# Whole Exome Sequencing

Whole Exome Sequencing provides a cost-effective alternative to whole genome sequencing, as it targets only the protein coding region of the human genome responsible for a majority of known disease-related variants. Whether you are conducting studies in rare mendelian disorders, complex disease, cancer research, or human population studies, Novogene's comprehensive human whole exome sequencing (hWES) service provides a high-quality, affordable, and convenient solution.

**Why Novogene?**

**Extensive experience with over 3000 projects**

**Industry-leading data quality guarantee**

**In house pipeline to meet different analysis requirement**

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## Sample requirements

| Sample Type | Amount | Volume | Concentration | Purity |
|---|---|---|---|---|
| Genomic DNA | ≥ 0.4 μg | ≥ 20 μL | ≥ 20 ng/μL | OD260/280=1.8-2.0<br>No degradation, no contamination |
| FFPE DNA | ≥ 0.8 μg | ≥ 20 μL | ≥ 20 ng/μL | Fragments should be longer than 1000 bp. |
| cfDNA/ctDNA | ≥ 0.05 μg | ≥ 20 μL | ≥ 20 ng/μL | Fragments should be in multiples of 170bp,<br>no genomic contamination |

## Sequencing parameters

| Exome Capture Strategy | Platform | Read length | Data quality |
|---|---|---|---|
| Agilent SureSelect Human All Exon V6 | Illumina NovaSeq 6000 | Pair-end 150bp | Q30 ≥ 85% |

## Publications using Novogene's expertise

**Cancer Research,** 2019.
Clonal Mutations Activate the NF-κB Pathway to Promote Recurrence of Nasopharyngeal Carcinoma

**Nature Communication,** 2019.
Recurrent GNAQ mutation encoding T96S in natural killer/T cell lymphoma

**Cancer Research,** 2019.
Multiregion Sequencing Reveals the Genetic Heterogeneity and Evolutionary History of Osteosarcoma and Matched Pulmonary Metastases

**Nature Communication,** 2018.
Copy number variation: A prognostic marker for young patients with squamous cell carcinoma of the oral tongue

**Scientific Reports,** 2018.
Csde1 binds transcripts involved in protein homeostasis and controls their expression in an erythroid cell line

**PLOS ONE,** 2018.
Strap associates with Csde1 and affects expression of select Csde1-bound transcripts

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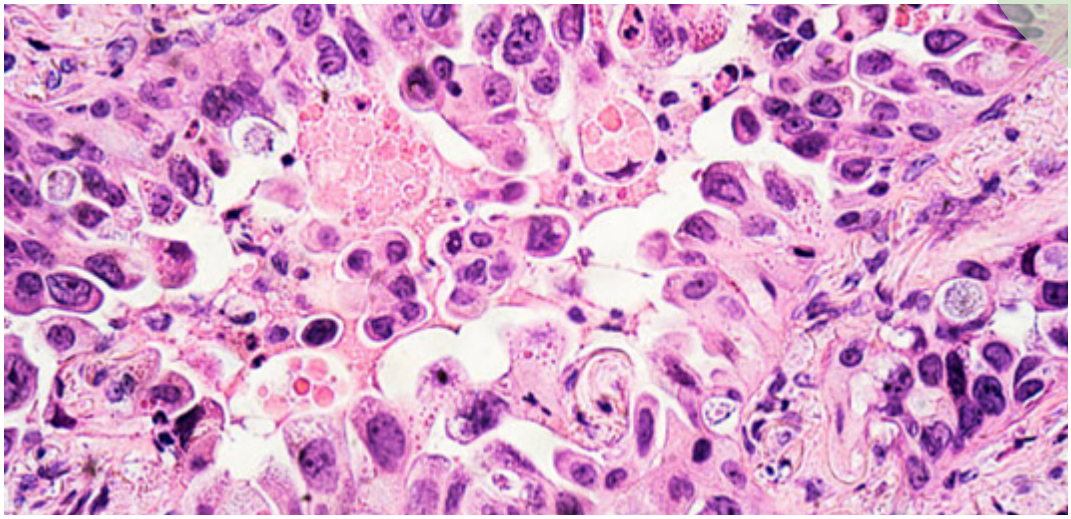
Case study

# Multiregion Sequencing Reveals the Genetic Heterogeneity and Evolutionary History of Osteosarcoma and Matched Pulmonary Metastases.

Wang et al., 2018. American Association for Cancer Research. DOI: 10.1158/0008-5472.CAN-18-1086



**Research objective:**
To investigate the dynamic evolutionary process and temporospatial heterogeneity of metastatic pulmonary osteosarcoma, to understand differences from primary tumors and provide insights for diagnosis and treatment of pulmonary metastasis.

**Sample collection:**
Samples were collected from patients that were diagnosed with osteosarcoma and developed pulmonary metastasis during follow-up. All samples (n=86) were utilised for WES.

**Sequencing strategy:**
PE150 on Illumina HiSeq platform.

**Data amount:**
>81Gb raw data generated with >99% coverage.

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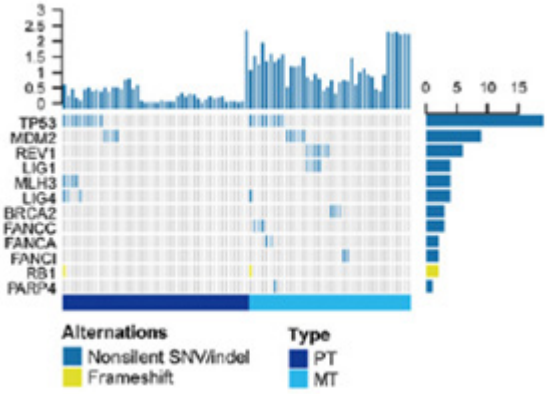
## Results

Osteosarcoma is the most common primary bone malignancy, and the lung is the most frequent site of metastasis. This study showed that these metastatic tumours exhibited higher tumour mutational burden (TMB) and genomic instability compared to primary tumours.

Mutated genes were enriched in the PI3K-Akt pathway in early and late stage evolution and the MAPK pathway at the metastatic stage. DNA damage response (DDR) genes were found to be deficient in metastatic tumours compared to primary tumours, confirming a previous hypothesis that DDR deficiency correlates with TMB, genomic instability, and osteosarcoma metastasis.

Linear evolution is the most common progression pathway from primary to metastatic tumour, with the assumption that only tumours with a genetic advantage can grow and disseminate, highlighting the importance of treating trunk events as early as possible. Four cases in this study showed a parallel mechanism of metastasis in osteosarcoma, a much less common mechanism, originating from independent tumours at a much earlier stage of tumorigenesis. This could be an important clinical application of this work for diagnosis and treatment.

**Figure 1**
Top: the TMB in each sample. Centre: the heatmap shows alterations of DDR pathway–related genes in each sample, including non-silent SNVs/Indels (blue) and frameshifts (yellow). Right: the histogram presents the number of alterations in every gene. Bottom: the bar presents the primary tumor (dark blue) samples and metastatic tumor (light blue) samples.




## Conclusions

This study highlighted the genetic heterogeneity between primary and metastatic pulmonary osteosarcomas. The detection of the dynamic evolution patterns in different patients was the first of its kind and provided insights into potential new diagnosis and treatment strategies.

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