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WARBURG MICRO SYNDROME IN TWO CHILDREN FROM A HIGHLY INBRED TURKISH FAMILY

BY M.S. YILDIRIM[1], A.G. ZAMANI[2] AND B. BOZKURT[1]

Summary: *Warburg micro syndrome in two children from a highly inbred Turkish family.* Warburg Micro syndrome (WMS) was first reported by Warburg in 1993. The cardinal features are microcephaly, microphthalmia, congenital cataract and intellectual disability. We report on two children from a highly inbred family with microcephaly, congenital cataract, optic atrophy, hypotonia and severe psychomotor retardation. This phenotype is similar to other reported rare entities and especially to the family reported by Warburg. Four other children in the same family may also have been affected. In this report, the symptoms and features of our cases are compared with the Warburg Micro syndrome patients in literature.

Key-words: Micro syndrome - Cataract - Consanguinity

INTRODUCTION

Warburg *et al.* first described this extremely rare, severe autosomal recessive genetic syndrome (WMS) in 1993 (7). This syndrome (OMIM 600118) is characterized by microcephaly, microcornea, congenital cataract, optic atrophy, intellectual disability, and hypogenitalism. The patients also have some dysmorphic features, such as beaked nose with a prominent nasal root, micrognathia, highly arched palate, hypertrichosis and large anteverted ears. Seizures and/or limb contractures may develop and delayed puberty is commonly observed (5).
WMS is caused by mutation in the *RAB3GAP* gene, located on chromosome 2. The *RAB3GAP* gene encodes a protein which is a key regulator of the Rab3 pathway implicated in exocytic release of ocular and neurodevelopmental trophic factors (2).
The symptoms and features of our cases are compared to the WMS patients in the literature.

CLINICAL REPORT

Two families were referred to our clinic with similar symptoms. Both families were reluctant to give detailed information to us but during pedigree analysis we found that they were from the same family

(1) Department of Genetics, Meram Medical Faculty, Necmettin Erbakan University, Konya, Turkey.
(2) Department of Opthalmology, Selcuklu Medical Faculty, Selcuk University, Konya, Turkey.