

QUALITY CONTROL IN MOLECULAR GENETICS LABORATORY: A LITERATURE REVIEW

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ABSTRACT

Relevance: Integrating molecular biomarkers with rigorous quality control (QC) measures in laboratory settings is essential for enhancing early detection strategies and prognostic evaluation in cancer patients. Precision and QC in laboratory diagnostics of oncological diseases have become particularly significant in the widespread implementation of targeted and personalized therapy.

The study aimed to review publications evaluating quality control in biomarker identification within molecular genetics laboratories, using ovarian cancer diagnostics as a case study.

Methods: This systematic literature review conducted in the framework of this study revealed 220 records, leading to 165 unique publications, of which 24 full-text articles were included in this review. The study followed Preferred Reporting Items for Systematic Reviews and Meta-Analyses 2020 (PRISMA 2020) guidelines.

Results: All analyzed sources showed that Implementing QC, including calibration, internal controls, and proficiency testing provided by the College of American Pathologists (CAP), significantly reduces errors despite ongoing funding constraints. The European Molecular Quality Network (EMQN) and CAP jointly offer proficiency testing programs to evaluate laboratory performance globally, ensuring consistency and reliability in testing outcomes.

Conclusion: Ensuring the accuracy and reliability of molecular diagnostic tests is critical in clinical settings, particularly for conditions such as ovarian cancer, where precise genetic analysis informs both diagnosis and treatment. Further advancements in early detection and personalized treatment can be achieved by integrating emerging technological innovations within robust QC framework, ultimately improving patient outcomes. Consequently, the establishment of standardized guidelines and standard operating procedures for molecular genetic testing, with a specific focus on the molecular genetic diagnosis of ovarian cancer, is imperative.

Keywords: molecular genetic testing, quality control (QC), BRCA1, BRCA2, polymerase chain reaction (PCR), next-generation sequencing (NGS).

Introduction: Modern medical personalization trends require the implementation and application of advanced diagnostic technologies. In recent years, this process has experienced significant advancements, notably in oncology. Various biomarkers play a pivotal role in personalization. Accurate detection of biomarkers and genetic alterations, in particular, using advanced and precise techniques of polymerase chain reaction (PCR) and next-generation sequencing (NGS), relies on stringent QC standards, which molecular genetics laboratories must uphold to ensure diagnostic reliability [1, 2]. We utilized the diagnosis of biological and molecular markers in ovarian cancer as a case study, considering the unique characteristics of disease diagnosis and progression, the application of detection methods, and the critical role of molecular markers in therapeutic decision-making.

The study aimed to review publications evaluating quality control in biomarker identification within molecular genetics laboratories, using ovarian cancer diagnostics as a case study.

Materials and Methods: A systematic review of literature conducted in the framework of this study revealed 220 records, leading to 165 unique publications, after which 70 full-text papers were analyzed. The study followed Preferred Reporting Items for Systematic Reviews

and Meta-Analyses 2020 (PRISMA 2020) guidelines [3] to assess worldwide QC procedures in molecular genetics laboratories that test for ovarian cancer. The research contained 25 unrelated ovarian cancer studies, excluded 15 works without QC information, 12 studies with imprecise methods, and five articles predating 2015. Eight studies passing the Critical Appraisal Skills Programme (CASP) checker with scores exceeding 80% were assembled for synthesis [4].

Search strategy: Literature sources from the PubMed, Scopus, Web of Science, and Google Scholar databases published between 2015 and 2025 were reviewed and analyzed. The research used the combination of "Quality Control" OR "Quality Assurance" together with "molecular genetics" OR "molecular diagnostics" supported by "ovarian cancer" AND "laboratory practices." A manual citation search was also performed, and references were organized using EndNote X9 to complete the research process. [5].

Exclusion criteria: Studies that failed to show laboratory or methodological details or were published before 2015 or in languages other than English were excluded.

Study selection process: The review proceeds through a clear workflow, which makes its findings strong and connected to the diagnostic QC of ovarian cancer, as shown in Figure 1.

