

Human Gene Therapy

Human cone visual pigment deletions spare sufficient photoreceptors to warrant gene therapy (doi: 10.1089/hum.2013.153)

has been peer-reviewed and accepted for publication, but has yet to undergo copyediting and proof correction. The final published version may differ from

TABLE 1. CLINICAL CHARACTERISTICS OF THE BCM PATIENTS

| Patient/ Family | Age at visits (y) | Visual acuity ^a | Refraction ^b | Nystagmus (in early life) ^c | Light aversion | Color vision (confusion axis) ^d | S-cone function ^e | ERG ^f amplitude Rod b- wave | Cone flicker |
|--------------------|-------------------|----------------------------|-------------------------|---|-------------------|---|---------------------------------|--|-----------------|
| P1/F1 | 5,8 | 20/100 | -12.50 | + (2-3 mos) | + | protan-deutan | + | ← np → | ← np → |
| P2/F1 | 8,10 | 20/80 | -3.75 | + (2-3 mos) | + | protan-deutan | + | ← np → | ← np → |
| P3/F1 | 11,13 | 20/63 | -12.75 | + (2-3 mos) | + | protan-deutan | + | ← np → | ← np → |
| P4/F1 | 48,50 | 20/100 | -10.25 | + (infancy) | + (disabling) | protan-deutan | + | N | A |
| P5/F2 | 7 | 20/100 | -3.25 | + (3-4 mos), †(12 mos), ‡(2.5 yr) | + | protan-deutan | + | N(b) | A |
| P6/F2 | 12 | 20/100 | -5.00 | + (9 mos), †(18 mos), ‡(4 yr) | + (not prominent) | protan-deutan | + | ← np → | ← np → |
| P7/F3 | 5,7 | 20/250 | -0.50 | + | no complaints | all three axes | + | N | A |
| P8/F3 | 10,19 | 20/100 | -5.00 | + (4 mos) | no complaints | protan-deutan | + | ← np → | ← np → |
| P9/F3 | 16,25 | 20/100 | -6.00 | + (6 mos) | + (disabling) | protan-deutan | np | N | A |
| P10/F3 | 19,28 | 20/125 | -6.50 | + (10 mos) | + | protan-deutan | + | N | A |
| P11/F4 | 7 | 20/80 | +2.00 | + | + | protan-deutan | np | N | A |
| P12/F4 | 41 | 20/160 | -13.00 | + | + | np | np | ← np → | ← np → |
| P13/F4 | 58 | 20/200 | -10.00 | + | + | np | np | N | A |
| P14/F5 | 11 | 20/100 | -7.50 | + (infancy) | + | protan-deutan | + | N(b) | A |
| P15/F6 | 14 | 20/100 | -4.00 | + (3 mos) | no complaints | protan-deutan | + | N | A |
| P16/F7 | 28 | 20/125 | -8.50 | + (<1 yr), †(later in life) | + (disabling) | protan-deutan | + | N | A |
| P17/F8 | 33 | 20/100 | -3.75 | + | + (disabling) | protan-deutan | + | ← np → | ← np → |
| P18/F9 | 35 | 20/63 | -5.50 | + | + (disabling) | protan-deutan | + | N(b) | A |
| P19/F10 | 43 | 20/80 | -6.00 | + (minimal) | glare | deutan | + | N | A |
| P20/F11 | 55 | 20/80 | -6.50 | + | + (disabling) | all three axes | + | ← np → | ← np → |

np, not performed; +, present.

^aBest corrected visual acuity at most recent visit; similar in the two eyes.

^bSpherical equivalent; average of both eyes.

^cAges when nystagmus was first noted or when changed; amplifying the comment 'in early life' when possible. The ages cited are from historical accounts by patients or parents (mos, months; yr, years). Arrows indicate whether there was recalled reduction or increase (and at what age).

^dFarnsworth D-15 color panel result.

^eDetection of 440 nm target on 100 phot.cd/m² yellow background, or detectable S-cone ERG.

^fElectroretinography was performed in different laboratories with different electrode types, protocols and normal limits. N, within normal limits of the laboratory; N(b), borderline normal - reports indicated blink or nystagmus artifact, imperfect cooperation, head not completely within Ganzfeld stimulus or other possible sources of reduced amplitude. A, abnormal.