Table 1. Demographic data and ocular findings of patients with congenital insensitivity to pain

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **Patient** | **Age** | **Sex** | **Consanguinity** | **Mutation** | **FU, months** | **Eye** | **Visual acuitya** | | **CO** | **CS** | **Refraction** | **SPK Gradeb**  **A D** | | **TBUT** | **Schirmer** | **Corneal abscess** | **Other** |
| **family 1** | **1** | **11** | **F** | **+** | ***PRDM12*** | **94** | **RIGHT** | **6/7.5** | **6/9** | **-** | **-** | **+2.25-3.50X175** | **3** | **3** | **NA** | **25** |  | **LT, PO** |
|  |  |  |  |  |  | **LEFT** | **6/21** | **6/9** | **+** | **-** | **+3.50-3.25X40** | **3** | **3** | **NA** | **30** | **2,10 yo** | **LT, PO** |
| **2** | **3** | **M** | **+** | ***PRDM12*** | **23** | **RIGHT** | **NF** | **LP** | **+** | **-** | **NA** | **3** | **3** | **NA** | **NA** | **1 yo** | **6AM+7LT, PO** |
|  |  |  |  |  |  | **LEFT** | **NF** | **NA** | **+** | **-** | **NA** | **3** | **3** | **NA** | **NA** | **1 yo** | **5AM+8LT, 2CCG, PO**  **ET, trichiasis** |
|  | **3** | **6.06** | **M** | **NA** | ***PRDM12*** | **53** | **RIGHT** | **CSM** | **6/60** | **+** | **NA** | **NA** | **NA** | **NA** | **NA** | **NA** | **1 yo** | **LT** |
|  |  |  |  |  |  |  | **LEFT** | **CSM** | **6/12** | **+** | **NA** | **+3.25-2.00X15** | **NA** | **NA** | **NA** | **NA** | **2 yo** | **LT** |
| **family 2** | **4** | **24.05** | **F** | **+** | ***SCN9A*** | **143** | **RIGHT** | **NF** | **6/60** | **+** | **+** | **-0.75-4.50X145** | **2** | **3** | **7** | **3** | **1 yo, 3LT** | **Amblyopia** |
|  |  |  |  |  |  | **LEFT** | **NF** | **6/9** | **-** | **+** | **+1.00-0.50X14** | **1** | **3** | **7** | **7** |  |  |
| **5** | **16.01** | **F** | **+** | ***SCN9A*** | **128** | **RIGHT** | **6/9** | **6/9** | **-** | **-** | **PLANO** | **0** | **0** | **3** | **<1** |  |  |
|  |  |  |  |  |  | **LEFT** | **6/9** | **6/9** | **-** | **-** | **PLANO** | **0** | **0** | **7** | **<1** |  |  |
| **6** | **13.06** | **F** | **+** | ***SCN9A*** | **121** | **RIGHT** | **6/18** | **6/24** | **+** | **+** | **+5.25-5.00 X40** | **1** | **3** | **2** | **<1** | **1 yo, LT,** |  |
|  |  |  |  |  |  | **LEFT** | **6/12** | **6/9** | **-** | **+** | **+4.25-1.00X179** | **1** | **3** | **2** | **<1** |  |  |

FU, follow up; CO, corneal opacity; CS, corneal sensitivity; CT, cotton thread; SPK, superficial punctate keratopathy; TBUT (seconds), tear break up time; M, male; F, female; mm, millimeters; yo, years old; LT, lateral tarsorrhaphy; AM, amniotic membrane; CCG, corneal covering graft; PO, punctal occlusion; ET, esotropia; NA, not applicable; NF, not found; CSM, central, steady, maintained; LP, light perception.

aBest corrected visual acuity at first visit and last visit.

bArea (A) ranges from A0, no staining, to A3, diffuse staining. Density (D) ranges from D0, no staining, to D3, lesions are dense and coalescent.