**Supplementary Table 1. Primary and associated clinical manifestations at diagnosis in the LDG**

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| --- | --- | --- | --- | --- | --- |
| Patient No | Age at diagnosis  (years) | Primary condition leading to diagnosis of HCU | | Associated presentations | |
| 1 | 5 | Unilateral lens subluxation | | Learning difficulties | |
| 2 | 10 | Bilateral lens subluxation | | Intellectual disability and learning difficulties | |
| 3 | 9 | Bilateral lens subluxation | | Learning difficulties and abnormal behaviors | |
| 4 | 10 | Bilateral lens subluxation | | Learning difficulties and abnormal behaviors | |
| 5 | 6 | Right lens subluxation (right) | | Learning difficulties and abnormal behaviors | |
| 6 | 6 | Bilateral lens subluxation and poor vision | | Learning difficulties | |
| 7 | 10 | Bilateral lens subluxation | | Learning difficulties and abnormal behaviors | |
| 8 | 6 | Bilateral lens subluxation | | None | |
| 9 | 10 | Bilateral lens subluxation | | Intellectual disability and learning difficulties | |
| 10 | 7 | Bilateral lens subluxation | | Learning difficulties | |
| 11 | 3.5 | Bilateral lens subluxation | | Developmental delay | |
| 12 | 4 | Left eye dislocation | | Developmental delay | |
| 13 | 6 | Left eye dislocation and pupillary block glaucoma (unilateral) | | Mild intellectual disability and abnormal speech | |
| 14 | 8 | Bilateral lens dislocation | | Learning difficulties | |
| 15 | 7 | Bilateral lens dislocation | | None | |
| 16 | 11 | Bilateral lens dislocation | | Mild learning difficulties | |
| 17 | 9 | Bilateral lens dislocation | | Hx of developmental delay and learning difficulties | |
| 18 | 10 | Bilateral lens dislocation | | Mild intellectual disability and learning difficulties | |
| 19 | 14 | Bilateral lens dislocation | | Intellectual disability, learning difficulties, Hx of abnormal behaviors | |
| 20 | 6 | Bilateral Lens subluxation | | Intellectual disability, learning difficulties | |
| 21 | 10 | Left surgical aphakia due to secondary glaucoma | | Mild intellectual and learning difficulties and abnormal behaviors | |
| 22 | 6.5 | Bilateral lens subluxation | | Intellectual disability and Hx of developmental delay | |
| 23 | 13 | Bilateral lens subluxation | | None | |
| 24 | 5 | Bilateral lens subluxation | | Learning difficulties | |
| 25 | 6 | Bilateral Lens subluxation | | Learning difficulties | |
| 26 | 8 | Right lens dislocation with secondary glaucoma | | Intellectual disability | |
| 27 | 7 | Bilateral high myopia | | Learning difficulties | |
| 28 | 7 | Bilateral lens subluxation | | None | |
| 29 | 10 | Bilateral lens dislocation | | Mild intellectual disability | |
| 30 | 7 | Bilateral lens dislocation | | Learning difficulties | |
| 31 | 9 | Bilateral lens subluxation with early cataract changes | | Hx of developmental delay with nocturnal enuresis | |
| 32 | 6 | Bilateral lens subluxation | | Hx of genu valgum at 4 yrs | |
| 33 | 8 | Bilateral lens subluxation | | None | |
| 34 | 12 | Bilateral lens subluxation | | Learning difficulties | |
| 35 | 6.5 | Developmental delay, intellectual disability, learning difficulties, Abnormal behaviors | | Skeletal deformity | |
| 36 | 12 | Intellectual disability | | Myopia | |
| 37 | 17 | Intellectual disability | | Bilateral lens dislocation | |
| 38 | 3 | Intellectual disability and developmental delay | | None | |
| 39 | 2 | Developmental delay in all milestones | | None | |
| 40 | 4 | Intellectual disability, speech delay, Hx of developmental delay | | None | |
| 41 | 7 | Intellectual disability | | Bilateral lens dislocation | |
| 42 | 7 | Intellectual disability, learning difficulties and abnormal behaviors | | Bilateral lens dislocation | |
