A)



B)



S1 Figure 1. Receiver operating characteristic (ROC) curves for CERAD total score. CERAD subtests and MMSE. A) Discrimination of asymptomatic+MCI vs Dementia. and B) Discrimination of asymptomatic vs MCI.

S1. Table 1. Comparison of AUCs for CERAD total score. CERAD subtests and MMSE in diagnosing MCI and dementia on a cohort of PSEN 1 E280 mutation carriers and non-carriers

|  |  |  |  |
| --- | --- | --- | --- |
|  | AUC | CI95% | |
| IL | SL |
| Asymptomatic vs Dementia |  |  |  |
| CERADTS/100 | 0.99 | 0.99 | 1.00 |
| VF/24 | 0.93 | 0.89 | 0.96 |
| BNT/15 | 0.84 | 0.80 | 0.89 |
| WLL/30 | 0.99 | 0.99 | 1.00 |
| WLR/10 | 0.99 | 0.98 | 0.99 |
| WLRD/10 | 0.98 | 0.97 | 0.99 |
| CP/11 | 0.92 | 0.89 | 0.96 |
| MMSE | 1.00 | 0.99 | 1.00 |
| Asymptomatic vs MCI |  |  |  |
| CERADTS/100 | 0.86 | 0.82 | 0.91 |
| VF/24 | 0.61 | 0.53 | 0.70 |
| BNT/15 | 0.53 | 0.43 | 0.63 |
| WLL/30 | 0.88 | 0.84 | 0.93 |
| WLR/10 | 0.93 | 0.90 | 0.96 |
| WLRD/10 | 0.84 | 0.77 | 0.91 |
| CP/11 | 0.62 | 0.54 | 0.71 |
| MMSE | 0.82 | 0.76 | 0.88 |

VF/24: Verbal fluency/24; BNT/15: Modified Boston Naming test/15;WLL/30: Word List Learning/30; WLR/10: Word List Recall/10; WLRD/10: Word List Recognition Discriminability/10;CP/11: Constructional Praxis/11 Copy; CERADTS/100: CERAD total score.MMSE: Minimental state examination

S1. Table 2. Sensitivity analysis adjusting for age and education for CERAD total score in diagnosing MCI and dementia on a cohort of PSEN 1 E280 mutation carriers and non-carriers

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | AUC | Cut off | Sensitivity | Specificity |
| (CI 95%) |  |  |
| Asymptomatic +MCI vs Dementia |  |  |  |  |
| Without covariates | 0.994 (0.989 - 0.999) | <58 | 97.4 | 91.9 |
| Adjusting for  Age | 0.976 (0.963 - 0.989) |  |  |  |
|  |  | <55 | 96.5 | 86.2 |
|  |  | <58 | 96.5 | 80.0 |
| Adjusting for  Age + Education | 0.981 (0.968 - 0.996) | <55 | 92,9 | 94,2 |
|  |  | <58 | 97.6 | 87.5 |
| Asymptomatic vs MCI |  |  |  |  |
| Without covariates | 0.862 (0.816 - 0.908) | <69 | 91.3 | 68.7 |
| Adjusting for  Age | 0.789 (0.734 - 0.843) |  |  |  |
|  |  | <66 | 0.833 | 0.563 |
|  |  | <69 | 0.889 | 0.445 |
| Adjusting for  Age + Education | 0.799 (0.744 – 0.855) |  |  |  |
|  |  | <66 | 0.922 | 0.616 |
|  |  | <69 | 0.956 | 0.791 |

AUC: Area under curve; MCI: Mild Cognitive Impairment.