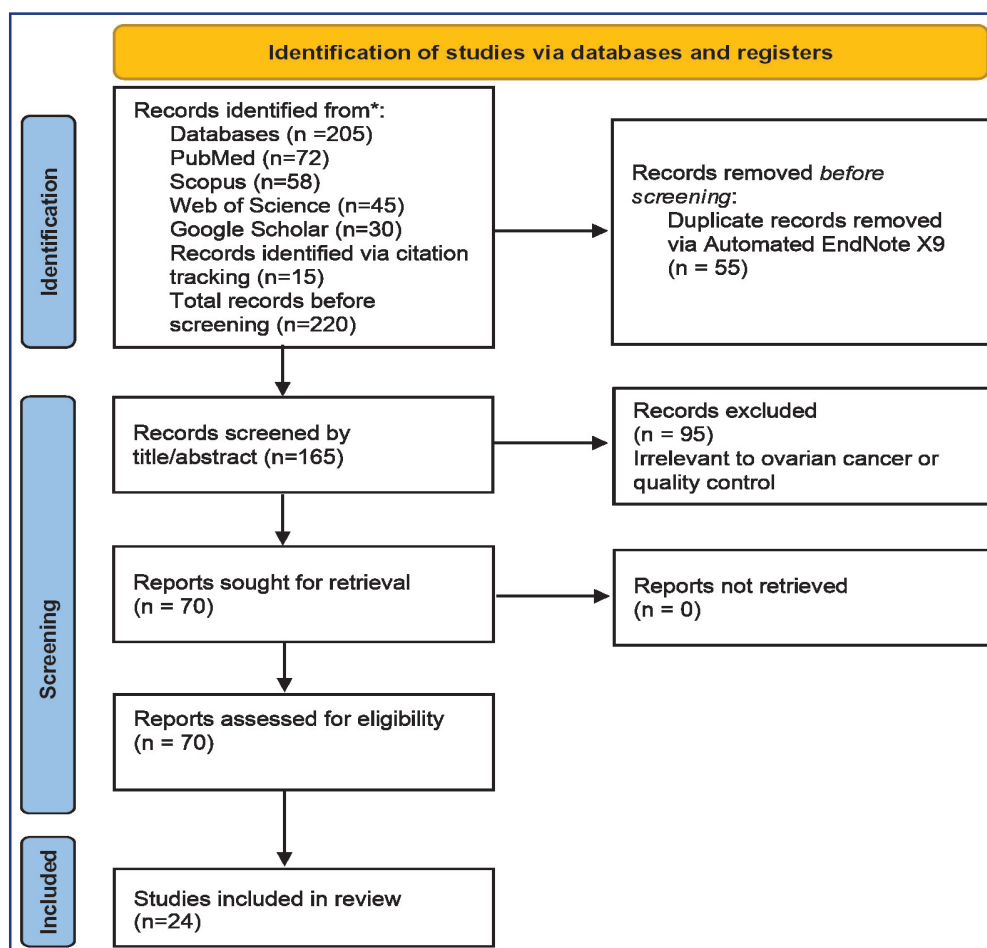


Figure 1 – PRISMA flow diagram

Results:

Quality control protocols in molecular genetics laboratories: With the rapid expansion of molecular genetic diagnostic methods in recent years, alongside the increasing number of tests and panels, the implementation and integration of rigorous QC systems in laboratories have become essential to ensure accuracy, reliability, and standardization. Since implementing genetic testing, the interest in and necessity for QC protocols to enhance testing accuracy have grown significantly. Figure 2 presents the evolution of QC protocols in molecular genetics laboratories, outlining key milestones in their development.

As shown in Figure 2, the Clinical Laboratory Improvement Amendments (CLIA) of 1988 established foundational standards, while the National Institutes of Health (NIH) and the Department of Energy (DOE) in 1997 emphasized the importance of Quality Assurance [6]. In 2009, the Centers for Disease Control and Prevention (CDC) outlined best practices for laboratory quality management [7]. The Minimum Information for Publication of Quantitative Real-Time Polymerase Chain Reaction Experiments (MIQE) guidelines, introduced in 2010, standardized quantitative PCR methodologies, followed by a QC framework in 2012 [8, 9]. The 2020 MIQE updates further refined QC measures addressing digital PCR advancements [10].

