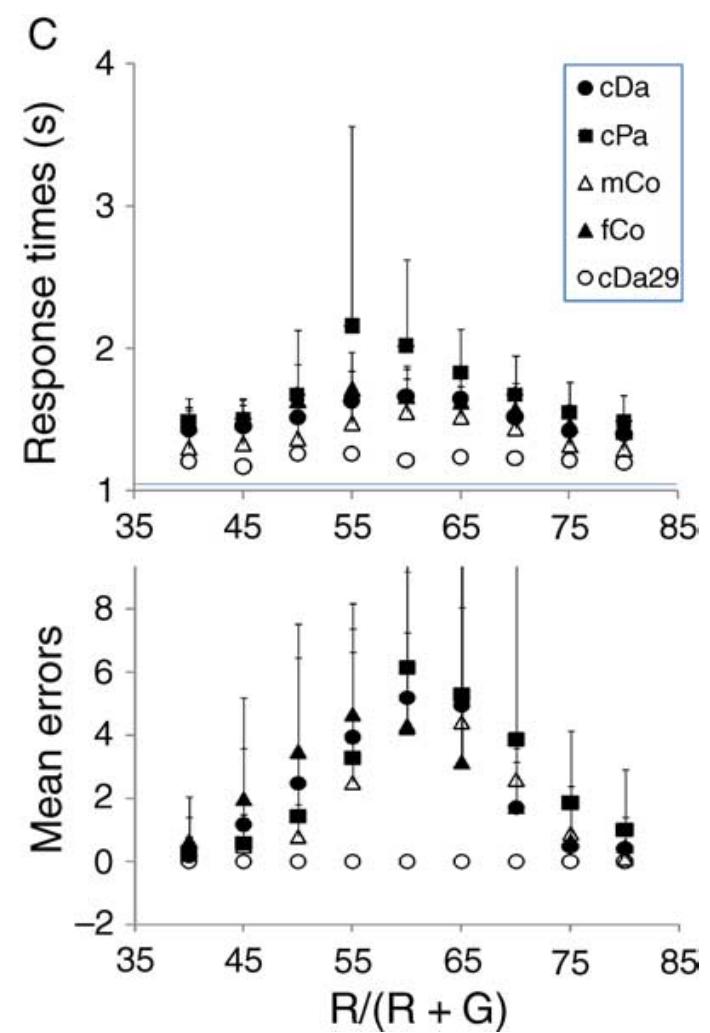


Tetrachromacy

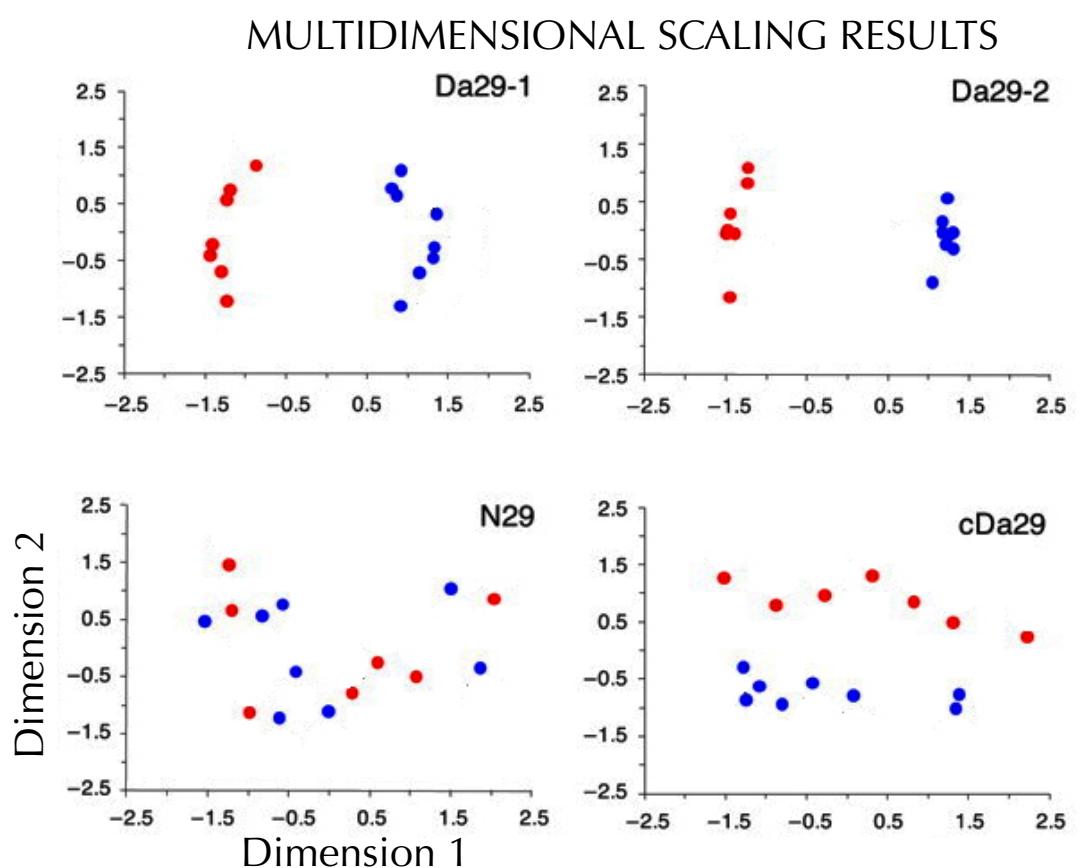
