HUMAN GENETICS EXPERT OPINION

Court-ordered expert opinion | Medical liability

Failure to recommend BRCA1/2 testing in cases of hereditary predisposition | Late diagnosis of breast cancer | Liability of the treating gynecologist

Case Number	LG [City] - Ref. [XX] O [XXXX]/[YY] (Sample Case)
Client	Regional Court [City], [Civil Division], Order for the Taking of Evidence dated [Date]
Plaintiff	Ms. Sandra W. [fictitious], born [Date]
Defendant	Dr. Klaus P. [fictitious], specialist in gynecology and obstetrics, practicing in [City]
Expert	Prof. Dr. Christian T. Thiel, MBA
Qualifications	Specialist in Human Genetics; University Hospital Erlangen; 30 years of clinical and scientific experience; expert witness for courts and insurance carriers
Date	March 28, 2026
Key Issue	Did the defendant gynecologist violate the duty, based on the current state of medical science, to recommend human genetic counseling and BRCA1/2 testing, and would timely testing have prevented or detected the breast cancer diagnosed later?

Declaration pursuant to Sections 407, 407a of the German Code of Civil Procedure (ZPO)

Pursuant to the decision of the Regional Court of [City], the undersigned was appointed as a neutral expert. He declares that he has prepared this expert opinion impartially, to the best of his knowledge and belief, and without regard to any interests of the parties. He affirms that he has no personal or financial relationship with the parties. The expert opinion is based on the time of treatment by the defendant physician (ex ante standard) and assesses whether the state of the art and guidelines at that time would have warranted a diagnosis.

1. Court's Mandate and Questions

By order for the taking of evidence dated [Date], the Regional Court of [City] commissioned the undersigned to prepare a human genetics expert opinion on the following questions:

1. Based on the documented family history, was there an indication at the time of the plaintiff's initial contact with the defendant gynecologist to refer her to a human genetics counseling center and/or to initiate BRCA1/2 germline testing in accordance with the state of medical science and the relevant guidelines applicable at that time?

2. What guidelines and recommendations were in effect at the time of the initial consultation regarding risk assessment and the determination of indications for BRCA1/2 testing?
3. Would guideline-based testing have detected the subsequently diagnosed BRCA1 germline mutation and the breast cancer with a reasonable degree of probability at an earlier stage?
4. What preventive measures would have been medically indicated if BRCA1 carrier status had been confirmed at an early stage?
5. How should the causal relationship between the failure to establish an indication and the resulting harm (late diagnosis, severity of the disease) be assessed?

2. Investigation Materials and Methodology

2.1 Submitted documents

- Complete medical records of the defendant, Dr. P., a gynecologist (treatment period [date]–[date])
- Hospital medical records Breast Center [Hospital] (Diagnosis and treatment documentation for breast cancer)
- Molecular Genetic Report: BRCA1/2 Diagnostics (Date of Diagnosis: [Date]): BRCA1 pathogenicity c.5266dupC (p.Gln1756ProfsTer74), ACMG Class 5
- Family history questionnaire (completed by the plaintiff)
- Death certificates and hospital reports of affected relatives (if available)
- Complaint and Answer to the Complaint, including the parties' submissions
- Relevant guidelines and recommendations (S3 Guideline on Breast Cancer, AGO Recommendations, DGGG/DKG Guidelines on Hereditary Breast and Ovarian Cancer, AWMF Guideline 015/065)

2.2 Expert Standard

The ex-ante standard is decisive for assessing the duty of medical care: Which measures corresponded to the state of the art and applicable guidelines at the time of the plaintiff's initial contact with the defendant? The standard of comparison is a diligent specialist in gynecology and obstetrics who adheres to guidelines at the relevant time.

3. Facts

3.1 Clinical Course

The plaintiff, Ms. Sandra W., born [date], first presented herself to the defendant gynecologist, Dr. P., for a routine checkup in [year]. She was [age] years old at that time. During the initial consultation, the plaintiff reported the following family history, which—according to the medical records—was documented:

Documented family history at the time of the initial consultation [Year]
Mother: Breast cancer, diagnosed at age 41, died at age 48 Maternal aunt (mother's sister): Ovarian cancer, diagnosed at age 44 years
Maternal grandmother: Breast cancer, diagnosed at age 52
Plaintiff at the time of initial contact: [age] years old, no personal history of cancer

Although the plaintiff was aware of a family history of cancer, the gynecologist failed to take appropriate measures. Thus, neither was a referral to a human genetics counseling center arranged, nor were appropriate diagnostic tests performed to detect BRCA1/2 mutations. Furthermore, the plaintiff was not informed about the possibility of such diagnostic testing.