QC measures in molecular genetics laboratories are pivotal in maintaining assay integrity, minimizing diag-

nostic errors, and ensuring reproducibility across different testing facilities.

External quality assessment (EQA) by organizations such as the European Molecular Genetics Quality Network (EMQN) and the College of American Pathologists (CAP) is used to evaluate laboratory performance on a global level [11, 12]. These programs provide standardized proficiency testing schemes that assess laboratory practices, monitor test consistency, and identify improvement areas. By benchmarking results against international standards, EQA programs contribute to the harmonization of molecular diagnostics and reinforce best practices in genetic testing. External quality assessments from CAP and EMQN strive to improve worldwide measurement standards by accrediting laboratories through proficiency testing programs that protect pathology and laboratory medicine quality. Programs and their brief description are shown in Table 1.

QC programs in laboratories typically comprise three distinct phases. The accuracy of molecular diagnostic equipment is contingent on stringent calibration protocols.

Laboratories in the United States and Europe adhere to International Organization for Standardization (ISO) guidelines, such as ISO 15189, to maintain thermal precision in PCR machines. These calibration standards are essential for ensuring consistent amplification conditions, thereby reducing variability in test results and enhancing diagnostic accuracy [13].

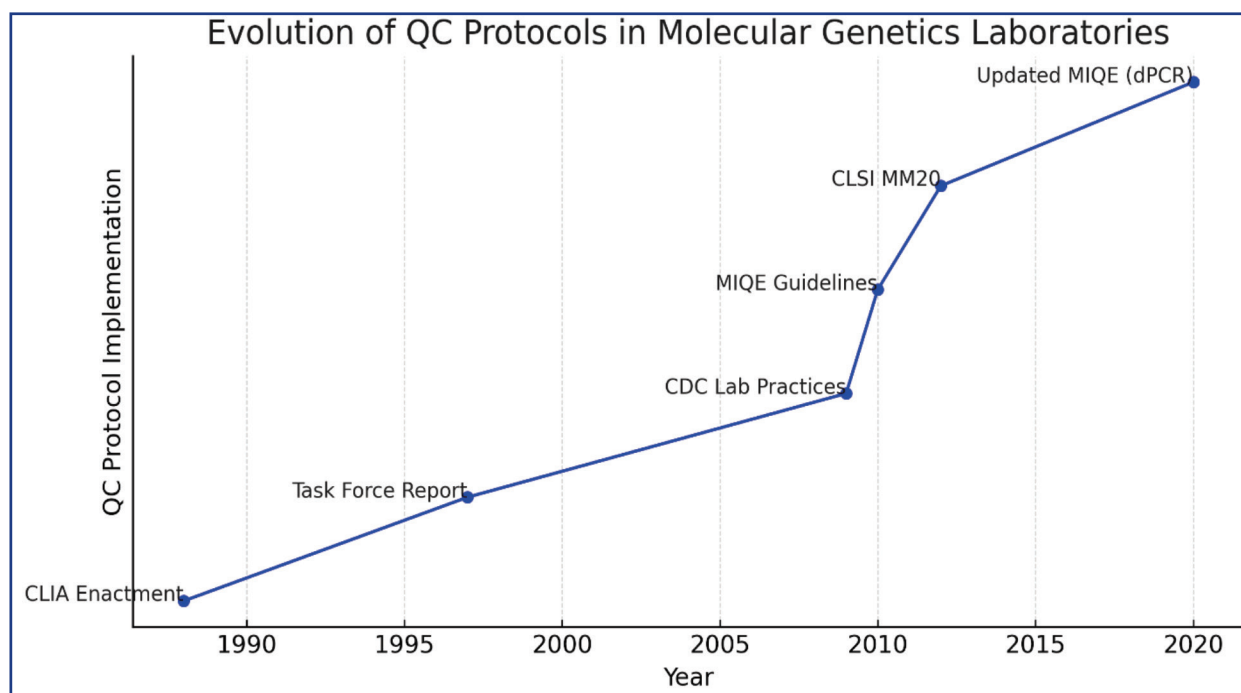


Figure 2 – Evolution of QC protocols used in Molecular Genetics Laboratories

Table 1 – External quality control programs and guidelines

Program	Description	Source
College of American Pathologists	Provides accreditation and proficiency testing for pathology and laboratory medicine.	CAP
European Molecular Genetics Quality Network	Offers external quality assessment for molecular genetics laboratories.	EMQN

Reliable molecular testing depends on rigorous assay validation and contamination detection protocols. Laboratories worldwide implement positive and negative control sample testing to assess assay performance and de-

tect potential contaminants. Well-characterized reference samples ensure that molecular assays produce consistent and reproducible results, further strengthening diagnostic reliability [2]. QC phases are provided in Table 2.