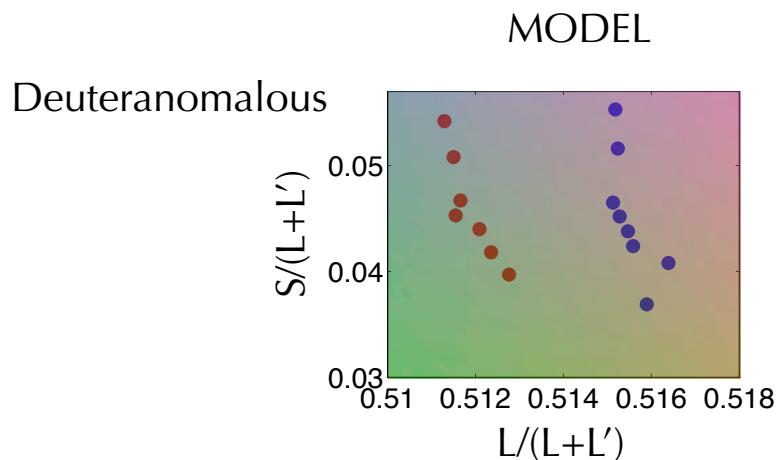
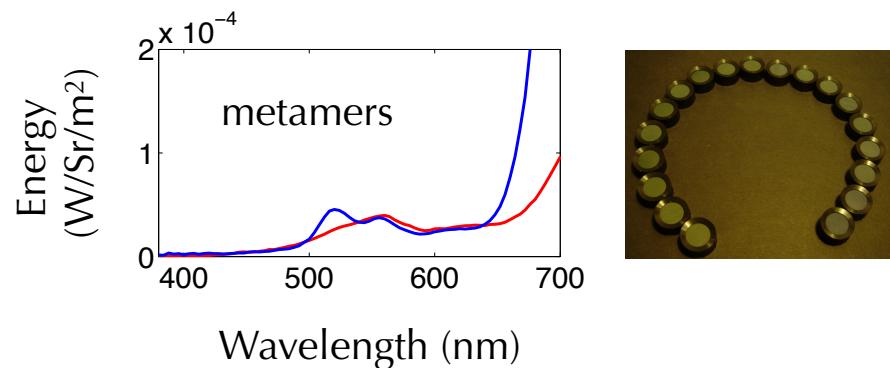
B



