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Clinical Features of Four Males and an Obligate Carrier in a Family with Lenz Syndrome. *N.M. Reynolds[1], S. Forrester[1], M.J. Kovach[1], R. Urban[2], V.E. Kimonis[1].* 1) Dept of Pediatrics, Southern Illinois University School of Medicine, Springfield, IL; 2) Dept 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 anophthalmia, microcephaly, mental retardation, cardiac, skeletal, urogenital, external ear, and digital, anomalies. We present three brothers (ages 15 years, 9 years, and 18 months) and a maternal uncle (age 27 years) with congenital anophthalmia, hypotonia and moderate to severe mental retardation. Dysmorphic features include dysplastic ears, high arched palate, pectus excavatum, finger and toe syndactyly, clinodactyly and fetal pads. Other features include scoliosis, cardiac and renal abnormalities. Obligate carriers have a history of recurrent spontaneous losses. One carrier has dysplastic ears and syndactyly of the 2-3rd toes bilaterally; features which may be helpful in identification. Fourteen previously reported cases were additionally reviewed. Mental retardation was present in 100%, 93% had growth retardation, 69% had microcephaly, 88% had ear anomalies, 76% had dental anomalies, 12% had a congenital heart defect, 50% had a urogenital anomaly, 47% had a spinal deformity and 69% had anomalies of the fingers or toes.

Linkage and haplotype analysis in this family indicates that the gene is located in a 17.65 cM region on chromosome Xq27-Xq28 flanked by microsatellite markers DXS1232 and DXS8043. This region overlaps the anopthalmos locus ANOP1, but excludes the OCRL locus for Lowe oculocerbrorenal syndrome. Candidate genes involved in neuronal development that map to this critical interval include CXORF1 and KIAA0006. This is the first report of linkage analysis in Lenz microphthalmia a unique disorder associated with mental retardation and multiple anomalies.