Table 2 – Quality control protocols in molecular genetics laboratories

Quality control protocol	Description	Global examples
Calibration	Regular calibration of equipment to maintain accuracy.	Laboratories in the USA and Europe follow ISO standards for calibration.
Internal Controls	Use of positive and negative controls in each assay to validate results.	Widely implemented in accredited molecular genetics labs globally.
Proficiency Testing	Participation in external quality assessment schemes to benchmark performance.	Programs like EMQN and CAP offer proficiency testing worldwide.

Key findings from studies on quality control in molecular genetics laboratories for ovarian cancer diagnostics. With the advancement of novel diagnostic methodologies for ovarian cancer, the standards and requirements for quality control programs have undergone significant evolution. The analyzed studies underscore significant advancements in genetic testing and QC systems, particularly molecular genetic diagnostics of ovarian cancer, regardless of techniques and methods used. In 2015, Strom et al. reported that NGS achieved 99% accuracy in detecting *BRCA1* and *BRCA2* using strict control systems and calibration methods [14]. Other study using multigene panel testing, showed that this panel matched 95% of all results obtained through Sanger sequencing but highlights the necessity for standardization practices [15]. According to C.R. Marshall et al., Whole Genome Sequencing provided 98% sensitivity, improving by regular-

ly implementing QC procedures [16]. D. Grafodatskaya et al. stated that testing accuracy for *BRCA1* and *BRCA2* improved when EQA was adopted along with a Limit of Detection that exceeded 10% [17]. In 2023, E.T. Kim et al. verified the use of NGS to analyze *BRCA1* and *BRCA2* genes in formalin-fixed paraffin-embedded samples by reaching 99% accuracy rates at sequencing depths exceeding 40x, eliminating unnecessary false positive test results [18]. The data from the Menon & Brash study (2023) showed that extended sequencing depth above 1000x and additional strict QC serve to reduce errors during rare variant detection [19]. And more recent study, published in 2024 by T. McDevitt et al. demonstrated reliable genetic testing through paired analysis by following EMQN guidelines and ISO 15189 standards to achieve maximum analytical sensitivity [20]. Key findings are documented in Table 3.

Table 3 – Key findings from studies on quality control (QC) in molecular genetics laboratories for ovarian cancer diagnostics

Authors	Key findings
Lincoln et al., 2015	Hereditary ovarian cancer testing through multigene panels detects more conditions yet needs strong QC procedures to achieve 95% accuracy compared to Sanger testing methods.
Strom et al., 2015	The NGS assay for BRCA1/2 reached 99% accuracy during validation through proper control implementation and calibration procedures that enhanced laboratory reliability to identify rare variants.
Marshall et al., 2020	The validation of Whole Genome Sequencing technology requires specific performance metrics to reach a sensitivity level of 98% when combined with standard QC procedures for complete ovarian cancer genetic analysis.
Grafodatskaya et al., 2021	EQA should be applied along with a Low Limit of Detection $\geq 10\%$ to improve the accuracy of BRCA1/2 testing for ovarian cancer.
Kim et al., 2023	The precision of NGS validation for BRCA in ovarian FFPE reaches 99% accuracy when the QC depth exceeds 40x.
Menon & Brash, 2023	The evaluation of NGS QC focuses on mutation detection at frequencies under 1000x depth and utilizes controls to prevent errors in ovarian cancer variant identification.
McDevitt et al., 2024	Applying EMQN guidelines requires implementing ISO 15189 standards, EQA participation, and paired testing for ovarian cancer, ensuring high analytical sensitivity.