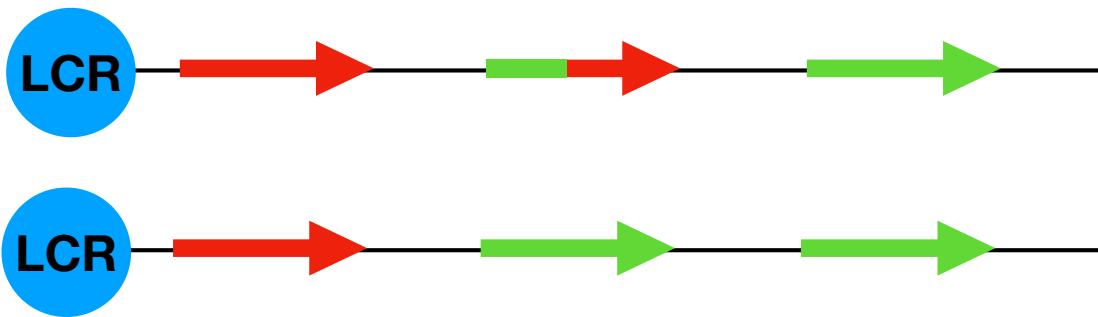
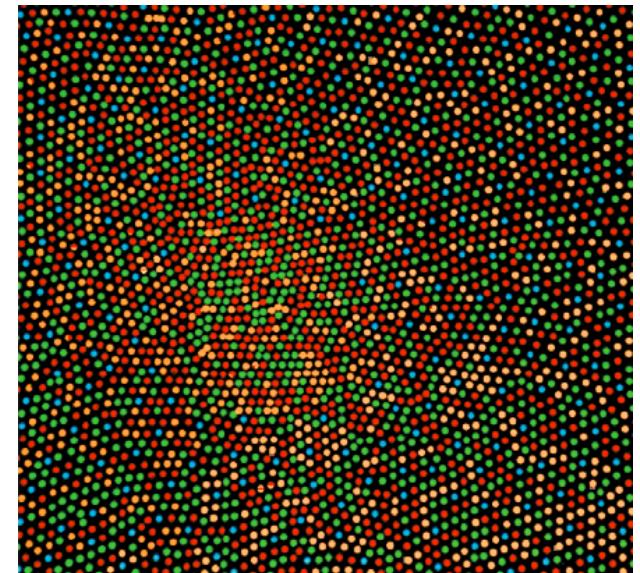
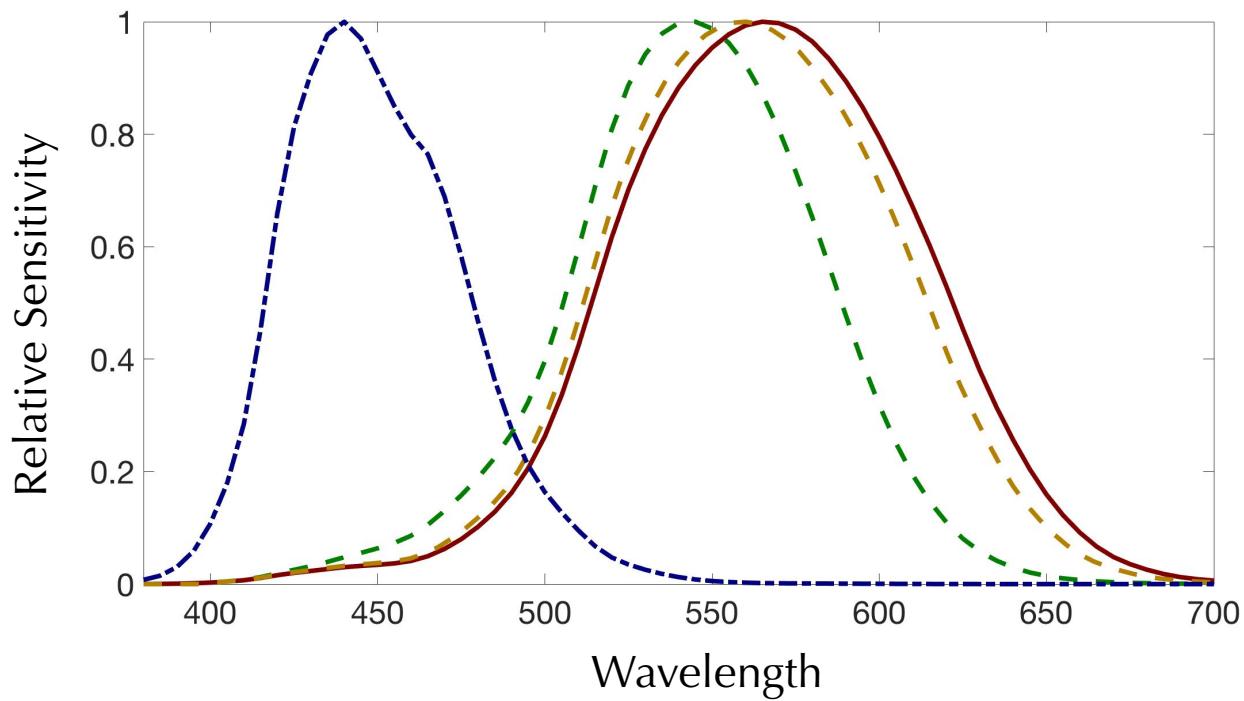
C