In the following years, the plaintiff was examined by the defendant at regular annual intervals. The clinical findings and the mammography results were unremarkable until the year [Year]. In [Month Year], the plaintiff, who was [Age] years old at the time of diagnosis, was diagnosed with invasive right-sided breast cancer (pT2, pN1, M0, G3, ER-positive, HER2-negative). Genetic testing was initiated, during which a so-called BRCA1 germline mutation (c.5266dupC, ACMG Class 5) was detected.

3.2 Chronology - Summary

Date	Event	Assessment
[Year]: Initial contact	Family history with 3 affected individuals (mother: breast, aunt: ovary, grandmother: breast) documented	**Indication for BRCA1/2 testing PRESENT - not acted upon**
[Year]–[Year]	Annual routine screening; no indication for genetic testing	**Repeated failure to act over several years**
[Year]	Breast cancer diagnosed (pT2 pN1 G3)	Injury
[Year]	BRCA1 mutation detected (c.5266dupC, Class 5)	Too late for preventive measures

4. Family history and risk assessment

4.1 Family history pattern

At the time of the initial consultation, the plaintiff's documented family history exhibits the following characteristics, which are typical of hereditary breast and ovarian cancer syndrome (HBOC) and are classified as a high-risk pattern according to all relevant guidelines:

- Breast cancer in the mother with early age at diagnosis (41 years) - Onset of the disease before age 45 is considered a strong BRCA risk factor
- Ovarian cancer in maternal aunt (age 44) - Ovarian cancer in a first-degree relative on the maternal line is a very strong BRCA indicator disease
- Breast cancer in the maternal grandmother (age 52) — third case of breast cancer in the maternal line
- Three affected individuals in the on line across three generations - Family tree findings with a clear vertical pattern of inheritance

According to the S3 guideline on breast cancer and the guidelines of the German consortia (Consortium for Hereditary Breast and Ovarian Cancer, DKG/DGGG/AWMF) in the version applicable at the time of treatment, clearly meets the criteria for a high-risk constellation with an indication for human genetic counseling and BRCA1/2 testing.

4.2 Quantitative Risk Assessment

Even without knowledge of the genetic findings, the plaintiff's a priori risk of developing the disease was significantly elevated based on the family history. Standardized risk models (BOADICEA, Tyrer-Cuzick) would have indicated a lifetime risk of breast cancer of [approx. 60–80%] for this family history—well above the threshold for intensive care measures and BRCA testing. The probability of a causative BRCA1/2 mutation in this scenario was [> 30–40%].

5. Relevant guidelines and indication criteria

5.1 Applicable guidelines at the time of the initial contact

At the time of the plaintiff's initial contact with the defendant ([year]), the following guidelines and recommendations, which are relevant for assessing the duty of medical care, were in effect:

- S3 Guideline on Breast Cancer (AWMF 032/045): Clear indication criteria for referral to a center for hereditary breast and ovarian cancer; already included in the versions from 2004 onward
- Guidelines of the German Consortium for Hereditary Breast and Ovarian Cancer (HBOC Consortium): Detailed inclusion criteria, including explicitly: two relatives with breast cancer (one of whom is under 50 years of age) OR one relative with breast cancer and one with ovarian cancer
- AGO Recommendations (Working Group on Gynecological Oncology): Annual updates with explicit BRCA indication lists for private practice gynecologists
- National Association of Statutory Health Insurance Physicians (KBV): Statutory health insurance entitlement to genetic testing in cases of relevant family history (Genetic Testing Act 2010, § 7 GenDG)

5.2 Indications according to the S3 guideline (at the time of treatment)

The S3 guideline explicitly listed the following as inclusion criteria for high-risk patients (simplified presentation):

