**Introduction/Aims:** Leigh syndrome (LS) is an inherited, progressive, mitochondrial encephalomyopathy with onset in early childhood. Symptoms include failure to thrive, hypotonus, psychomotor regression and brain stem dysfunction. Several different gene mutations in nuclear or mitochondrial DNA can cause LS. The aim of this study was to describe ophthalmological findings in children with LS at diagnosis and over time, and relate the results to genetic findings and the results of a sex and age matched reference group.

**Methods:** A retrospective study was performed on 44 children with LS (19 female), with a median age of 2.4 years (range 0.6-14.2) at diagnosis. They were investigated and diagnosed by a multidisciplinary team at the Queen Silvia Children’s hospital in Gothenburg, Sweden, during 1987-2013. Twenty-seven children had known genetic mutations. The children underwent an ophthalmological examination including visual acuity (VA), eye motility, refraction, slit-lamp examination, ophthalmoscopy, and a full-field electroretinogram (ff-ERG). Seventeen of the children were available for follow-up over a mean time of 5.4 years (range 0.6 to 12.3). The results of the 17 children were compared with an age and sex matched reference group of healthy Swedish children (n=119).
**Results:** 36/44 (82%) of the children had ophthalmological findings. Most common findings were refractive errors (n=16/25), low VA (n=9/36), strabismus (n=8/42), reduced eye motility (n=8/42), optic atrophy (n=7/41), retinal macular pigmentation (n=6/39), and nystagmus (n=6/42). Several ophthalmological findings appeared over time. In 5/15, ff-ERG showed retinal dystrophy. No significant correlation between phenotype/genotype was found. The children with LS had significantly lower VA, more astigmatism and strabismus compared with the reference group at follow-up.

**Discussion/Conclusions:** In this unique cohort of children with LS, a vast majority showed ophthalmological findings at diagnosis, which increased over time. Therefore, we recommend that all children diagnosed with Leigh syndrome should be followed up with regular ophthalmological examinations.