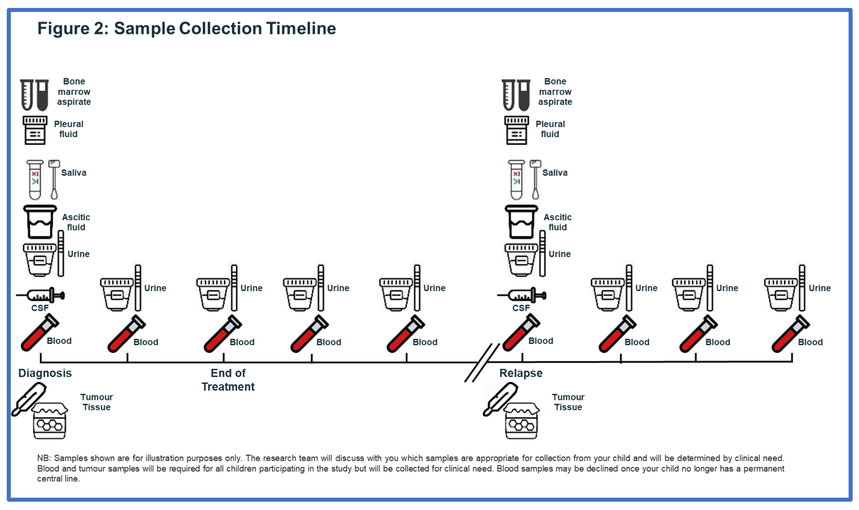
Liquid biopsies multi-modal analysis correlates with disease burden and response to treatment in paediatric cancer.

Paola Angelini*, Carolin Sauer*, Pooja Balasubramanian, Courtney Himsworth, Marilena Nicolaidou, Laura Rey Portela, Nicholas Tovey, Reda Stankunaite, Deborah Hughes, Joanne Stockton, Jake Micallef, Michael Hubank, Louis Chesler#, Andrew D. Beggs#, Isidro Cortes-Ciriano#, John Anderson#.
*equally contributed
senior authors

Background/Aims:

- The analysis of cfDNA for the detection of ctDNA offers a minimally invasive tool for diagnosis.
- New technologies provide additional capabilities for detection of ultra-low template reads, methylation profiles and nucleosome signatures, in addition to somatic copy number aberrations (SCNAs) and single nucleotide variants (SNV).
- We compared results by multi-modal analysis of plasma cfDNA and correlated with clinical information from paediatric cancer patients.



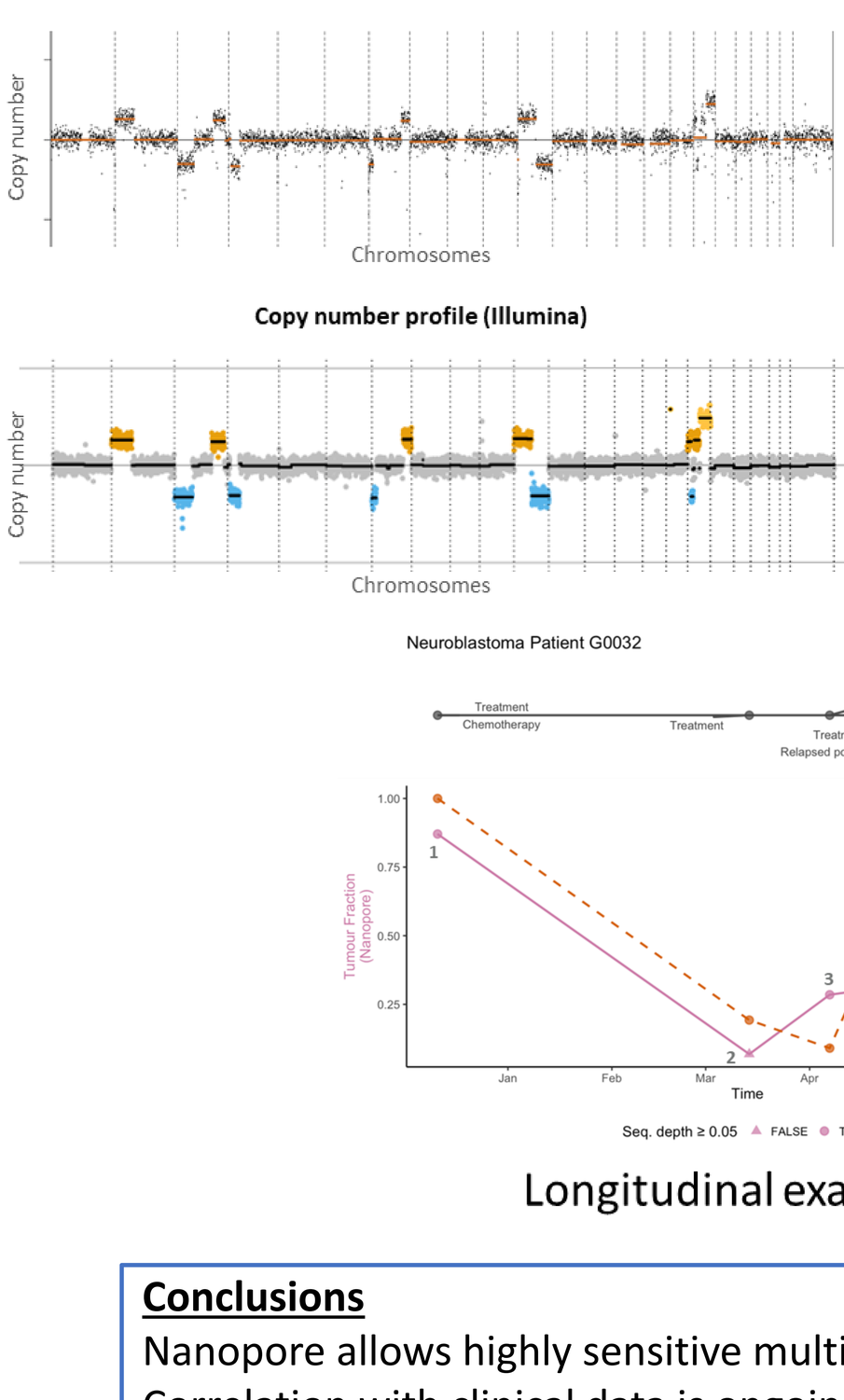
Patients and Methods:

