The First Clinical CNV Database in Russia

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In Russia, the chromosomal microarray analysis (CMA) for clinical purposes is performed mostly by private laboratories. We use CMA for postnatal diagnosis in children with multiple congenital anomalies (MCA), developmental delay (DD) and dysmorphic facial features (DF), for prenatal diagnosis if chromosomal anomalies in fetus are suspected, and for testing products of conception and FFPE tumor samples. Over the course of 5 years, we created the largest CNVs database in Russia that includes information on 6155 samples tested in our laboratory (Fig. 1). Implementing this database not only solved the problem of storing CNVs data in a standardized form, but also made it possible to classify CNVs by type, size, gene content, pathogenicity, major clinical features in patient, the kind of the sample and the array type. In particular, we evaluated the prevalence of microdeletion syndromes and pathogenic CNVs in different patient groups (Fig. 2). In a group of 3299 patients with MCA, DD and DF, pathogenic CNVs were detected in 565 cases (17.1%) (Fig. 3).

The database contains information on CNVs detected mainly in populations of Russia, and it is used to determine the clinical significance of CNVs previously not described in other databases, e.g. DGV or DECIPHER, and as an additional tool for evaluation of pathogenicity of all other CNVs.

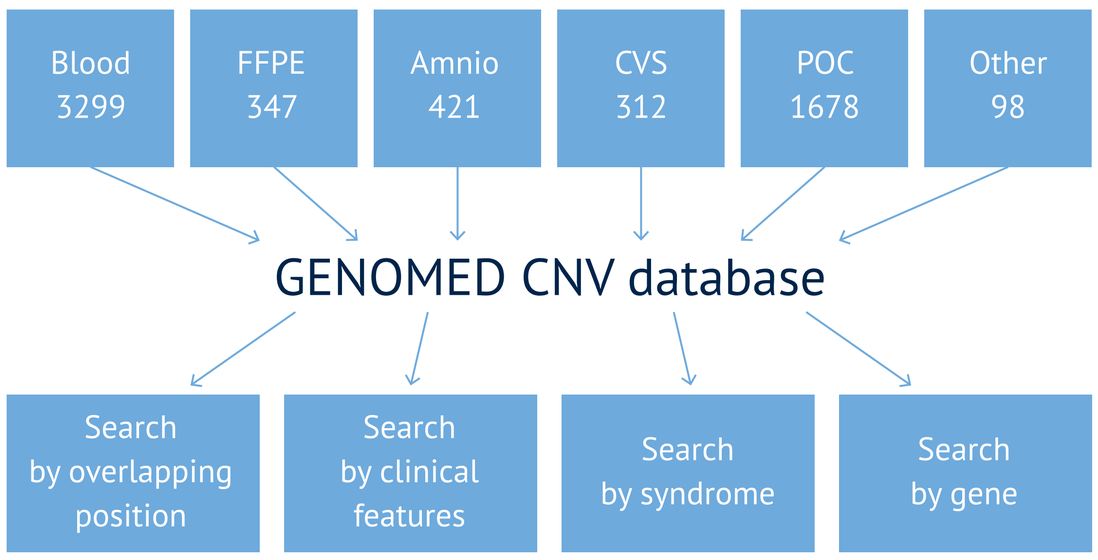
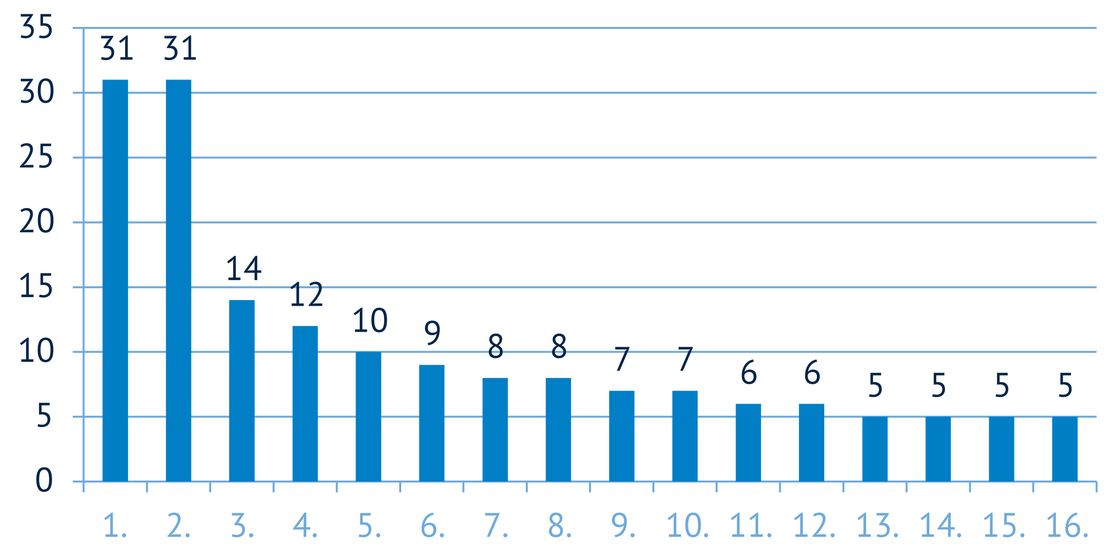