| 43 | 7 | Intellectual disability, learning difficulties and abnormal behaviors | | Bilateral lens dislocation | |
| 44 | 2 | Developmental delay and abnormal behaviors | | None | |
| 45 | 5 | Mild intellectual disability, Hx of developmental delay | | Bilateral lens dislocation | |
| 46 | 11 | Developmental delay, intellectual disability, learning difficulties, | | Bilateral lens dislocation | |
| 47 | 8 | Intellectual disability and abnormal walking | | Left eye dislocation | |
| 48 | 6 | Intellectual disability and Hx of developmental delay | | None | |
| 49 | 6 | Learning difficulties, enuresis | | Skeletal changes | |
| 50 | 10 | Severe intellectual disability | | Bilateral lens dislocation | |
| 51 | 6 | Learning difficulties | | Bilateral lens subluxation | |
| 52 | 11 | Learning difficulties | | Bilateral lens subluxation | |
| 53 | 8 | Learning difficulties, poor school performance | | Myopia | |
| 54 | 3 | Developmental delay in milestones | | None | |
| 55 | 7 | Intellectual disability with Hx of DD and nocturnal enuresis | | Abnormal behavior and skeletal changes | |
| 56 | 6 | Learning difficulties and poor school performance | | Myopia | |
| 57 | 3 | Mild intellectual disability, Hx of developmental delay | | None | |
| 58 | 3 | Developmental delay in all milestones | | Hx of seizure | |
| 59 | 6 | Intellectual disability and abnormal behaviors | | Sever myopia | |
| 60 | 6 | Learning difficulties | | Poor vision | |
| 61 | 5 | Severe intellectual disability and abnormal behavior | | Myopia | |
| 62 | 7 | Learning difficulties, Hx of developmental delay | | Myopia | |
| 63 | 9 | Learning difficulties | | Refractive errors | |
| 64 | 1 | Sagittal sinus thrombosis with convulsion | | None | |
| 65 | 1.5 | Superior sagittal sinus thrombosis with seizures | | None | |
| 66 | 3 | Stroke | | None |
| 67 | 4 | | Hyperactivity and abnormal behaviors | None |
| 68 | 10 | Epilepsy | | Learning difficulties, poor school performance | |
| 69 | 5 | Recurrent seizures | | Intellectual disability | |

**Supplementary Table 2. Age of diagnosis and clinical presentation, course of disease, and adherence to treatment for patients in the FSG**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Patient No | Age at diagnosis  (years) | Significant clinical presentation | Course of disease | Adherence to treatment |
| 1 | 1 | Skeletal, behavioral | High arch palate, sometimes aggressive | Non-adherent |
| 2 | 0.4 | None | No abnormalities | Adherent |
| 3 | 5 | Skeletal, ocular | Fracture of right fifth metacarpal bone at 12 years, high arch palate, refractive error at 9 years | Non-adherent |
| 4 | 4 | None | No abnormalities | Non-adherent lately |
| 5 | 5 | Skeletal, Ocular, Vascular,  Scoliosis | High arch palate and mild scoliosis at 10 years, mild mitral prolapse at 16 years,  refractive error | Non-adherent |
| 6 | 0.5 | Skeletal and ocular | High arch palate and scoliosis and spondylitis by MRI, refractive error, Hx of seizures once at 14 years | Non-adherent |
| 7 | 1.3 | Skeletal | High arch palate and mild scoliosis at age 13 years | Non-adherent |
| 8 | 1 | None | Artificial left eye due to Hx of congenital left retinoblastoma at 7 months | Adherent |
| 8 | 0.75 | Skeletal and rare complication | High arch palate, Hx of chronic abdominal pain and weight loss for 4 months | Adherent |
| 10 | 2 | Skeletal and rare complication | High arch palate biliary pancreatitis with gallbladder stone and laparoscopic cholecystectomy at 15 years | Non-adherent |
| 11 | 3 | Skeletal | High arch palate | Adherent |
| 12 | 0.1 | Ocular | Refractive error | Adherent |
| 13 | 1 | Scoliosis | Mild scoliosis at 14 yrs | Non-adherent |