positive cases. TMAs included 535 cases reported within our institution (DHM Pathology) over a 10 year period. Four cases showed moderate to strong positivity with ALK immunohistochemistry. Cases included a pleomorphic rhabdomyosarcoma (FOXO1-/SS18-/EWSR1-/MDM2-; 1/11), metastatic Ewing's sarcoma (EWSR1 FISH-proven; 1/16), an undifferentiated pleomorphic sarcoma (1/68) and uterine myxoid leiomyosarcoma (1/118). No significant staining was identified in myxofibrosarcomas ($n=97$), well-differentiated liposarcomas (LPS)/atypical lipomatous tumours ($n=95$), dedifferentiated LPS ($n=39$), myxoid LPS ($n=28$), angiosarcomas ($n=17$), synovial sarcomas ($n=17$) and pleomorphic LPS ($n=11$). ALK gene fusion analysis is pending but this preliminary data confirms the high specificity and sensitivity of the ALK antibody with negative stain in 99.25% of cases, restricting the necessity of molecular testing to 0.75% of cases. If ALK translocations are confirmed, this broadens the range of mesenchymal tumours in which ALK rearrangements may be present.

CLINICAL IMPLICATIONS OF DISCORDANT MASSARRAY AND SANGER SEQUENCING RESULTS IN CYSTIC FIBROSIS NEWBORN SCREENING

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Background: Agena's MassArray platform is used for high throughput genotyping in our service. This method combines primer extension termination polymerase chain reaction (PCR) with mass spectrometry (MADI-TOF) and proprietary genotype calling algorithms.

Method: The Agena iPLEX CFTR 74 v3 off-the-shelf assay is used as a 'common variant screen' for cystic fibrosis (CFTR) genotyping, including infants with an immunoreactive trypsin in the top 1% on newborn blood spot screening. Variants detected by this assay are not routinely confirmed by Sanger sequencing.

Case report: Cascade testing identified a variant called by the MassArray platform as c.2175dupA to be discordant with capillary electrophoresis sequencing, which identified this as c.2173G>A. Many of the variants on this screening panel are rare in our experience. The pathogenic c.2175dupA variant had not been previously detected in clinical testing in our service. The c.2173G>A variant is classified as a variant of unknown significance and would not usually be reported in a newborn screening patient.

Discussion: A primer extension product with a specific mass to charge ratio can detect different PCR products. The presence of rare SNPs is listed as a method limitation, but in this Case, resulted in variant re-classification.