Fig. 1. Genomed CNV database.



1. Chromosome 22q11.2 deletion syndrome
2. Prader-Willi/Angelman syndrome
3. Williams syndrome
4. Phelan-McDermid syndrome
5. Chromosome 1p36 deletion syndrome
6. Wolf-Hirschhorn syndrome
7. Down syndrome
8. Chromosome 22q11.2 duplication syndrome
9. Trisomy 9 mosaicism syndrome
10. Chromosome 15q11.2 deletion syndrome
11. Smith-Magenis syndrome
12. Koolen-De Vries syndrome
13. Chromosome 1q21.1 deletion syndrome
14. Chromosome 15q13.3 microdeletion syndrome
15. Mental retardation, autosomal dominant, type 1
16. Chromosome 5q14.3 deletion syndrome

Fig. 2. The most common syndromes in postnatal cohort.

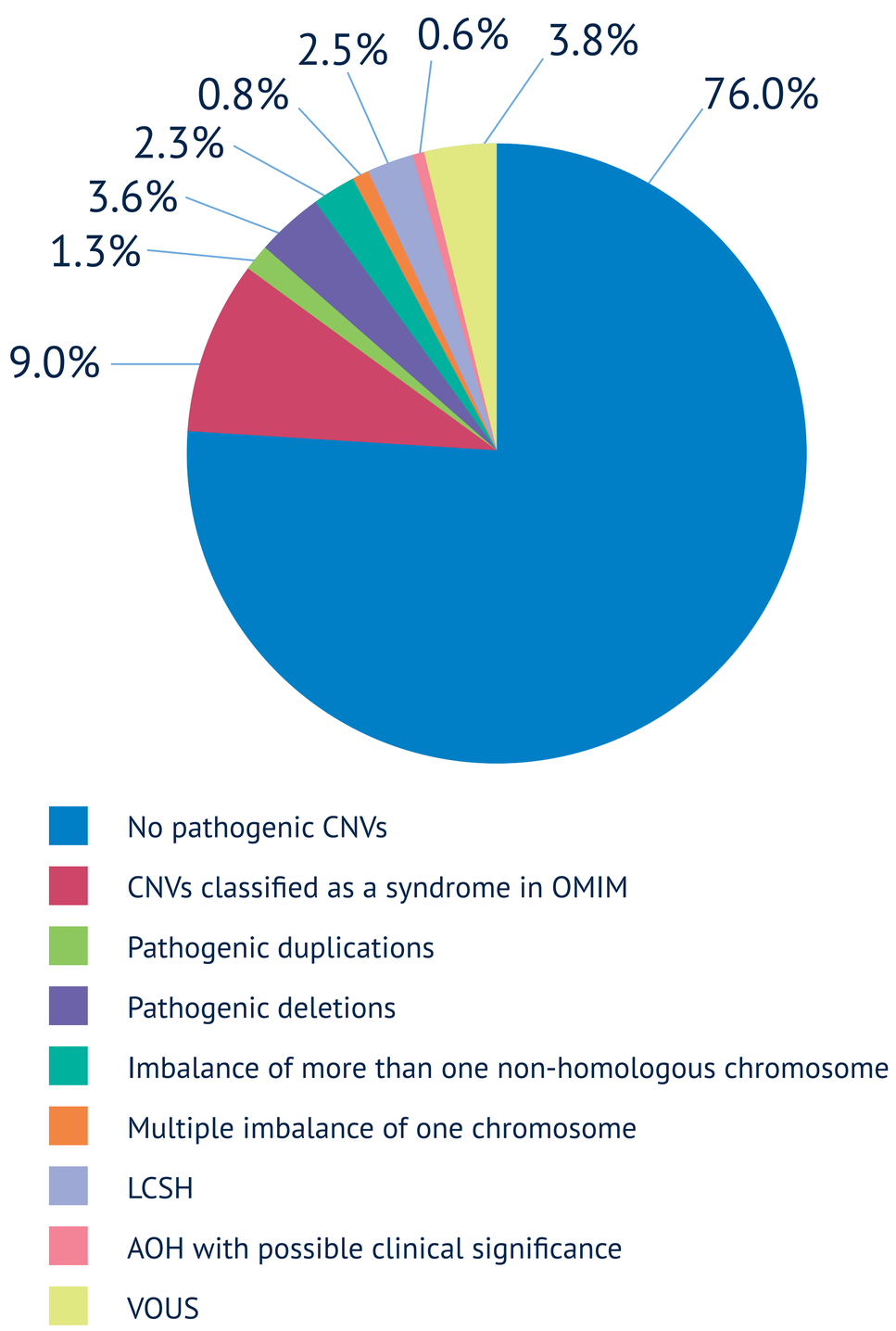


Fig. 3. Chromosomal microarray analysis of 3299 postnatal samples using Affymetrix Cytoscan HD, 750k and Optima arrays.

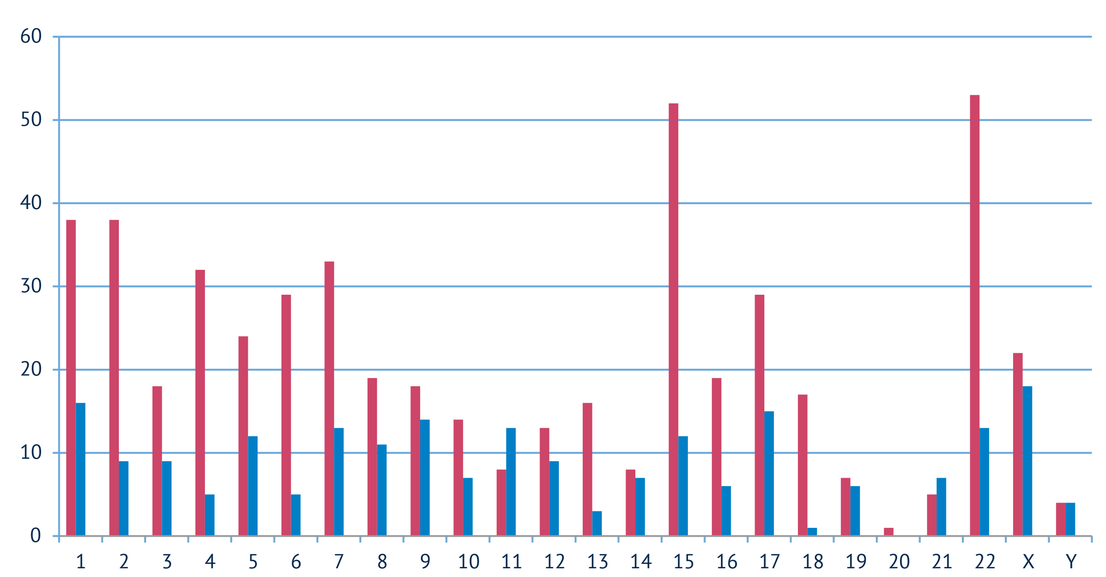


Fig. 4. Distribution of CNVs by chromosomes in patients with MCA, DD and DF.