**A recurrent character and a high frequency of 11p13 deletion affecting *PAX6* downstream regulatory regions in aniridia patients from Russia**

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**Purpose**. Aniridia (OMIM [106210](http://omim.org/entry/106210)) is autosomal dominant congenital panocular disorder caused by *PAX6* gene damage by large deletions and small mutations. The study aimed to determine *PAX6* mutation spectrum in Russian aniridia patients.

**Methods**. 121 unrelated families with congenital aniridia (147 patients) underwent ophthalmic examination and DNA testing: Sanger sequencing and MLPA followed by loss of heterozygosity of STR markers analysis.

**Results**. 16 patients from 10 unrelated families share the same 11p13 0.5–1.5 Mb deletion affecting *PAX6* downstream regulatory regions: 5 familial and 5 sporadic. The frequency of this deletion, 8.3% (10/121), is higher than *PAX6* hotspot c.718C>T rate (7/121, 5.7%).

**Conclusions**. The high rate of the deletion in studied cohort suggests a common underlying mechanism of its formation and points to 11p13 genomic region instability.

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