Criterion according to the S3 guideline	Met at the time of the plaintiff's initial contact?	Assessment
At least 2 relatives with breast cancer, one of whom is under 50	Yes (mother: 41 years old; grandmother: 52 years old)	Met
At least 1 relative with ovarian cancer AND 1 with breast cancer	Yes (Aunt: ovarian; Mother + Grandmother: breast)	Met
Affected individuals across multiple generations of the same lineage	Yes (3 generations on the maternal side)	Met
Personal risk of developing the disease > 20–25% (risk model)	Yes (estimated 60–80%)	Met
Indication for referral / counseling	**DEFINITELY YES - by defendant**	**BREACH OF DUTY OF CARE**

6. Molecular genetic findings and evaluation

6.1 Detected mutation

BRCA1 germline mutation - Findings report [Date of diagnosis]
Gene: BRCA1 (Chromosome 17q21; NM_007294.4)
Variant: c.5266dupC (p.Gln1756ProfsTer74) - heterozygous
Variant type: Frameshift (duplication, leading to premature stop codon)
Classification: Pathogenic (ACMG Class 5) – one of the most common BRCA1 founder mutations worldwide
Disease risk for BRCA1 carriers: Lifetime risk of breast cancer approx. 72%; lifetime risk of ovarian cancer approx. 44% [1,2]

6.2 Classification of the variant

The identified variant c.5266dupC (formerly known as 5382insC) is one of the best-characterized pathogenic BRCA1 variants worldwide and has been known since the early 1990s. [1,3] It leads to a frameshift and a premature stop codon, causing a loss of function of the BRCA1 protein due to haploinsufficiency. As a tumor suppressor, BRCA1 is essential for DNA repair (homologous recombination); its loss of function leads to genomic instability and an increased risk of cancer. [1,2]

At the time of the plaintiff's initial contact with the defendant, this variant was listed as pathogenic in ClinVar, LOVD, and all relevant BRCA databases. **It is neither rare nor difficult to classify—it is among the longest-known and most well-documented BRCA1 mutations.**

7. Expert Analysis of Errors

7.1 Duty to provide a medical indication

The defendant gynecologist's duty of care in this case included:

1. Collection and evaluation of the family history regarding hereditary tumor diseases—this was carried out according to the medical records (documentation available)
2. Identification of a high-risk constellation based on the documented family history—this was mandatory according to current guidelines
3. Informing the patient about the increased familial risk and the possibility of genetic counseling and BRCA1/2 testing
4. Referral or at least a recommendation to consult a center for hereditary breast and ovarian cancer

The defendant physician did not document any of these steps (2–4) or—according to his own statement in the answer to the complaint—take them**. This constitutes a breach of the duty of medical care as defined by the state of scientific knowledge at the time.**

7.2 Was the indication clear or only borderline?

This point is decisive for the question of liability. If the family history had shown only a weak or borderline risk profile, one could speak of a reasonable margin of medical discretion. In the present case, there are three serious BRCA indicators:

- Breast cancer in the mother under the age of 45 (strongest single criterion)
- Ovarian cancer in a first-degree relative on the maternal side (strongly independent)
- Third breast cancer in the family tree—genealogical clustering

This combination is not borderline but **prototypical of the high-risk constellation, which has been listed as a mandatory indication for referral in all guideline versions since at least [year].** This is not a matter of discretion but a clear guideline requirement.

7.3 Hypothetical course of events had the guidelines been followed

Had the defendant physician acted in accordance with the guidelines, the following course of events would have occurred with a high degree of probability:

1. Referral to a human genetics counseling center or an HBOC center in [Year]
2. BRCA1/2 germline testing with a high probability in [Year]/[Year]
3. Detection of the BRCA1 c.5266dupC mutation—this mutation had been known and diagnosable for years
4. Classification as a high-risk carrier and initiation of enhanced screening: biannual breast MRI, biannual clinical examination, and, if necessary, increased frequency of mammography
5. If necessary, prophylactic salpingo-oophorectomy and/or risk-reduction counseling regarding prophylactic mastectomy—in BRCA1 carriers, prophylactic mastectomy reduces the risk by >90% [2,4]

6. With early intensive screening: If the breast cancer diagnosed later would have developed anyway, it would very likely have been detected earlier—as a T1 tumor rather than a T2, without lymph node involvement

8. Assessment of causation

8.1 Causality giving rise to liability

The causal link between the failure to establish an indication and the harm must be assessed according to the standard of preponderance of probability (in tort law: “with a probability bordering on certainty” or “highly probable,” depending on the harm).