Discussion: Despite significant progress in establishing QC systems worldwide and accuracy levels of *BRCA* ½ mutations with 99% detection achieved through NGS mirror global findings, some laboratories face technical difficulties that differ from the international research focus and align with resource constraints discussion [18-19, 21, 22]. The combined initiative of standardization practice using CAP and EMQN frameworks establishes a comprehensive system beyond the diverse perspectives described by different authors [15, 21]. The work by Wang X in 2024 and Hamidi et al. (2023), along with other emerging technologies, puts this study ahead of domestic biomarker research while demonstrating the significance of QC for precision medicine advancement [23, 24].

Conclusion: In recent years, molecular genetic research has been increasingly incorporated into the routine practice of oncology institutions in the Republic of Kazakhstan, as well as into diagnostic and treatment protocols for oncological diseases, particularly ovarian cancer. Ensuring the accuracy and reliability of molecular diagnostic tests is critical in clinical settings, particularly for conditions such as ovarian cancer, where precise genetic analysis informs both diagnosis and treatment. Standardized protocols, combined with calibration, internal controls, and proficiency testing, enhance diagnostic accuracy, as demonstrated by global research studies. The continuity of laboratory standardization relies on sustained efforts aligned with international quality benchmarks, such as ISO 15189, CAP, and EMQN. Further advancements in early detection and personalized treatment can be achieved by integrating emerging technological innovations within a robust QC framework, ultimately improving patient outcomes.

References:

- Vashisht V., Vashisht A., Mondal A. K., Woodall J., Kolhe R. From Genomic Exploration to Personalized Treatment: Next-Generation Sequencing in Oncology // *Curr. Iss. Mol. Biol.* – 2024. – Vol. 46, No. 11. – P. 12527-12549. <https://doi.org/10.3390/cimb46110744>
- Moldovan E., Moldovan V. Controls in real-time polymerase chain reaction-based techniques // *Acta Marisensis – Ser. Med.* – 2020. – Vol. 66, No. 3. – P. 79-82. <https://doi.org/10.2478/amma-2020-0025>
- Page M. J., McKenzie J. E., Bossuyt P. M., Boutron I., Hoffmann T. C., Mulrow C. D., Shamseer L., Tetzlaff J. M., Akl E. A., Brennan S. E., Chou R., Glanville J., Grimshaw J. M., Hróbjartsson A., Lalu M. M., Li T., Loder E. W., Mayo-Wilson E., McDonald S., Moher D. The PRISMA 2020 statement: An updated guideline for reporting

systematic reviews // *BMJ.* – 2021. – Vol. 372. – Art. no. 71. <https://doi.org/10.1136/bmj.n71>

4. CASP. Critical Appraisal Checklists [Internet]. – *Critical Appraisal Skills Programme*, 2024. – Дана docmyna: 25.03.2025. – Aðpec docmyna: <https://casp-uk.net/casp-tools-checklists/>

5. Bustin S. A., Benes V., Garson J. A., Hellemans J., Huggett J., Kubista M., Mueller R., Nolan T., Pfaffl M. W., Shipley G. L., Vandesompele J., Wittwer C. T. The MIQE guidelines: minimum information for publication of quantitative real-time PCR experiments // *Clin. Chem.* – 2009. – Vol. 55, No. 4. – P. 611-622. <https://doi.org/10.1373/clinchem.2008.112797>

6. Braga F., Pasqualetti S., Aloisio E., Panteghini M. The internal quality control in the traceability era // *Clin. Chem. Lab. Med.* – 2021. – Vol. 59, No. 2. – P. 291-300. <https://doi.org/10.1515/cclm-2020-0371>

7. Chen B., Gagnon M., Shahangian S., Anderson N.L., Howerton D.A., Boone J.D., Centers for Disease Control and Prevention (CDC). Good laboratory practices for molecular genetic testing for heritable diseases and conditions // *MMWR. Recommend. Rep.* – 2009. – Vol. 58, No. RR-6. – P. 1-4. <https://www.cdc.gov/mmwr/preview/mmwrhtml/rr5806a1.htm>

