

Diagnostic Challenges: Preclinical Presentation Of Dementia With Lewy Bodies In A Middle-aged Adult

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Positive Biomarkers, Preserved Function.

How should we communicate risk before diagnostic criteria for DLB have been met?

Background and Clinical Context:

- DLB is often underrecognized in its earliest stages of development.
- Multidomain testing can be used to evaluate cognition, mood, and subtle changes using validated scales and ancillary tests.
- Biomarkers can identify synucleinopathy before substantial functional decline.
- Early disclosure may support planning and research participation but may also increase anxiety and uncertainty.
- Evidence-based disclosure frameworks are currently limited.
- Early detection of DLB can raise clinical, ethical, and communication challenges.

Participant Profile

Characteristic	Finding
Age	Late 50s
Sex	Female
Family History	Positive for DLB
Functional Status	Independent
Cognitive Concerns	1-2 years
Informant Concern	Present

Results

Cognitive performance WNL, with mild domain specific weaknesses in attention. Mood characteristics were clinically relevant to disclosure planning.

Measure	Score
MoCA	26/30
AD8	4/8
HADS-Anxiety	10/21
GDS	2

BIOMARKER FINDINGS:

MRI: No gross abnormalities
α-Synuclein Skin Biopsy: 2/3 sites positive
DAT SPECT: Mildly abnormal
Cardiac MIBG: Negative
UPSIT: Mild hyposmia

Findings consistent with a preclinical/prodromal DLB risk state.

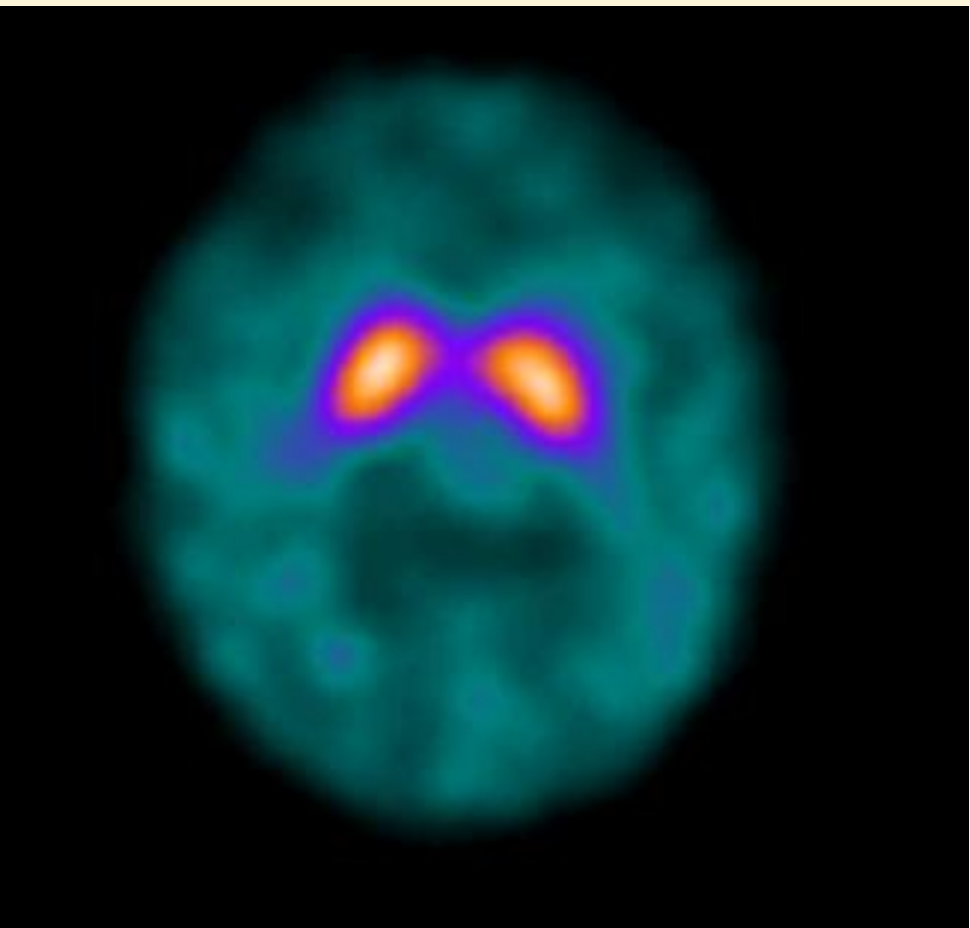
Results were reviewed through consensus, emphasizing symptom trajectory, multimodal biomarkers, and risk communication.