- All patients age 0-24 with diagnosis of paediatric solid tumours were eligible.
- Prospective serial collection of biological fluids (blood, CSF, bone marrow).
- ctDNA was extracted from plasma and 25-50 ng of ctDNA were analyzed by nanopore and Illumina NGS panel.

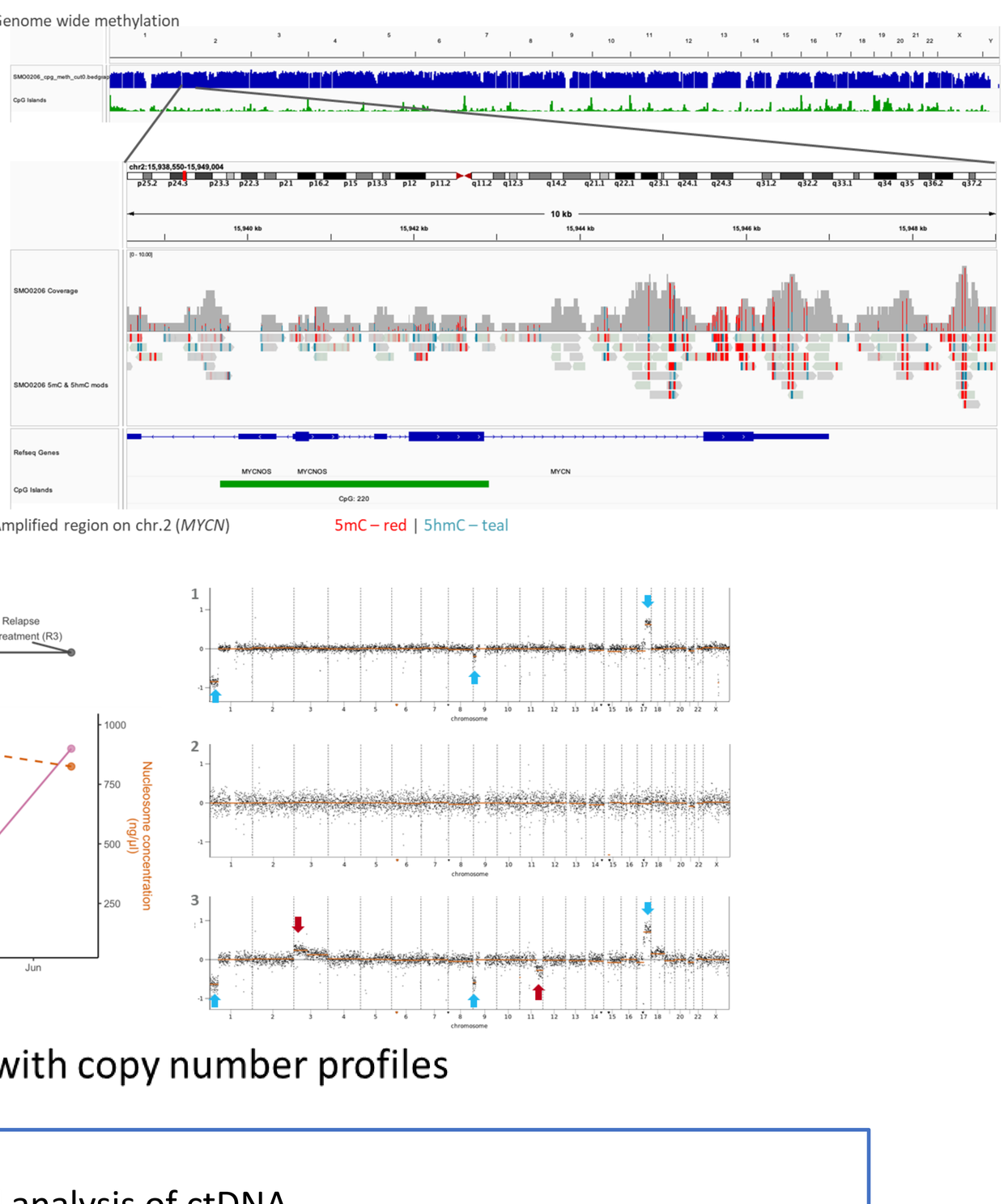
	N	%
Total patients	267	
Gender Male	150	56
Female	117	44
Diagnosis		
NB	98	37
RMS	28	10
Other sarcoma	23	9
Hepatoblastoma	7	3
Wilms tumour	13	5
Others	98	37

Results:

Comparison Illumina vs Nanopore



Detection of 5mC and 5hmC methylation from cfDNA



Amplified region on chr.2 (*MYCN*) 5mC – red | 5hmC – teal

Longitudinal example with copy number profiles

Conclusions

Nanopore allows highly sensitive multi-modal analysis of ctDNA.
Correlation with clinical data is ongoing.

Paola.angelini@nhs.net