8. Williams L.O., Cole E.C., Lubin I.M., Iglesias N.I., Jordan R.L., Elliott L.E. Quality assurance in human molecular genetics testing: status and recommendations // *Arch. Pathol. Lab. Med.* – 2003. – Vol. 127, No. 10. – P. 1353-1358. <https://doi.org/10.5858/2003-127-1353-QAIIHMG>

9. Clinical & Laboratory Standards Institute. CLSI MM20. Quality Management for Molecular Genetic Testing [Internet]. – 1st ed. – CLSI, 2023. – Дана docmyna: 25.03.2025. – Aðpec docmyna: <https://clsi.org/standards/products/molecular-diagnostics/documents/mm20/>

10. Huggett J. F., Whale A. S., De Spiegelaere W., Trypsteen W., Nour A. A., Bae Y.-K., Benes V., Burke D. G., Corbisier P., Cleveland M. The digital MIQE guidelines update: Minimum information for publication of quantitative digital PCR experiments for 2020 // *Clin. Chem.* – 2020. – Vol. 66, No. 8. – P. 1012-1029. <https://doi.org/10.1093/clinchem/hvaa125>

11. European Molecular Genetics Quality Network (EMQN). EMQN's EQA schemes are designed to test the whole analytical process of a genomics diagnostic laboratory [Internet]. – Дана docmyna: 25.03.2025. – Aðpec docmyna: <https://www.emqn.org/our-eqa-schemes/>

12. College of American Pathologists. Proficiency Testing [Internet]. – Дана docmyna: 25.03.2025. – Aðpec docmyna: <https://www.cap.org/laboratory-improvement/proficiency-testing>

13. ISO. Online Browsing Platform (OBP). ISO 15189:2022(en). Medical laboratories – Requirements for quality and competence [Internet]. – Дана docmyna: 25.03.2025. – Aðpec docmyna: <https://www.iso.org/obp/ui/en/#iso:std:iso:15189:ed-4:vi:en>

14. Strom C.M., Rivera S., Elzinga C., Angeloni T., Rosenthal S.H. Development and Validation of a Next-Generation Sequencing Assay for BRCA1 and BRCA2 Variants for the Clinical Laboratory // *PLOS ONE.* – 2015. – Vol. 10, No. 8. – Art.no. e0136419. <https://doi.org/10.1371/journal.pone.0136419>

15. Lincoln S. E., Kobayashi Y., Anderson M.J., Yang S., Desmond A.J., Mills M.A., Nilsen G.B., Jacobs K.B., Monzon F.A., Kurian A.W.,

Ford J.M., Ellisen L.W. A systematic comparison of traditional and multigene panel testing for hereditary breast and ovarian cancer genes in more than 1000 patients // *J Mol Diagn.* – 2015. – Vol. 17, No. 5. – P. 533-544. <https://doi.org/10.1016/j.jmoldx.2015.04.009>

16. Marshall C.R., Chowdhury S., Taft R.J., Lebo M.S., Buchan J.G., Harrison S.M., Rowsey R., Klee E.W., Liu P., Worthey E.A., Jobanputra V., Dimmock D., Kearney H.M., Bick D., Kulkarni S., Taylor S.L., Belmont J.W., Stavropoulos D.J., Lennon N.J., Medical Genome Initiative. Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease // *NPJ Genom. Med.* – 2020. – Vol. 5. – Art. no. 47. <https://doi.org/10.1038/s41525-020-00154-9>

17. Grafodatskaya D., O'Rielly D. D., Bedard K., Butcher D. T., Howlett C. J., Lytwyn A., McCready E., Parboosingh J., Spriggs E. L., Vaags A. K., Stockley T. L. Practice guidelines for BRCA1/2 tumour testing in ovarian cancer // *J. Med. Genet.* – 2022. – Vol. 59. – P. 727-736. <https://doi.org/10.1136/jmedgenet-2021-108238>

18. Kim E.T., Jeong H.E., Yoon H.J., Kim K.H., Suh D.S. Validation of multigene panel next-generation sequencing for the detection of BRCA mutation in formalin-fixed, paraffin-embedded epithelial ovarian cancer tissues // *Taiwan. J. Obstet. Gynecol.* – 2023. – Vol. 62, No. 1. – P. 66-70. <https://doi.org/10.1016/j.tjog.2022.07.010>

