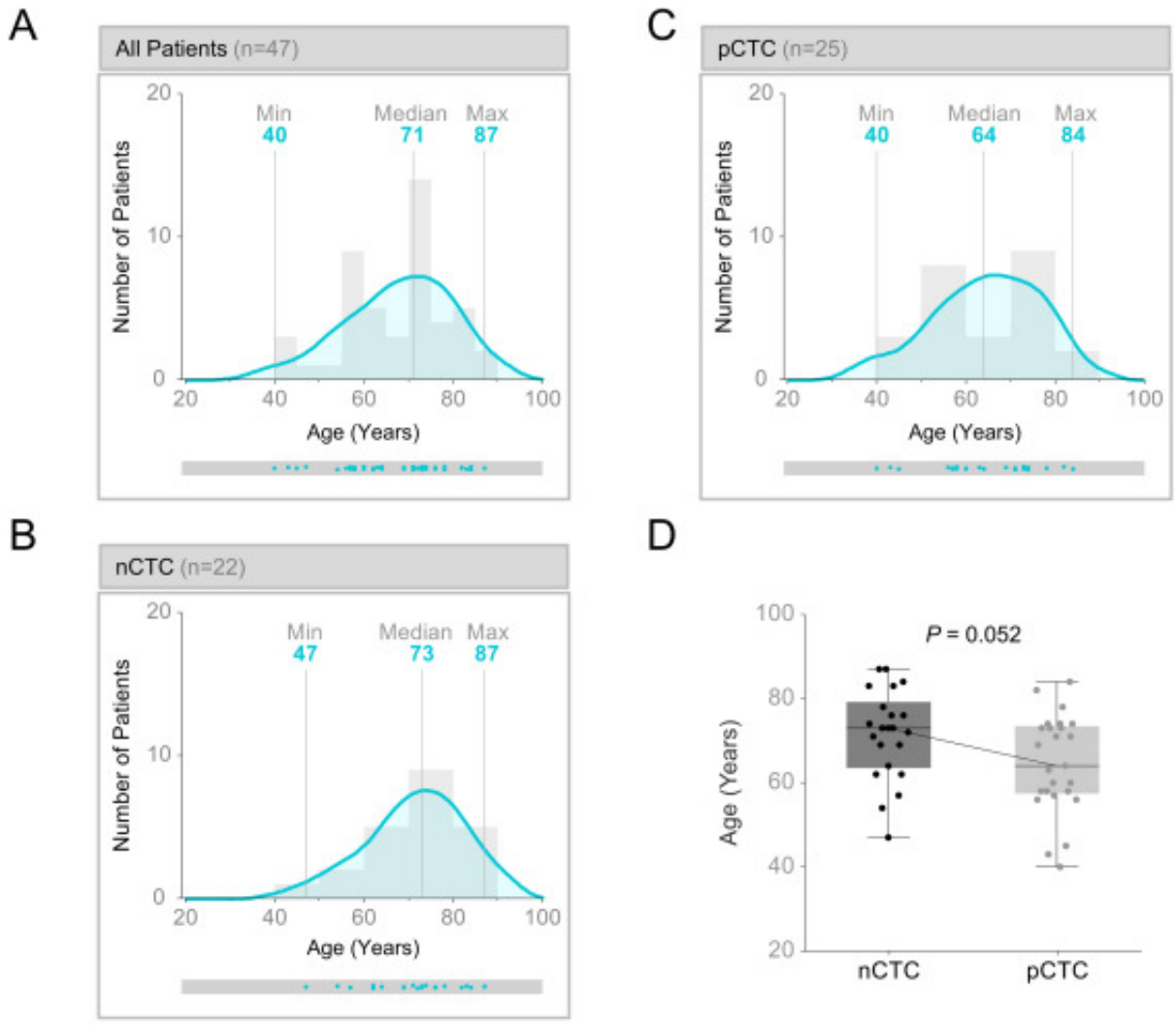
Detection of circulating tumor cells in patients with small choroidal melanocytic lesions



Purpose

To determine the presence of circulating tumor cells (CTC) in patients with indeterminate small choroidal melanocytic lesions (SCML).

Design

Retrospective case series.

Subjects

Forty-seven patients with choroidal melanocytic lesions 2.5 mm or less in tumor thickness and 10 mm or less in largest basal diameter (LBD).

Methods

Blood samples were analyzed for CTC and the presence of Monosomy 3 (M3) in CTC. Tissue biopsy was performed in the patients who were positive for CTC.

Main Outcome Measures

Presence and M3-status of the CTC with regard to the clinical characteristics and result from tissue biopsy.

Results

Median thickness of all (n=47) lesions was 1.1 mm (range: 0.2–2.5 mm) and LBD 5.6 mm (range: 2.0–10.0 mm). CTC were found in twenty-five patients (n=25). This group was classified as positive (pCTC) and compared to the negative (nCTC) consisting of twenty-two (n=22) patients. Median tumor dimensions in the pCTC versus the nCTC group were 1.6 mm (range: 0.6–2.5 mm) versus 0.5 mm (range: 0.2–2.5 mm) for thickness and 6.6 mm (range: 4.1–10.0 mm) versus 4.0 mm (range: 2.0–8.0 mm) for LBD. Both LBD and thickness were positively associated ($P<0.001$) with the presence of CTC. Compared to the nCTC group, a higher percentage of the pCTC patients exhibited LBD >5mm (36% versus 88%), subretinal fluid (9.1% versus 56%), orange pigment (4.5% versus 60%),

sonographic hollowness (9.1% versus 60%), and the presence of multiple risk factors (0% versus 68% for three or more factors) with $P<0.001$ for all parameters. No significant difference was detected in the clinical parameters of the patients who had disomy-3 (n=7) versus monosomy-3 (n=17) in their CTC. The tissue biopsy confirmed the uveal melanoma (UM) in 22 of the 25 pCTC patients (88%), whereas no conclusive diagnosis could be given in the remaining three cases due to insufficient or invalid material.

Conclusions

We report, for the first time, compelling evidence for the potential of liquid biopsy as an additional tool to screen SCML for malignancy. These findings pave the way towards the implementation of liquid biopsy to detect small UM and monitor melanocytic lesions.