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Pearls and Pitfalls of ^{18}F-FDG PET/CT for Suspected Alzheimer's Disease in Patient with Down Syndrome

Alzheimer Hastalığından Şüphelenilen Down Sendromlu Olgunun Beyin ^{18}F-FDG PET/BT'sindeki İnciler ve Tuzaklar

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Abstract

Dementia in individuals with Down syndrome (DS) is the leading cause of early-onset cognitive decline occurring before the age of 50. However, establishing an accurate diagnosis remains particularly challenging due to pre-existing intellectual disability, variability in baseline cognitive function, and the limited reliability of standard neuropsychological assessments. ^{18}F-fluorodeoxyglucose positron emission tomography (^{18}F-FDG PET) plays a critical role in identifying underlying neurodegenerative processes and supporting the diagnosis. We present a case of DS with multiple known and unknown pathologies identified on brain ^{18}F-FDG PET and a confirmed diagnosis of early-onset mild Alzheimer's disease.

Keywords: ^{18}F-FDG, dementia, Alzheimer, Down syndrome

Öz

Down sendromlu (DS) bireylerde demans, 50 yaş altındaki erken başlangıçlı bilişsel gerilemenin en sık nedenidir. Ancak, önceden var olan entelektüel yetersizlik, başlangıç bilişsel düzeyindeki bireysel farklılıklar ve standart nöropsikolojik testlerin sınırlı güvenilirliği nedeniyle doğru tanı koymak oldukça güçtür. ^{18}F-florodeoksiglukoz pozitron emisyon tomografisi (^{18}F-FDG PET) altta yatan nörodejeneratif süreçlerin ortaya konulmasında ve tanının desteklenmesinde kritik bir rol oynamaktadır. Burada, beyin ^{18}F-FDG PET incelemesinde hem bilinen hem de atipik patolojilerin saptandığı ve erken başlangıçlı hafif Alzheimer hastalığı tanısı konulan DS'li. bir olguyu takdim ediyoruz.

Anahtar Kelimeler: ^{18}F-FDG, demans, Alzheimer, Down sendromu

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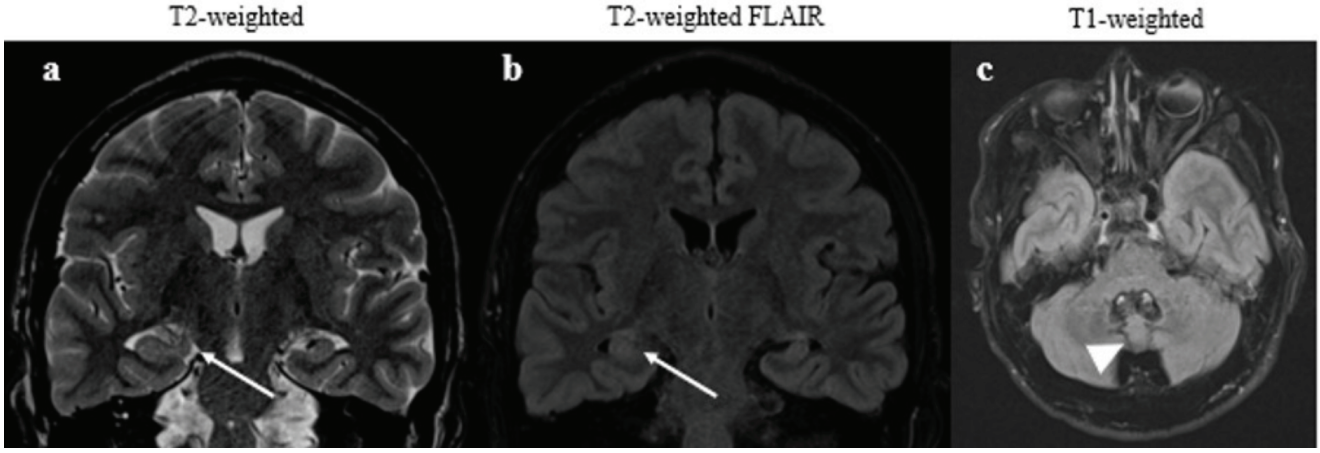

Figure 1. Thirty-five-year-old man patient with Down syndrome admitted to hospital with forgetting tasks, decrease in expression of wants/desires. After an episode leading to a fall/presumed seizure 5 years ago, his cognition and demeanor have changed. Electroencephalography revealed stiffening behaviors that are inconsistent with seizures. The Montreal Cognitive Assessment score was 6/30. Brain magnetic resonance imaging revealed mild volume loss of both hippocampi without abnormal T2/FLAIR signal, which likely represents degenerative changes associated with Trisomy-21 (a and b; arrows). Vermian hypoplasia with prominent extra-axial spaces was detected (c; arrowhead).