19. Menon V., Brash D.E. Next-generation sequencing methodologies to detect low-frequency mutations: "Catch me if you can" // *Mutat. Res. Rev. Mutat. Res.* – 2023. – Vol. 792. – Art. no. 108471. <https://doi.org/10.1016/j.mrrrev.2023.108471>

20. McDevitt T., Durkie M., Arnold N., Burghel G.J., Butler S., Claes K.B.M., Logan P., Robinson R., Sheils K., Wolstenholme N., Hanson H., Turnbull C., Hume S. EMQN best practice guidelines for genetic testing in hereditary breast and ovarian cancer // *Eur. J. Hum. Genet.* – 2024. – Vol. 32, No. 5. – P. 479-488. <https://doi.org/10.1038/s41431-023-01507-5>

21. O'Daniel J.M., McLaughlin H.M., Amendola L.M., Bale S.J., Berg J.S., Bick D., Bowling K.M., Chao E.C., Chung W.K., Conlin L.K., Cooper G.M., Das S., Deignan J.L., Dorschner M.O., Evans J.P., Ghazani A.A., Goddard K.A., Gornick M., Farwell Hagman K.D., Hambuch T., Rehm H.L. A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories // *Genet. Med.* – 2017. – Vol. 19, No. 5. – P. 575-582. <https://doi.org/10.1038/gim.2016.152>

22. Nkengasong J. N., Yao K., Onyebujoh P. Laboratory medicine in low-income and middle-income countries: Progress and challenges // *Lancet.* – 2018. – Vol. 391, No. 10133. – P. 1873-1875. [https://doi.org/10.1016/S0140-6736\(18\)30308-8](https://doi.org/10.1016/S0140-6736(18)30308-8)

23. Wang X., Xie C., Lu C. Identification and Analysis of Gene Biomarkers for Ovarian Cancer // *Genet. Test. Mol. Biomarker.* – 2024. – Vol. 28, No. 2. – P. 70-81. <https://doi.org/10.1089/gtmb.2023.0222>

24. Hamidi F., Gilani N., Arabi Belaghi R., Yaghoobi H., Babaei E., Sarbakhsh P., Malakouti J. Identifying potential circulating miRNA biomarkers for the diagnosis and prediction of ovarian cancer using machine-learning approach: Application of Boruta // *Front. Digit. Health.* – 2023. – Vol. 5. – Art. no. 1187578. <https://doi.org/10.3389/fdgh.2023.1187578>

АНДАТПА

МОЛЕКУЛАЛЫҚ ГЕНЕТИКА ЗЕРТХАНАСЫНДАҒЫ САПАНЫ БАҚЫЛАУ: ӘДЕБИЕТКЕ ШОЛУ

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Өзектілігі: Онкологиялық пациенттерде ерте диагностикалау мен болжамды бағалаудың тиімділігін арттыру үшін молекулалық биомаркерлерді анықтауды зертханалық жағдайларда сапаны қатаң бақылаумен біріктіру аса маңызды. Онкологиялық ауруларды зертханалық диагностикалауда дәлдік пен сапаны бақылау, нысаналы және дербестендірілген терапияны кеңінен енгізу аясында, ерекше мәнге ие болуда.

Зерттеу мақсаты – аналық без обырын диагностикалау мысалында молекулалық-генетикалық зертханаларда биомаркерді идентификациялау сапасын бақылауды бағалауға арналған жарияланымдарды шолу.

Әдістері: Осы зерттеу аясында жүргізілген жүйелі әдебиеттерге шолу 220 жазбаны анықтады, нәтижесінде 165 бірегей жарияланымдар алынды, оның ішінде 24 толық мәтінді мақала осы шолуға қосылды. Зерттеу жүйелі шолулар мен мета-талдаулар 2020 (PRISMA 2020) үшін артықшылықты есеп беру элементтері ұсынымдарына сәйкес жүргізілді.