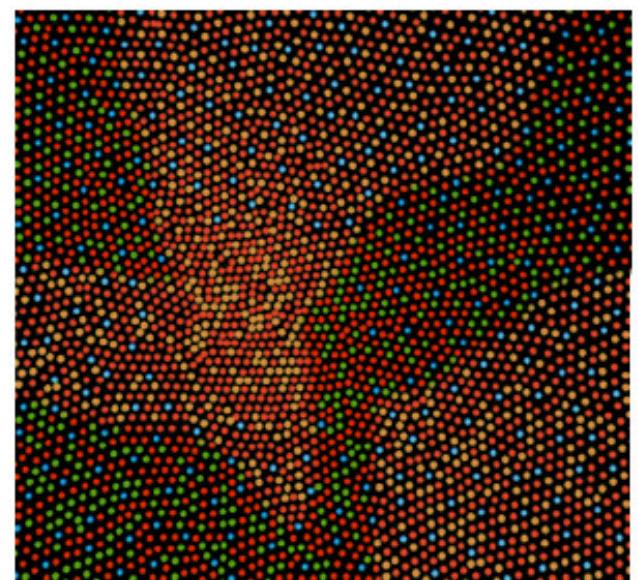
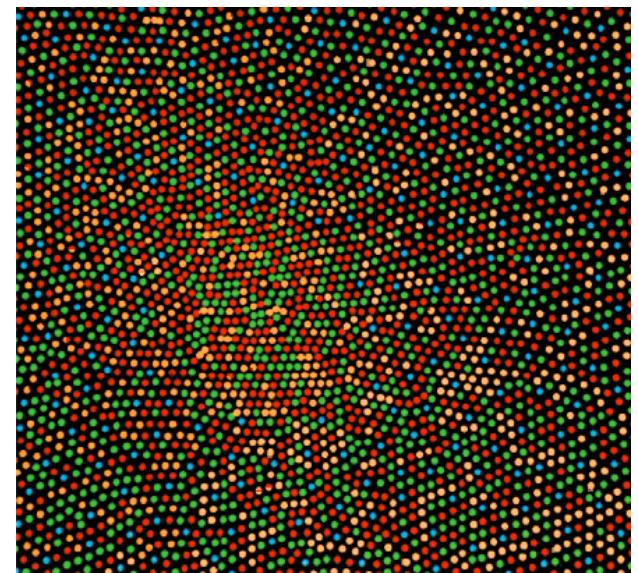
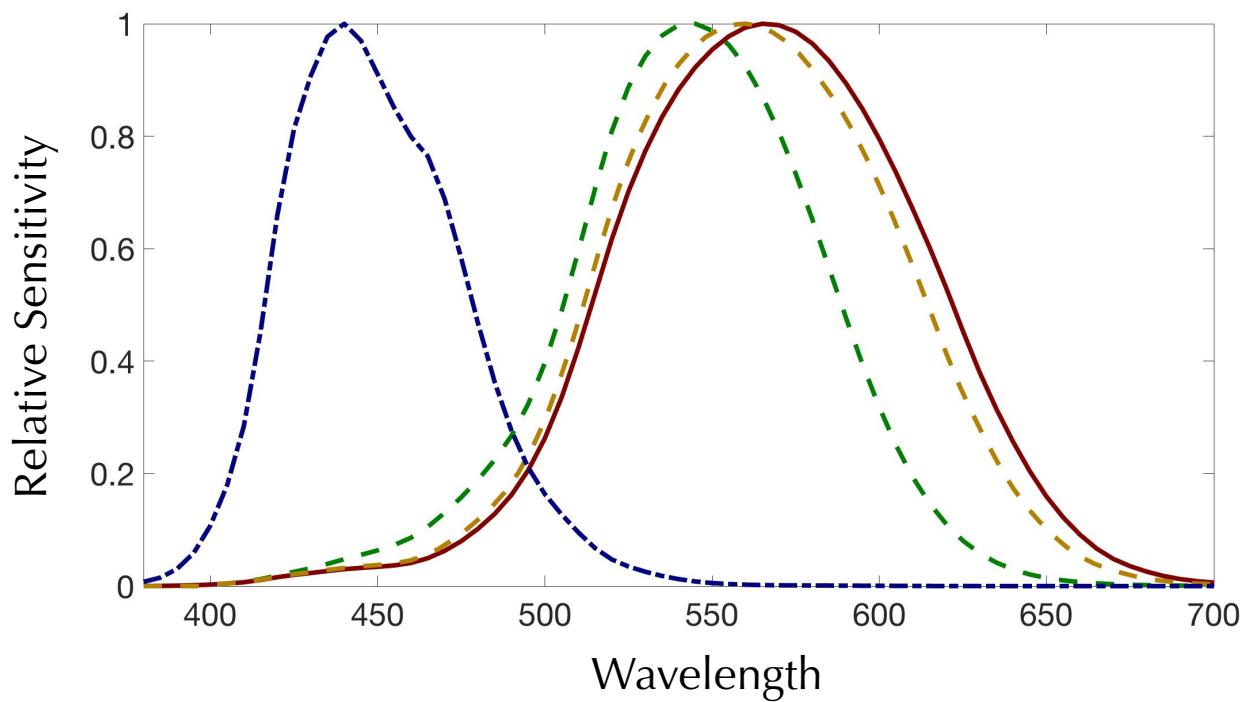
Tetrachromacy