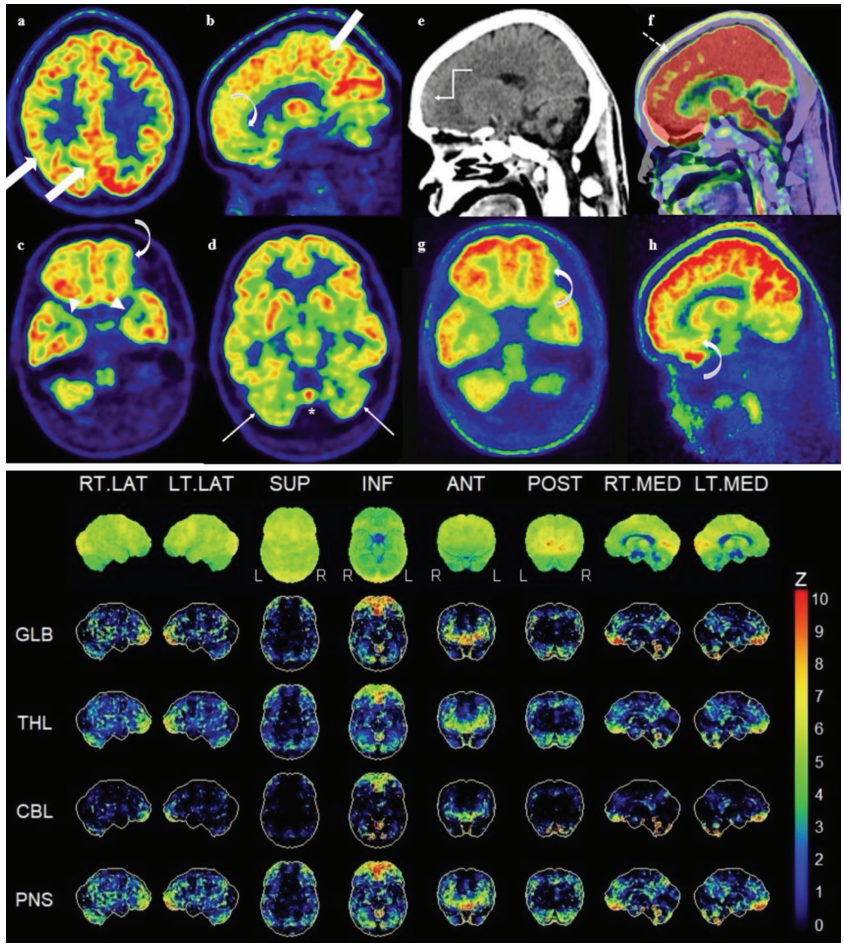

Figure 2. ^{18}F-fluorodeoxyglucose positron emission tomography/computed tomography (^{18}F-FDG PET/CT) scan revealed mildly decreased uptake in the parieto-temporal lobes and precuneus bilaterally which were consistent with Alzheimer's disease (AD) (a and b; thick arrows).
Markedly decreased uptake was detected bilaterally in the anteromedial temporal lobes, secondary to mild hippocampal atrophy (c; arrowheads).
Because of cerebellar vermian hypoplasia, uptake was decreased in the cerebellar hemispheres (d; thin arrows) and in the midline cerebellum (d; asterisk).
Decreased uptake was detected bilaterally in the anterior-inferior frontal lobes (b and c; right-curved arrow) without lobar atrophy on CT images (e; elbow-arrow). In the fused images, approximately 5 mm of head motion in the axial plane was detected between the CT and PET scans (f; dashed-arrow). Hypometabolism in the frontal lobes may have been an attenuation-correction artefact, but, in the light of non-attenuation-corrected (NAC) images, it is probably indicative of true hypometabolism (g and h; left-curved arrow).
The three-dimensional Stereotactic Surface Projections (3D-SSP) demonstrate the AD-related hypometabolic pattern by projecting stereotactically normalized ^{18}F-FDG PET data onto a standardized cortical surface and displaying deviations from an age-matched normative database. Unfortunately, no biomarker test was performed.

DS dementia is the most common form of dementia under the age of 50-years (1). Diagnosing dementia in Down syndrome (DS) could be challenging because of learning disabilities, unknown accuracy of cognitive tests, difficulties performing mental tests, and heterogeneity of intellectual functions. Similar pattern of hypometabolism in AD diagnosed cases either sporadic or DS, suggesting similar neurodegeneration processes (2). Although the diagnosis of AD was not confirmed with biomarker tests in our cases, a strong association between metabolic changes with biomarker levels was already reported (2). 3D-SSP analysis which was proved to be useful in objectively revealing early metabolic changes confirmed AD (3,4,5).

Neuroimaging presents some limitations in DS. Impaired communication may produce artifacts resulting from movement during acquisition. It has been reported that a spatial mismatch of 5-10 mm may result in errors of approximately 10-25% in measured ^{18}F-FDG PET activity (6,7,8). In our cases, we confirmed hypometabolism in the frontal lobes on NAC images. As a reflection difference in brain development, DS participants with or without dementia show lower metabolic rate and gray matter volume in frontal regions (9). Besides, larger involvement of frontal regions in DS cases with AD than sporadic AD cases was also reported (2).

^{18}F-FDG PET was critical not only because it revealed metabolic changes in anatomical pathologies that had already been detected by magnetic resonance imaging, but also because it clarified the AD diagnosis relevant to his current complaints. However, possible artefacts and variations must be well known in this specific patient group.

Ethics

Informed Consent: The written and verbal informed consent was obtained from the patient and his custodian.

Footnotes

Authorship Contributions

Surgical and Medical Practices: E.A., Concept: E.A., S.M., Design: E.A., S.M., Data Collection or Processing: E.A., Analysis or Interpretation: E.A., S.M., Literature Search: E.A., Writing: E.A.

Conflict of Interest: No conflicts of interest were declared by the authors.

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