International Agency for Research on Cancer

World Health Organization

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# Rare Disease Day 2024: the IARC Rare Cancers Genomics Team

# Questions and Answers (Q&A)

Rare cancers are cancer types with fewer than 6 newly diagnosed cases per 100 000 people per year. Although individually these cancer types occur rarely, taken together, rare cancer types account for about 25% of all cancer diagnoses. The lack of data on rare cancers results in slow progress in understanding the biology of these tumours. This contributes to poorer outcomes: the 5-year overall survival rate for rare cancers (~49%) is significantly lower than that for more common cancers (~63%).

At the International Agency for Research on Cancer (IARC), the Rare Cancers Genomics Team leads projects to improve the understanding of rare cancers. One example is the lungNENomics project, which aims to improve the diagnosis and clinical management of lung neuroendocrine tumours. To mark Rare Disease Day, scientists from the Rare Cancers Genomics Team answer questions about this project.

### 1. Why is it important to study lung neuroendocrine tumours?

Currently, pathologists and researchers classify lung neuroendocrine tumours into low-grade “typical” tumours and intermediate-grade “atypical” tumours. Whether a tumour is classified as typical or atypical informs both patient prognosis and treatment decisions, because atypical tumours tend to be faster-growing and more aggressive.

Distinguishing between typical and atypical tumours can be difficult, even for experts. As a result, the current classification system fails to accurately identify typical tumours that develop aggressively and atypical tumours that progress more slowly. This leads to unexpected recurrence for some patients and stressful, expensive, and unnecessary long-term monitoring for others. If we can uncover the signs that actually show which tumours are more aggressive, we can close this knowledge gap to give clarity to doctors and improve treatment for patients.

### 2. Which innovative technologies have you used to address the knowledge gap?

The Rare Cancers Genomics Team at IARC has adapted artificial intelligence algorithms for their application in tumour diagnostics to uncover currently unknown differences between aggressive and non-aggressive tumours.

Whether a lung neuroendocrine tumour is classified as typical or atypical depends on the number of cell divisions and the presence of dying cells. The question of how to estimate this proliferative activity, and in particular the optimal threshold for separating typical tumours from atypical tumours, has been debated for more than 30 years, leading to a substantial waste of resources.

The Rare Cancers Genomics Team has collected data from a large cohort of patients whose tumours have been reviewed by a panel of six expert pathologists. These data and the application of original statistical methods enable the team to examine the strengths and limitations of the current diagnostic criteria, while exploring the use of alternative markers to improve diagnostic accuracy.

### 3. What were the findings, and what are the next steps?

The researchers in the LungNENomics project have highlighted the limitations of how current criteria differentiate between typical and atypical tumours. Using original methods and artificial intelligence, they have found new associations between tumour aggressiveness, morphological features, and previously reported molecular groups.

Overall, these findings open a new avenue towards a more clinically relevant classification of lung neuroendocrine tumours, based on both what they look like and their molecular characteristics, which could go on to improve diagnosis for patients.

**For more information, please contact**

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