For the assessment, a distinction must be made between two levels of harm:

Level of harm 1: Prevention of preventive measures

It is highly probable that, had the BRCA1 mutation been detected, the plaintiff would have considered a prophylactic bilateral salpingo-oophorectomy and possibly a prophylactic mastectomy. **Studies show that prophylactic mastectomy reduces the risk of breast cancer in BRCA1 carriers by over 90%. [2,4]** The failure to pursue this option constitutes an independent harm, consisting in the denial of the opportunity for risk reduction.

Level of Harm 2: Late Diagnosis Due to Lack of Intensive Screening

Even if one assumes that prophylactic surgery is uncertain, intensive screening in accordance with guidelines (MRI every 6 months) would have detected the breast cancer at an early stage with a high degree of probability. The mammography-MRI combination achieves a sensitivity of >90% for T1 tumors in BRCA1 carriers. [5,6] The pT2 status and lymph node involvement (pN1) documented at the time of diagnosis indicate that, had the tumor been detected early, it could have been diagnosed as T1 and node-negative, which would have significantly improved treatment intensity, prognosis, and chances of survival.

8.2 Questions of burden of proof

Note to the Court: According to the Federal Court of Justice’s case law on medical liability (including BGH VI ZR 252/20), the plaintiff may be entitled to a relaxation of the burden of proof in cases of grossly negligent failure to fulfill a duty to diagnose or provide informed consent. It is up to the court to determine whether gross medical malpractice occurred in this case; from an expert perspective, it should be noted that the indication for referral was clear and, in accordance with guidelines, mandatory.

9. Conclusion / Expert Assessment

Key Expert Findings

1. Breach of the duty of care: The defendant gynecologist violated the clearly established obligation, based on the current state of scientific knowledge and applicable guidelines (S3 Breast Cancer Guideline, HBOC Guidelines), to indicate the need for human genetic counseling and BRCA1/2 testing. The documented family history clearly met the inclusion criteria for a high-risk constellation at the time of the initial consultation, without any borderline cases.
2. No discretion: The constellation (breast cancer in the mother < 45 years, ovarian cancer in the aunt, breast cancer in the grandmother) is prototypical for a high-risk BRCA indication and is not identified as a discretionary case in any version of the guidelines from the relevant period.
3. Causal connection: Had the defendant acted in accordance with the guidelines, a BRCA1 mutation would most likely have been detected at an early stage. This would have enabled preventive measures (prophylactic surgeries, intensive screening) and/or resulted in an early diagnosis of breast cancer at stage T1 node-negative instead of T2 N1.
4. Damages: As a result of the omission, the plaintiff lost the right to know her genetic risk status, the opportunity to take risk-reducing measures, and the chance for an early diagnosis of breast cancer. These damages are adequately causally attributable to the defendant's omission.

The foregoing expert findings are rendered to the best of my knowledge and belief, impartially and conscientiously, in accordance with Sections 407 and 407a of the German Code of Civil Procedure (ZPO).

9a. Current treatment recommendations for the plaintiff

Regardless of the outcome of the legal dispute, the following measures are currently indicated for the plaintiff from a human genetic perspective:

- Intensive follow-up care for breast cancer according to standard oncological protocols
- Ophthalmological and gynecological monitoring (risk of ovarian cancer with BRCA1 approx. 44%)
- Discussion of prophylactic salpingo-oophorectomy following completion of family planning
- Human genetic family counseling: first-degree relatives (sisters, daughters) have a 50% risk of a BRCA1 mutation; offer predictive testing
- Psycho-oncological care: The combination of a BRCA1 mutation and cancer poses a significant psychological burden

10. References

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7. S3 Guideline on Early Detection, Diagnosis, Treatment, and Follow-up of Breast Cancer. AWMF Registry Number 032-045OL. Current version and previous versions from [year].

8. Guideline on Hereditary Breast and Ovarian Cancer. German HBOC Consortium. AWMF 015/065. All relevant versions.

9. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants. Genet Med. 2015;17(5):405-424.

10. Robson M, Im SA, Senkus E, et al. Olaparib for Metastatic Breast Cancer in Patients with a Germline BRCA Mutation. N Engl J Med. 2017;377(6):523-533.

Erlangen, March 28, 2026

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