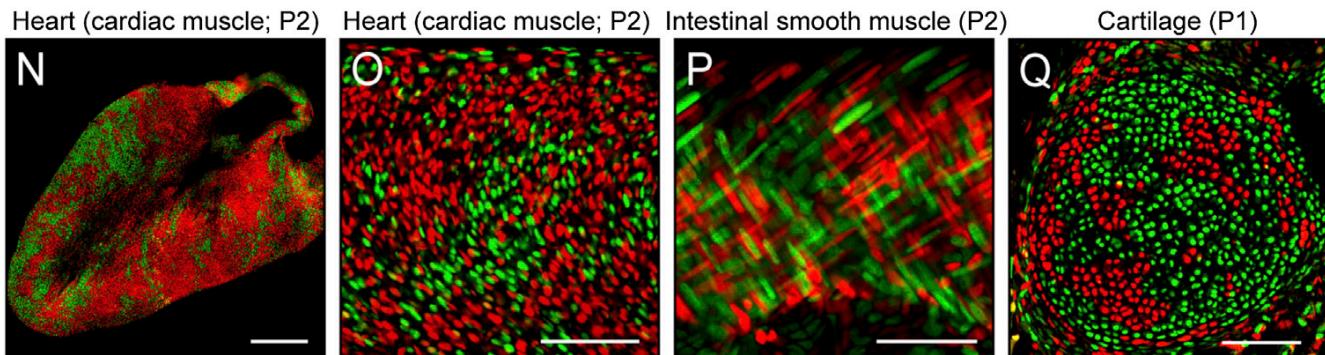
Tetrachromacy



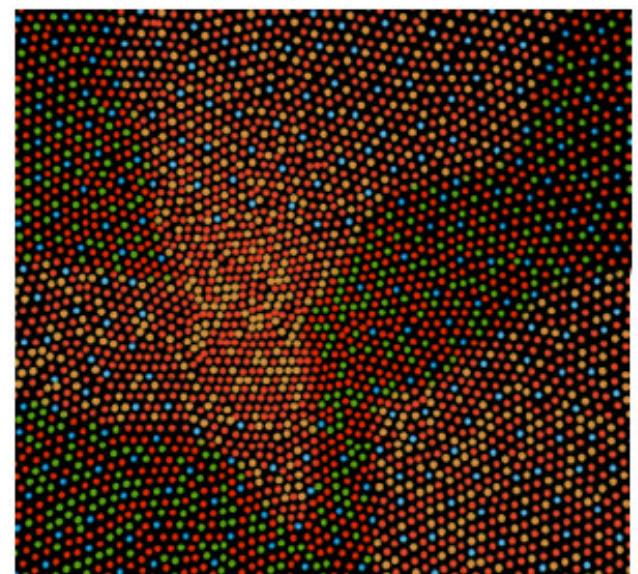
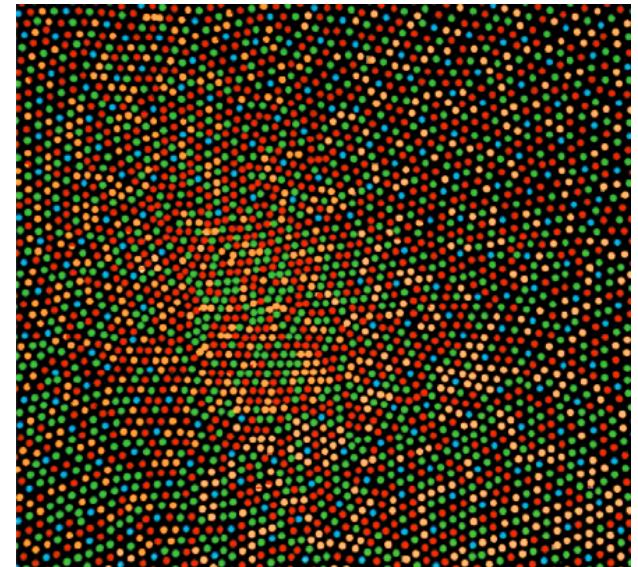
Tetrachromacy