Нәтижелері: Сапаны бақылау (Quality Control, QC) шараларын енгізу, соның ішінде калибрлеу, ішкі бақылау және біліктілікті тестілеу, Американдық патологтар колледжінің (CAP) ұсынымдарын пайдалана отырып жүргізілген жағдайда, қателіктердің санын айтарлықтай азайтады. Қаржыландырудағы шектеулерге қарамастан, Еуропалық молекулалық сапа желісі (EMQN) және CAP зертханалық тәжірибені ғаламдық деңгейде бағалауға бағытталған біліктілікті тестілеу бағдарламаларын ұсынады, бұл диагностикалық нәтижелердің үйлесімділігі мен сенімділігін қамтамасыз етеді.

Қорытынды: Клиникалық практикада молекулалық-генетикалық диагностикалық тесттердің дәлдігі мен сенімділігін қамтамасыз ету өте маңызды, әсіресе аналық без қатерлі ісігі сияқты ауруларда, онда нақты генетикалық талдау диагностикалық және емдеу стратегиясын анықтайды. Ерте диагностикалау мен дербестендірілген терапиядағы одан әрі жетістіктер жаңа технологияларды инновацияларды сенімді сапаны бақылау жүйесімен біріктіру арқылы жүзеге асырылуы мүмкін, бұл ақыр соңында пациенттерді емдеу нәтижелерін жақсартуға әкеледі. Осыған байланысты молекулалық-генетикалық тестілеудің, әсіресе аналық без қатерлі ісігін диагностикалауға арналған, стандартталған нұсқаулықтары мен стандартты операциялық процедураларын әзірлеу өзекті әрі қажетті міндет болып табылады.

Түйінді сөздер: Молекулалық-генетикалық тестілеу, сапаны бақылау (QC), BRCA1, BRCA2, полимеразалық тізбекті реакция (PCR), келесі ұрпақтың секвенирлеуі (NGS).

АННОТАЦИЯ

КОНТРОЛЬ КАЧЕСТВА В ЛАБОРАТОРИИ МОЛЕКУЛЯРНОЙ ГЕНЕТИКИ: ОБЗОР ЛИТЕРАТУРЫ

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Актуальность: Интеграция детекции молекулярных биомаркеров со строгим контролем качества в условиях лаборатории молекулярной генетики имеет ключевое значение для повышения эффективности ранней диагностики и оценки прогноза

у онкологических пациентов. Точность и контроль качества в лабораторной диагностике онкологических заболеваний приобретают особую значимость в связи с широким внедрением таргетной и персонализированной терапии.

Цель исследования – обзор публикаций, посвященных оценке контроля качества идентификации биомаркеров в лабораториях молекулярной генетики на примере диагностики рака яичников.

Методы: Систематический обзор литературы, проведенный в рамках данного исследования, выявил 220 записей, что привело к 165 уникальным публикациям, из которых 24 полнотекстовые статьи были включены в данный обзор. Исследование проводилось в соответствии с рекомендациями Preferred Reporting Items for Systematic Reviews and Meta-Analyses 2020 (PRISMA 2020).

Результаты: Внедрение мер контроля качества (Quality Control, QC), включая калибровку, внутренний контроль и тестирование квалификации, используя рекомендации Колледжа американских патологов (CAP), существенно снижает количество ошибок, несмотря на сохраняющиеся ограничения в финансировании. Европейская молекулярная сеть качества (EMQN) совместно с CAP предлагают программы тестирования квалификации, направленные на оценку лабораторной практики в глобальном масштабе, обеспечивая согласованность и надежность результатов тестирования.

Заключение: Обеспечение точности и надежности молекулярно-генетических диагностических тестов критически важно в клинической практике, особенно при заболеваниях, таких как рак яичников, где точный генетический анализ определяет стратегию диагностики и лечения. Дальнейшие достижения в раннем выявлении и персонализированной терапии могут быть достигнуты за счет интеграции новых технологических инноваций в рамках надежной системы контроля качества, что в конечном итоге приведет к улучшению результатов лечения пациентов. Следовательно, разработка стандартных руководств и стандартных операционных процедур для молекулярно-генетического тестирования, с особым акцентом на диагностику рака яичников, является насущной необходимостью.

Ключевые слова: молекулярно-генетическое тестирование, контроль качества, BRCA1, BRCA2, полимеразная цепная реакция (ПЦР), секвенирование нового поколения (NGS).

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