



CASE SERIES

IRCCS Istituto Auxologico Italiano

Federico Verde

email
federico.verde@unimi.it

15/07/2022

1 Case Series Description

Age Range: 30-90

Size N: 750 subjects

Recruitment: 2010 - 2022

Data collection: 2010-2022

Healthy controls: yes (n: 150)

Only at-risk gene carriers included: yes

Diseases studied:

Alzheimer Disease (AD)
Subjective Cognitive Complaints (SCC)
Mild Cognitive Impairment (MCI)
Vascular Dementia (VaD)
Lewy Bodies Dementia (DLB)
Parkinson Disease (PD)
Progressive Supranuclear Palsy (PSP)
corticobasal Degeneration (CBD)
Creutzfeldt-Jakob Disease (CJD)
Amyotrophic Lateral Sclerosis (ALS)
Huntington's disease (HD)

Clinical Evaluation:

- Neuropsychological Assessments (tests performed): MMSE (Measso et al., 1993); Matrici Attenziali (Spinnler and Tognoni, 1987); Trail Making Test (TMT) (Giovagnoli et al., 1996); Stroop test (Caffarra et al., 2002); FAB (Apollonio et al., 2005); Matrici Progressive di Raven – 47 (Basso et al., 1987); Clock Drawing Test (Mondini et al., 2003); Digit Span avanti (Monaco et al., 2013); Digit Span indietro (Monaco et al., 2013); Span Spaziale avanti (Monaco et al., 2013); Span Spaziale indietro (Monaco et al., 2013); Breve Racconto (Novelli et al., 1986); Rievocazione immediata e differita delle 15 Parole di Rey (RAVLT) (Carlesimo et al., 1985); Test della Figura Complessa di Rey-Osterrieth (ROCF) (Caffarra et al., 2002); Denominazione Visiva (Sartori et al., 1988); BADA Denominazione Nomi (Miceli et al., 1994); Fluenza fonologica FAS (Carlesimo et al., 1995); Fluenze Verbali fonemiche FPL (Novelli et al., 1986); Fluenza semantica Frutta-Animali-Marche d'auto (Novelli et al., 1986); FCSRT (Frasson et al., 2011); Aprassia Costruttiva (Spinnler and Tognoni, 1987); Street's Completion Test (Spinnler and Tognoni, 1987); Digit Symbol Test (Amodio et al., 2008); ECAS (Poletti et al., 2016)
- Behavioural Assessments (tests performed): Frontal Behaviour Inventory (Alberici et al., 2007); Neuropsychiatric Inventory (NPI) (Cummings et al., 1994); Beck Depression Inventory (Beck et al., 1961); State Trait Anxiety Inventory (STAI) (Spielberger et al., 2007).

Imaging and Neurophysiology:

- EEG
- MRI 3 T
- MRI 1.5 T
- FDG-PET (rarely)
- Amyloid PET (rarely)
- DaT SPECT (in selected cases)
- Myocardial MIBG scintigraphy (rarely)

Genotyping: yes

- C9orf72, ALS2, ANG, ANXA11, APOA1, APOE, APP, ARPP21, ATP13A2, ATP1A3 (DYT12), ATP6AP2, CHCHD10, CHMP2B, CST3, DAO, DCTN1, DJ1 (PARK7), DNAJC13, DNAJC6, EIF4G1, FBXO7 (PARK15), FUS, GBA, GCH1 (DYT5), GIGYF2 (PARK11), GRN, GSN, HNRNPA1, HNRNPA2/B1, HTRA2, LRKK2, MAPT, MATR3, NEFH, NEK1, NOTCH3, OPTN, PACRG, PARK2, PFN1, PINK1 (PARK6), PLA2G6 (PARK14), PRKRA (PARK16), PRNP, PRPH, PSEN1, PSEN2, RAB39B, SETX, SNCA, SOD1, SORL1, SPAST, SPG11, SQSTM1, TAF1 (DYT3), TARDBP, TBK1, TH (DYT5), TREM2, TTR, TUBA4A, UBQLN2, UCHL1, VAPB, VCP, VPS13C, VPS35

Digital Data obtained from patients through electronic devices: no **Biological Samples:**

- CSF
- DNA
- Plasma
- Serum
- Fibroblast (in selected cases)
- iPSC (in selected cases)

Follow-up:

- Number of Follow-ups planned: 200 (tot)
- Number of Follow-ups completed: 400 (tot)
- Average duration between follow-ups (years): 0.5 year
- Follow-ups type: Clinical evaluation, neuropsychological evaluation (in selected cases)