# Analysis of clinical pathological association with somatic mutations through next-generation sequencing

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## Pupose

The next generation sequencing technology has the advantages of high speed, high throughput and high accuracy. Because of these advantages, it is used in various cancer fields. Several gene pannels have been applied to breast cancer to assess risk and determine treatment direction accordingly. The purpose of this study was to improve the prognosis and future treatment of patients with breast cancer by applying NGS.

## Method

From January 2018 to December 2018, we studied patients who underwent surgery at Kosin University Gospel Hospital. The study patients were from stage 1 to stage 3 of breast cancer. Patients who were not able to undergo surgery or who had more than stage 4 patients were excluded. This study included patients who underwent Neo-systemic therapy(NST). NGS was performed postoperatively. And in patients who underwent NST, NGS proceeded to pre-chemotherapy specimens.

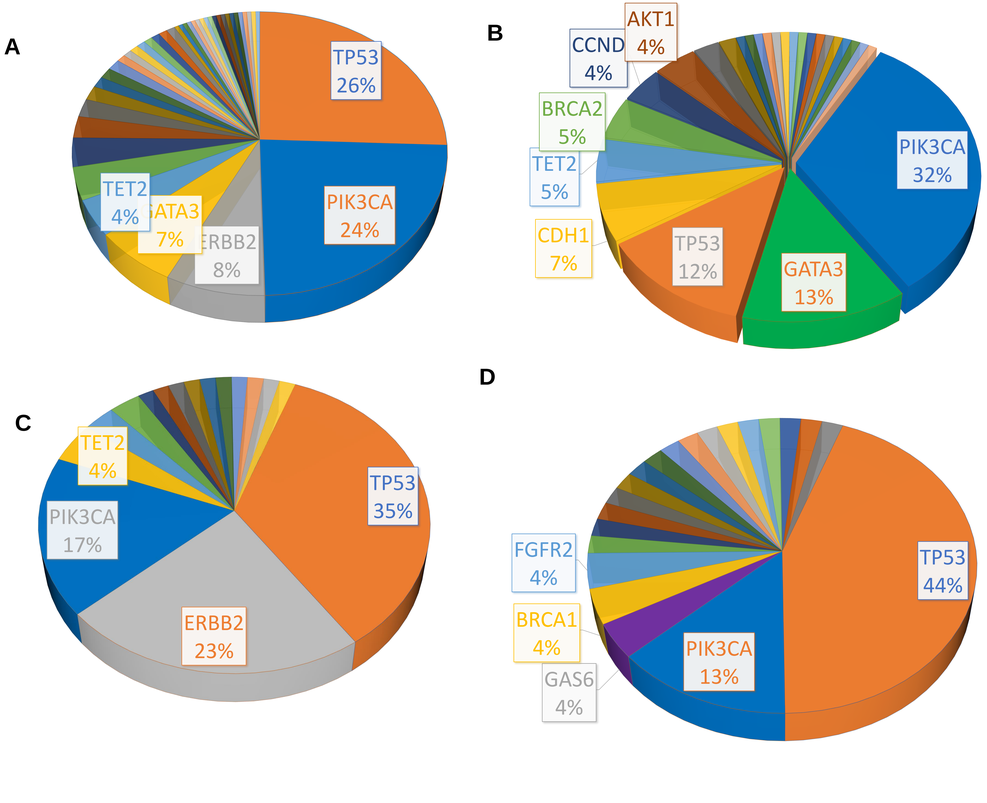


Fig. 1 : A :Somatic mutation for breast cancer patients.(N=160, total mutation number : 242),
B : Luminal type, C : Her2 type, D : TNBC

## Result

The expression of somatic mutation was different for each type of breast cancer. Most of them have been observed to have more than two mutations. It shows the expression ratios of each gene in figure 2. Overall, TP53, PIK3CA, and ERBB2 showed high expression frequencies. figure1 shows the frequency of mutation incidence frequent in each type of patient.