Tetrachromacy

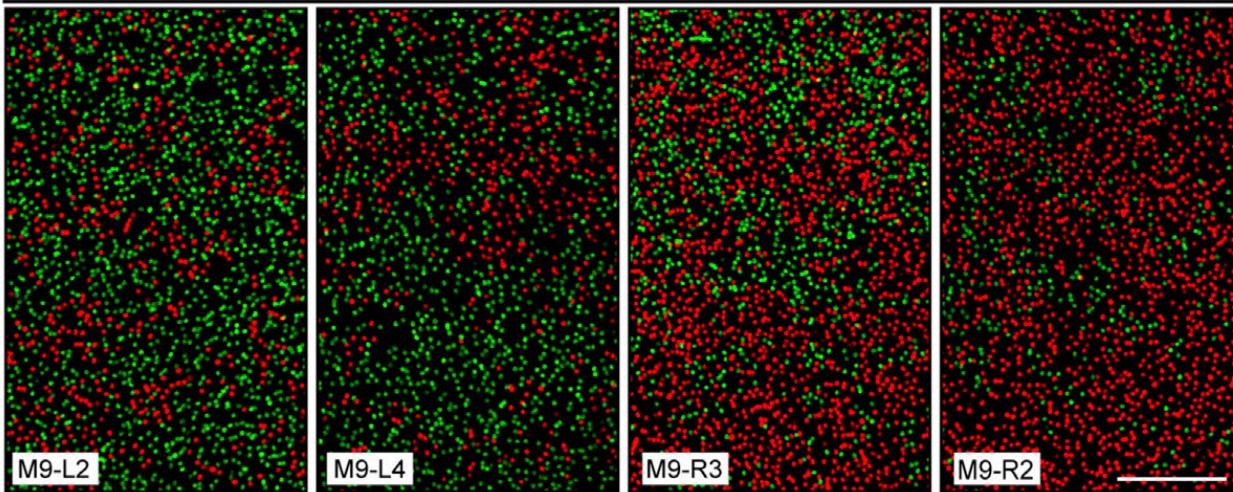


Wu et al. (2014)

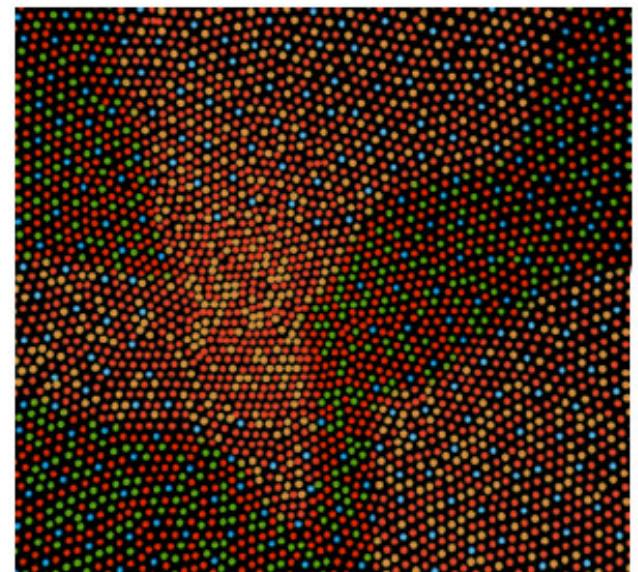
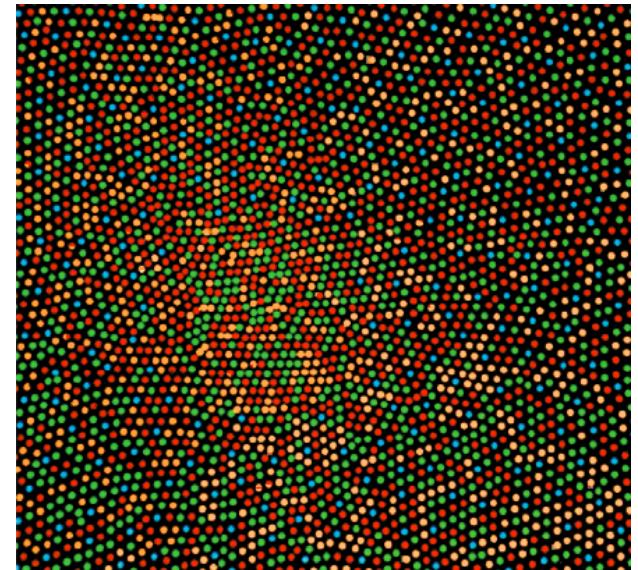


Tetrachromacy

A Cone photoreceptors from a single ~P30 mouse; retina flat mounts (*Cone-Cre;Hprt^{LSL-GFP/LSL-tdT}*)



Wu et al. (2014)



Summary

- Genetic polymorphisms in the L and M opsin gene array lead to a variety of colour vision phenotypes, including measurably different phenotypes even among ‘normal’ trichromats
- The colour vision of individual observers can be modelled using cone sensitivity *nomograms* with adjustments for lens, optical density and macular pigment
- Colour discrimination is predicted to correlate with the spectral separation between the M and L cones, but meta analysis shows the relationship is weak
- Anomalous trichromats may postreceptorially compensate for reduced signals sent from their M and L cones to relatively normalise their colour vision
- Notch filters can enhance perceived colour saturation in anomalous trichromacy, but are most effective for broadband spectra
- The existing evidence is consistent with the existence of strong tetrachromacy in a minority of women, but perhaps alternative explanations have not yet been ruled out

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