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MedGenome unveils Indiaâ??s first study on clinical impact of RNA-based NGS in sarcoma diagnosis

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Represents a major advancement in establishing evidence-based precision diagnostics for sarcomas



On the occasion of World Health Day, Bengaluru-based MedGenome has announced the outcomes of India's first prospective study assessing the clinical utility of RNA-based next-generation sequencing (NGS) for soft tissue and bone sarcoma diagnosis.

Aligning with this year's theme *"Together for health. Stand with science,"* the study highlights the importance of science-led, collaborative approaches in strengthening access to accurate diagnosis and improving cancer care.

This study, conducted in collaboration with India's leading hospitals between 2022 and 2025 and published in the Journal of Global Oncology, has demonstrated that introducing targeted RNA sequencing into a multidisciplinary team (MDT) workflow can substantially enhance diagnostic clarity and aid treatment decisions for Sarcomas, especially in patients with ambiguous or complex presentations.

For this study, 68 patients with unclear diagnoses based on their pathological indications were considered for evaluation after an MDT review. All samples were analysed with MedGenome's CAP-accredited indigenous RNA fusion panel, which is custom-designed for sarcomas and implemented in concordance with the WHO 2021 classification.

The study showed that the RNA-based NGS provided clinically informative findings in half of all evaluated cases. Notably, diagnostic refinement or reclassification could be achieved in over 40% of patients, while treatment decisions changed in more than a quarter of cases according to molecular results.

The study also highlights the opportunity of implementing cost-effective genomic testing in low and middle-income countries. MedGenome's sarcoma panel enables the detection of both canonical and ultra-rare gene fusions, many of which could have gone undetected by universal testing methods. By integrating these results into MDT discussions, clinicians can avoid ineffective chemotherapy regimens, confirm rare histologic subtypes, and make more informed therapeutic decisions.

Speaking about the importance of the research, an author of the study Charu Bahl, PhD, General Manager - Scientific Affairs, MedGenome Labs Ltd, said, "Sarcomas often sit at the crossroads of overlapping morphology and unpredictable clinical behaviour. For a large proportion of patients, conventional tools such as Histopathology, Immunohistochemistry (IHC), or Fluorescence In Situ Hybridization (FISH) are not enough to arrive at a definitive diagnosis. This prospective study distinctly shows that RNA-based NGS offers the resolution we need to classify difficult tumours correctly and, in many cases, course-correct treatment plans before precious time is lost."