



Supplementary Figure S1: ESCS phenotype of proband II-1 in the American family. **A)** ERGs of the proband and a normal subject. The light source was a Grass stimulator recessed in a Ganzfeld. The subjects were studied in the dark- (above) and light-adapted state (below) using four different Wratten filters providing red (Wratten 72B), yellow (Wratten 21Y), green (Wratten 74) and blue (Wratten 98) light. Responses are detected at the cornea by Burian-Allen bipolar contact lens electrodes and amplified by a Nicolet computer system with low noise amplifiers, averaging usually 50-100 responses to the same flash presented every second. The proband has all characteristics of ESCS. In the dark-adapted state, her response to all wavelengths are similar with a more delayed and prolonged response, especially reduced in amplitude to those wavelengths that strongly affect rods, *i.e.* yellow, green and blue. In the light-adapted state, where rod responses are normally eliminated by saturation, her responses to red, yellow and green, which strongly affect long and middle wavelength cones, are reduced in amplitude, but the response to blue, which strongly affects S-cones, is much larger than normal and abnormal in waveform with a more pronounced negative a-wave than is normally seen in the S-cone ERG. The calibration, lower right, indicates 7 μ V vertically and 20 msec horizontally. **B)** Spectral sensitivity curve of the proband's ERG based on constant threshold responses. Eight different spectrally selective Wratten filters were used to determine the spectral sensitivity of the ERG, *i.e.* the reciprocal of energy needed to obtain a threshold response (Gouras et al, 1993, IOVS 34:2437-42). Intensity/amplitude functions for the responses to each wavelength were plotted and then the energy needed to produce an equal and quasi-threshold response for each wavelength determined. By this means the spectral sensitivity (reciprocal of energy on a quantal basis) of the ERG was obtained in the light- and dark-adapted state. The peak sensitivity in both the dark- and light-adapted state is in the blue region of the spectrum, which is an effective wavelength for eliciting

S-cone responses. This is unique to ESCS. C) ERG to white light of proband II-1 in the light- and dark-adapted state. The gain is reduced in the lower trace. The vertical lines indicate the measurement points used to obtain b-wave amplitude. The vertical line at the peak of the b-wave was used to determine peak latency of the b-wave. The light flash occurs at the start of the trace. Each trace is 0.1 seconds in duration.