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Clinical Features of Four Males and an Obligate Carrier in a Family with Lenz Syndrome. *S. Forrester[1], S. Rothert[1], R. Urban[2], M. Kovach[1], V. Kimonis[1]*. 1) Department of Pediatrics, SIU School of Medicine, Springfield, IL; 2) Department of Ophthalmology, Tampa General Hospital, Tampa, FL.

Lenz syndrome is a rare X-linked recessive syndrome first described by Lenz in 1955. Clinical features include anophthalmos or microphthalmos, microcephaly, mental retardation, external ear, digital, cardiac, skeletal, and urogenital anomalies (Traboulsi, et al 1988). Graham, et al (1991) localized the gene to Xq27-Xq28 in a Northern Ireland family with X-linked anophthalmos. However, the affected members in this family lacked digital, cardiac, and urogenital anomalies typically seen in Lenz syndrome. We present three brothers (ages 15 years, 9 years, and 18 months) and a maternal uncle (age 27 years) with Lenz syndrome. All four males have congenital anophthalmos with absence of the globes, optic nerve, and optic chiasm, and the youngest male has unilateral microphthalmos with corneal sclerosis. Delayed milestones and moderate to severe mental retardation were evident in the four males, and three of them have behavior problems. On physical examination, a high arched palate, pectus excavatum, finger and toe syndactyly and clinodactyly, fetal pads on all fingers and toes, and decreased muscle tone were noted in all four males. Scoliosis and outer ear anomalies were noted in three of the four males. The oldest brother has dilated left atrium of the heart and duplicated renal system, and the maternal uncle has right ventricular hypertrophy. An obligate carrier was noted to have syndactyly of the 2-3rdtoes bilaterally and dysplastic ears. We believe this family will add to the spectrum of clinical features of Lenz syndrome. Many of the cases published to date for Lenz syndrome are isolated reports. Linkage analysis in this family is in progress to identify the gene responsible for Lenz syndrome.