Research Clinician Disclosure Strategy

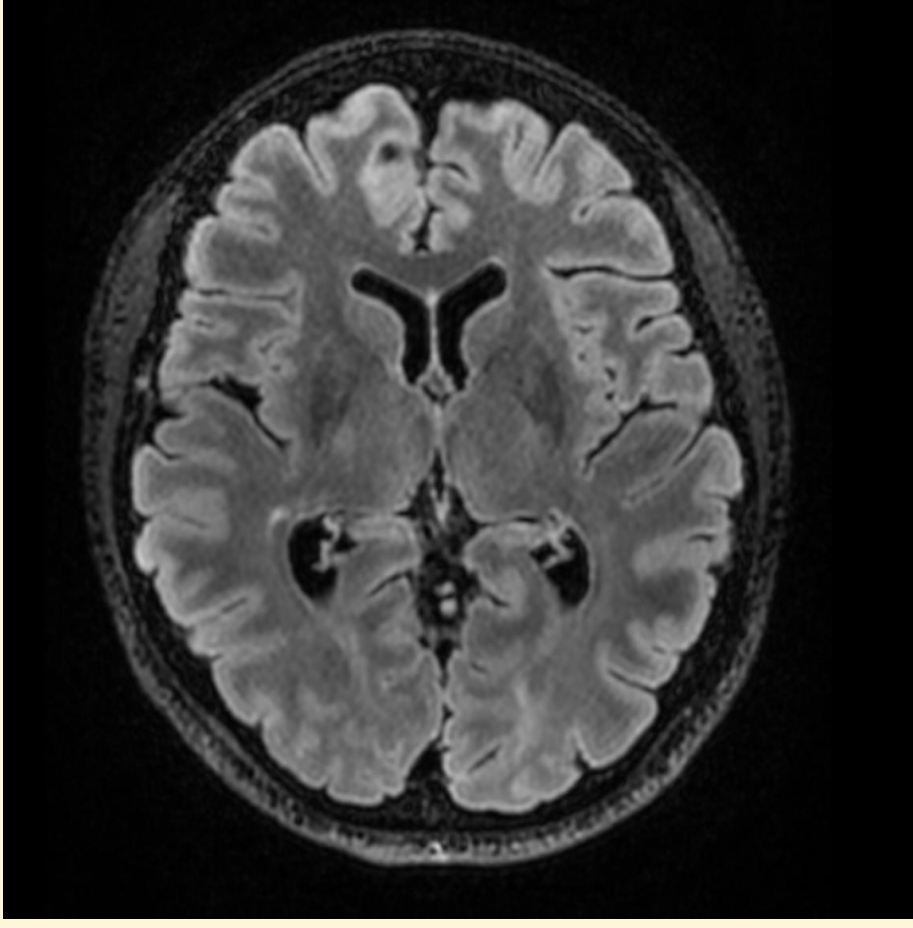
Participant-Centered Communication

- Assess baseline anxiety
- Review uncertainty transparently
- Avoid premature diagnosis
- Discuss a monitoring plan
- Offer support resources
- Consider optional genetic counseling to promote risk understanding and education

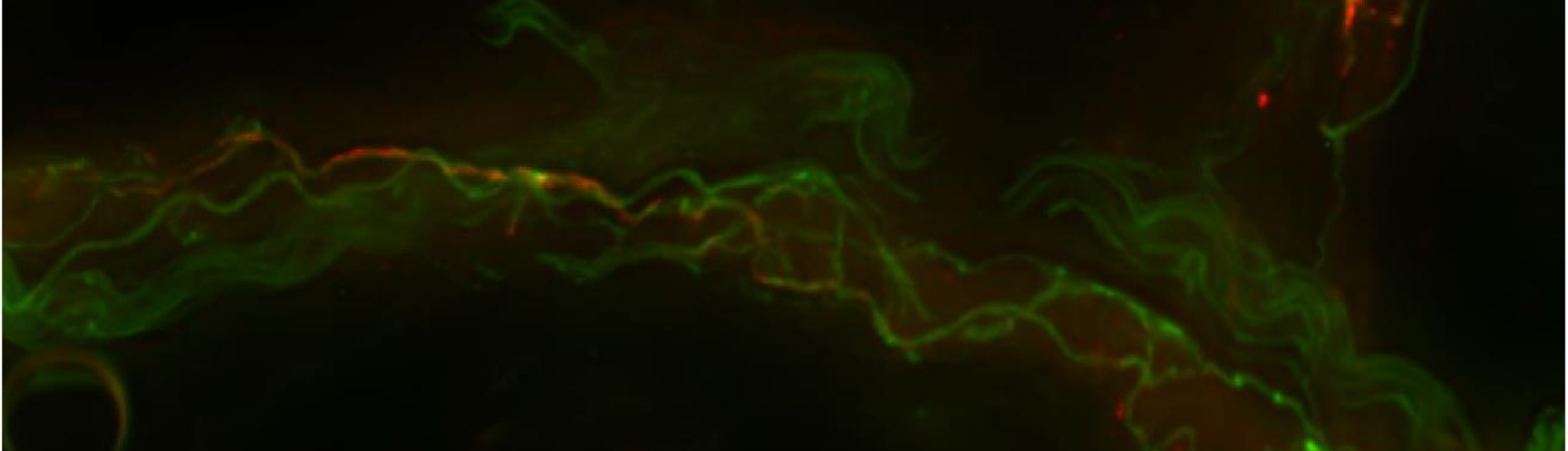
DAT SPECT*



MRI*



SKIN BIOPSY*



Actual subject image. The green regions indicate PGP9.5 immunostained nerve fibers. The orange regions are immunostained deposits of phosphorylated alpha-synuclein within nerve fibers. This photo illustrates evidence of phosphorylated alpha-synuclein deposition.

Outcome and Interpretations

- Participant received structured disclosure and enrolled in longitudinal follow-up study
- Communication is as important as biomarker testing in early DLB risk states
- Disclosure should be individualized and psychologically informed
- Longitudinal monitoring is essential for interpreting early biomarker findings
- Research participation and counseling provide support while avoiding premature diagnosis

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