HEREDITARY DISEASES AND CONGENITAL MALFORMATIONS REGISTRATION AND MONITORING INFORMATION SYSTEM

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Introduction. Database on hereditary diseases is one of the main tasks of healthcare. Register of hereditary pathology allows to assess accurately the frequency of childbirths with various hereditary diseases and congenital malformations in the population. The purpose of this work is to present the frequency and structure of congenital and hereditary pathology based on the use of the original genetic database for monitoring of congenital malformations and hereditary diseases.

Methods: Clinical-genealogical, cytogenetic, FISH method, molecular-genetic (MLPA, mass spectrometry and fluorimetry, Sanger sequencing, NGS), statistical.

Results: Up to date, National Research Center for Maternal and Child Health's genetic register database has registered 4564 patients and fetuses with congenital and hereditary pathologies. Congenital malformations prevail in the database with 2317 cases (50,8%), including congenital malfunctions of obligatory registration according to International Register (EUROCAT). Chromosomal pathology is identified in 1283 (28,1%) patients, monogenic – in 964 (21,1%) cases. In congenital malformations the largest proportion belongs to congenital malformations of blood circulation – 694 cases (29,9%), followed by congenital malformations of gastrointestinal tract – 304 cases (13,1%), multiple malformations – 275 cases (12,8%), congenital malformations of urinary system – 298 patients (12,8%), congenital malformations of nervous system – 217 cases (9,4%), congenital malformations of facial structures (187 patients, 8%) and of musculoskeletal system with 175 cases (7,5%). The most common of the chromosomal pathologies was registered Down syndrome with 616 cases (48,1%), Edwards syndrome – in 122(9,5%). Turner syndrome – in 101(7,9%) cases, Kleinfelter's syndrome – in 66 (5,1%), Patau syndrome – in 30 (2,3%), other gender chromosome pathologies – 19 (1,5%), structural rearrangements – 329 (25,6%). Monogenic pathology was detected in 964 (21,1%) cases. Among monogenic pathology, the most common were osteogenesis imperfecta, chondrodystrophy, adrenogenital syndrome, congenital hypothyroidism, mucopolysaccharidosis, Prader-Willi syndrome.

Conclusion: The formation of a database of congenital and hereditary pathologies with full coverage of all fetuses, newborns and children at an older age, a comprehensive examination of families and clarification of the diagnosis allows to establish the frequency, structure and dynamics of congenital malformations and hereditary pathology.