ENDEMIC GLAUCOMA IN THE MUNE-BRГUD AREA

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SUMMARY – In the past century, research into the prevalence of congenital glaucoma among the Mune and Brгud villagers showed that glaucoma was present in 38% of the population. Genealogical studies established the presence of congenital glaucoma through six generations of the villagers. Tradition has it that members of the Šori family have diseased eyes from time immemorial and that other families have through generations been linked to this family, in a repeated circle of conjugal relationships. From the considerable documentation collected, it appears that approximately 50% of the population are genetically linked to the central Šori family. In this group, about half of the offspring show symptoms of this congenital disease. Clinical characteristics of the Mune-Brгud congenital glaucoma are similar to those of the late congenital glaucoma. It is caused by a congenital anomaly of the iridocorneal angle, and is clinically manifested between the age of 10 and 40. It is characterized by goniodysgenesis and high insertion of the iris, remnants of undifferentiated mesodermal embryonic tissue, widening of the trabecular meshwork, decreased aqueous outflow, and regularly present markedly deep anterior chamber. Megalocornea, hypoplasia of the corneal stroma and moderate myopia are present sporadically.

Key words: Mune-Brгud glaucoma, hereditary glaucoma, congenital glaucoma

The villages of Mune and Brгud are situated in the north Adriatic region, on the slopes of the Učka Mountain (Fig. 1). In the past century, the research into the prevalence of congenital glaucoma among villagers of the Mune and Brгud villages showed that glaucoma was present in 38% of the population. Genealogical studies established the presence of congenital glaucoma through six generations of the Mune and Brгud villagers.

Generally, the anomaly that results in congenital glaucoma is genetically determined and shows the recessive inheritance pattern in most cases. This means that both parents are heterozygous and have a single defective gene. Twenty-five percent of their offspring would be expected to have two defective genes and thus to exhibit infantile glaucoma.

In the past generations, all Mune-Brгud family members had glaucoma, and considering its clinical manifestation, the disease had a "dominant character". Since it is a specifically structured clinical manifestation in a geographically restricted population, the genetic material accumulated (modifier) in the heterozygote to the homozygote with a very high expression of the mutant gene where, through the evolution of dominance, a new form of glaucoma developed, known as the Mune-Brгud type of glaucoma.

Genealogical studies report on typical family names of families with glaucoma that recur in conjugal relationships through several generations. These family names are Afrić, Ujičić, Hrvatin, Štenberger, Stambul, Brentin, Gregorić and others, and experience confirms that a glaucomatous disease can be seriously suspected in relation to the family name.

It has been reported that the Mune and Brгud villagers have a traditional 200-year-old lifestyle and are strictly confined to their villages. The elders have by common consent rejected the possibility of the railroad passing through their village because it might disturb the livestock, and to prevent strangers from coming to their village (Fig. 2).

The villagers lived a simple, modest and neighbor-dependent life. They were mainly farmers and foresters, while the eminent family nicknamed Šori that had

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27 members 200 years ago, carried on trade. Even today people talk about them. Their nickname could have come from signori, Šiort in the dialect, meaning Italian gentlefolk, which meant that they were the most significant and the most distinguished family in the village, smarter and richer than others. They were the first in the village and they made important decisions regarding the community as the village elders.

Close relatives married each other. To this day, the elderly of the region say that it is better that the grandson marries a girl from the family in order that everyone remains in the house, the family stays united and the property preserved.

Tradition has it that members of the Šori family have diseased eyes from time immemorial and that through generations other families have been linked to this fami-
2nd generation family members and in 75% of the 3rd generation family members. In the 4th generation of subjects, the disease has not appeared so far (Fig. 4).

On the genealogical tree of the Afrić and Brentini families, consanguinity is not observed in the 3rd generation (Fig. 5).

These deeply rooted characteristics, established through a particular lifestyle and linked by consanguinity of the villagers, disappeared for the most part after World War I and World War II, and as the result of migration, mostly to the USA, which has only reflected in the last two generations. Mixed marriages are no longer taking place as the result of modern way of life and better communications. This is the main reason why the manifestation of congenital glaucoma in these villages has been decreasing, however, the inheritance component of the disease is still observed depending on the expression of the disease in the new marital relationships.

Clinical characteristics of the Mune-Brgud congenital glaucoma are similar to those in the late congenital glaucoma (glaucoma congenitum tardum). It is caused by a congenital anomaly of the iridocorneal angle, and is clinically manifested between the age of 10 and 40. It is characterized by goniodysgenesis and high insertion of the iris, remnants of undifferentiated mesodermal embryonic tissue, widening of the trabecular meshwork, decreased aqueous outflow, and a regularly present markedly deep anterior chamber. Megalocornea, hypoplasia of the corneal stroma, and moderate myopia are present sporadically (Fig. 6).

Treatment is administered according to the principles of treatment for congenital glaucoma. The method of choice is antiglaucomatous filtering surgery. In clinical practice, we have observed that even when the dis-
ease is medicamentously stabilized, sudden relapse may occur with elevated intraocular pressure, quick development of glaucomatous cupping, and rapid visual field deterioration leading to blindness, all in one year. For this reason, almost all of our registered patients with glaucoma of the Mune-Brugd type underwent surgery. Newly detected cases of glaucomatous disease that either by inheritance or by characteristic family names or by typical clinical manifestations pertain to the families from Mune and Brugd are under intensive follow up 4 to 5 times a year.

References

Sažetak
ENDEMSKI GLAUKOM U NASELJIMA MUNE I BRGUD

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Ključne riječi: glaukom Mune-Brugd, nasljedni glaukom, kongenitalni glaukom