SARKOMFÖRENINGEN

Whole Genome Sequencing

A Paradigm Shift in the Fight Against Sarcoma and Other Rare Cancers

In 2022-2023, a study was conducted at Karolinska University Hospital in Stockholm, using whole-genome sequencing, WGTS, on **200** patients with suspected sarcoma. The study was led by pathologist and associate professor Felix Haglund de Flon, with strong support from the Swedish Sarcoma Association.

Some of the findings:

In more than **10%** of cases , the previous diagnosis was revised, allowing for correct treatment, which in turn increases survival chances. This reduces the risk of both over- and under-treatment.

6% of the patients had a tumour which was initially believed to be malignant but was actually benign. This allowed patients to go from being cancer patients returning to their lives and planning for the future after a more modest surgery.

15% of patients with high- grade sarcoma which currently lack an efficient treatment had targetable mutations. Many patients thereby avoid ineffective chemotherapy, and in some cases, limbs that would otherwise have been amputated can be saved.

What were the study's consequences?

The hospital immediately decided to introduce whole-genome sequencing, WGTS, as part of the clinical routine for all suspected sarcomas.

What are the Sarcoma Association's recommendations after the project?

- All patients with suspected sarcoma and other rare cancers should request whole-genome sequencing from their healthcare provider.
- All sarcoma centres should adopt whole-genome sequencing as the clinical standard for suspected sarcomas.