

Expanded Syndromic Retinal Disease Questions

Hearing loss -- Hearing aids? Present at birth? Static or progressive? Cochlear implants? Slow to walk (after 14 months)? Abnormal speech?

Polydactyly -- Postaxial? Which limbs?

Obesity -- Notably greater than parents and sibs?

Diabetes mellitus? -- Diabetes in mother or maternal relatives?

Cognitive issues? -- Static or progressive? Insistent perseverative questioning of examiner?

Seizures, ataxia, dysarthria, other brain abnormalities?

Renal disease? -- Bedwetting or frequent nocturia? Small kidneys on ultrasound? Abnormal renal function tests? Unexplained hypertension in children?

Heart abnormalities?

Abnormal dentition?

Cleft palate? Dysmorphic facies and/or malpositioned ears?

Other skeletal or joint abnormalities?

Pedal edema, skin or hair abnormalities?

Abnormal bruising or bleeding or frequent infections?

Malabsorption or muscle weakness?