## Conclusion

Various types of somatic mutations are also expressed in breast cancer, and they are different according to each type. These various manifestations may be associated with the prognosis of breast cancer. Further studies are needed to determine for them.

| Characteristic | Status | No NST(110) | |
|---|---|---|---|
| Age | (mean, range) | 54.29(35-86) | |
| T stage | T1 or 1mic | 69 | 62.7% |
| | T2 | 37 | 33.6% |
| | T3 | 4 | 3.6% |
| N stage | N0 | 89 | 80.9% |
| | N1 | 17 | 15.4% |
| | N2 | 2 | 1.8% |
| | N3 | 2 | 1.8% |
| Grade | G1 | 23 | 20.9% |
| | G2 | 54 | 49.0% |
| | G3 | 31 | 28.1% |
| | unknown | 2 | 1.8% |
| ER | POSITIVE | 75 | 68.1% |
| | NEGATIVE | 35 | 31.8% |
| PR | POSITIVE | 72 | 65.4% |
| | NEGATIVE | 38 | 34.5% |
| HER2 | POSITIVE | 20 | 18.1% |
| | NEGATIVE | 90 | 81.8% |
| Ki67 | <10 | 44 | 10.0% |
| | 10-20 | 23 | 20.9% |
| | >20 | 43 | 39.0% |

Table 1. Characteristics of patients not receiving neo-systemic therapy.

| Subtype | Total : 160 |
|---|---|
| Luminal A | 54(34%) |
| Luminal B | 38(24%) |
| HER2 | 33(21%) |
| TNBC | 33(21%) |
| NST(Neo-systemic therapy) | 50(31%) |

Table 3. Subtype analysis for total patients.

| Characteristic | Status | NST(50) | |
|---|---|---|---|
| Age | (mean, range) | 50.04(34-66) | |
| cT stage | T1 | 4 | 8% |
| | T2 | 28 | 56% |
| | T3 | 14 | 28% |
| | Unknown | 4 | 8% |
| cN stage | N0 | 13 | 26% |
| | N1 | 23 | 46% |
| | N2 | 10 | 20% |
| | N3 | 1 | 2% |
| | Unknown | 3 | 6% |
| Pre NST Nuclear Grade | G1 | 2 | 4% |
| | G2 | 30 | 60% |
| | G3 | 8 | 16% |
| | unknown | 10 | 20% |
| ypT stage | No residual or Tis | 15 | 30% |
| | T1 or 1mic | 14 | 28% |
| | T2 | 17 | 34% |
| | T3 | 2 | 4% |
| | Unknown | 2 | 4% |
| ypN stage | N0 | 28 | 56% |
| | N1 | 14 | 28% |
| | N2 | 4 | 8% |
| | N3 | 2 | 4% |
| | Unknown | 2 | 4% |
| ypGrade | G1 | 4 | 8% |
| | G2 | 21 | 42% |
| | G3 | 9 | 18% |
| | unknown | 16 | 32% |
| ER | POSITIVE | 26 | 52% |
| | NEGATIVE | 24 | 48% |
| PR | POSITIVE | 15 | 30% |
| | NEGATIVE | 35 | 70% |
| HER2 | POSITIVE | 13 | 26% |
| | NEGATIVE | 37 | 74% |
| Ki67 | <10 | 20 | 40% |
| | 10-20 | 9 | 18% |
| | >20 | 21 | 42% |

Table 2. Characteristics of patients receiving neo-systemic therapy.

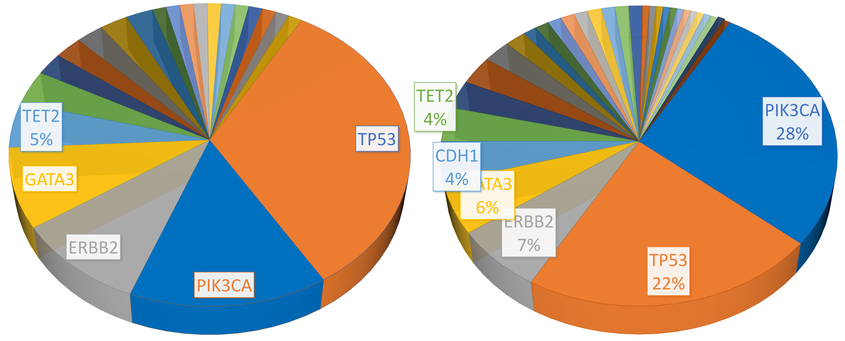


Fig. 3 : A :Somatic mutation for breast cancer patients receiving NST(N=50),
B: No receiving NST