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Breast cancer: Determining the genetic profile from ultrasound-guided percutaneous biopsy specimens obtained during diagnostic workups☆

J.A. López Ruiz[a,*], I. Zabalza Estévez[b], J.A. Mieza Arana[c]

[a] *Diagnóstico e Intervencionismo de Mama, Grupo Preteimagen, Bilbao, Spain*
[b] *Jefe del Servicio de Anatomía Patológica, Hospital de Galdakao-Usansolo, Usansolo, Bizkaia, Spain*
[c] *Ginecología Oncológica, Hospital Universitario de Cruces-Barakaldo, Bizkaia, Spain*

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Breast cancer;
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Abstract
Objectives: To evaluate the possibility of determining the genetic profile of primary malignant tumors of the breast from specimens obtained by ultrasound-guided percutaneous biopsies during the diagnostic imaging workup.
Material and methods: This is a retrospective study in 13 consecutive patients diagnosed with invasive breast cancer by B-mode ultrasound-guided 12 G core needle biopsy. After clinical indication, the pathologist decided whether the paraffin block specimens seemed suitable (on the basis of tumor size, validity of the sample, and percentage of tumor cells) before sending them for genetic analysis with the MammaPrint® platform.
Results: The size of the tumors on ultrasound ranged from 0.6 cm to 5 cm. In 11 patients the preserved specimen was considered valid and suitable for use in determining the genetic profile. In 1 patient (with a 1 cm tumor) the pathologist decided that it was necessary to repeat the core biopsy to obtain additional samples. In 1 patient (with a 5 cm tumor) the specimen was not considered valid by the genetic laboratory. The percentage of tumor cells in the samples ranged from 60% to 70%. In 11/13 cases (84.62%) it was possible to do the genetic analysis on the previously diagnosed samples.
Conclusion: In most cases, regardless of tumor size, it is possible to obtain the genetic profile from tissue specimens obtained with ultrasound-guided 12 G core biopsy preserved in paraffin blocks.

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* Corresponding author.
E-mail address: jalopez@preteimagen.com (J.A. López Ruiz).

PALABRAS CLAVE
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Cáncer de mama: determinación del perfil genético a partir de la biopsia percutánea ecoguiada diagnóstica

Resumen
Objetivos: Evaluar la posibilidad de obtener el perfil genético de los tumores primarios malignos de la mama a partir de las muestras obtenidas mediante la biopsia percutánea ecoguiada realizada durante el proceso diagnóstico.
Material y métodos: Estudio retrospectivo sobre 13 pacientes consecutivas diagnosticadas de cáncer infiltrante de mama mediante biopsia ecoguiada (Modo B) con aguja gruesa (BAG) de calibre 12 G. Tras indicación clínica, el anatomopatólogo determinó, sobre los bloques de parafina, la aparente idoneidad de las muestras, antes de enviarlas al laboratorio para análisis genético con la plataforma MammaPrint®. Se han evaluado los siguientes aspectos: tamaño tumoral, validez de la muestra y porcentaje de células tumorales.
Resultados: El tamaño ecográfico tumoral osciló entre 0,6 cm y 5 cm. En 11 pacientes la muestra conservada se consideró ''válida'' y apta para la determinación del perfil genético. En una paciente (tamaño tumoral de 1 cm) y a juicio del anatomopatólogo, hubo que repetir la BAG para obtener muestras adicionales. En otra paciente (tamaño tumoral de 5 cm), la muestra no fue considerada ''válida'' por el laboratorio genético. El porcentaje de células tumorales, entre las muestras válidas, osciló entre el 60% y 70%. En 11 de 13 (84,62%) casos fue posible el análisis genético a partir de las muestras diagnósticas previas.
Conclusión: Resulta posible obtener el perfil genético en la BAG ecoguiada con agujas de 12 G, a partir de las muestras diagnósticas conservadas en bloques de parafina, en la mayoría de los casos, independientemente del tamaño tumoral.

Introduction

Breast cancer is a heterogeneous disease; therefore, knowledge of the so-called profile (''fingerprint'', ''signature'') of tumor gene expression (genotype) has become an important tool when it comes to establishing prognosis and planning therapy in patients affected by breast cancer.[1–5]

This information is usually obtained through surgical tumor samples. However, several situations occur in which it is necessary to obtain such information non-surgically (especially if implementing neoadjuvant therapy is being considered) through breast percutaneous biopsy (PB).

There are not a lot of specific bibliographic references regarding the exclusive use of the paraffin blocks resulting from diagnostic PB; therefore, our goal is to determine whether diagnostic PB samples are adequate to evaluate the tumor genetic profile through the genetic platform MammaPrint®.

Material and methods

Retrospective study of 13 consecutive patients between January 2012 and September 2015. Inclusion criterion: diagnosis of infiltrating breast cancer through PB later confirmed through surgery, with a clinical indication to determine the genetic signature from the samples obtained during the diagnostic process. No exclusion criteria have been considered.

The protocols established in our centers to access information in the medical histories have been followed in order to be able to publish it. All patients gave their written informed consent for the determination of the genetic profile. It was not necessary to request authorization from the Hospital Ethics Committee since no resource or procedures different from those already registered in everyday practice were used.

The percutaneous biopsies corresponded to a B-mode ultrasound-guided biopsy with a thick needle on masses suspicious of malignancy and were conducted by the same radiologist using a 12 G Trucut needles (Magnum®, BARD). The number of samples per patient ranged from 3 to 4 and was obtained by selecting the ''long'' (22 mm) advance and that of different areas of the lesion following our usual protocol.

After the clinical request, the pathologist would review the paraffin blocks and determine the suitability of the samples, before referring them to perform the genetic workup using the MammaPrint® software (Agendia Inc., Amsterdam, The Netherlands). The sample was considered valid whenever the genetics lab would say so, considering the specimen suitable in the presence of at least 30% tumor cells.[6] After the genetic analysis, the sample was returned to the anatomical pathological lab.

The genetic analysis classified the patients into two groups, ''high'' or ''low'' based on the level of risk of developing recurrence/metastatic disease.

The following aspects of each case were evaluated: tumor size, sample validity and percentage of tumor cells.

Results

Table 1 shows the list of cases with the patients' ages, tumor size, percentage of tumor cells in the samples analyzed and level of risk obtained in the analysis.