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Objectives: To report the clinical, psychological and neuroradiological features of a family with a mutation in the VCP gene and study any premanifest cognitive or behavioural changes.

Materials: Five members of a large family with IBMPFD were recruited at the Dementia Research Centre (University College London Institute of Neurology).

Methods: The subjects underwent a standardized interview, neurological examination, neuropsychometry, brain MRI (except for subject 5) and genetic analysis. The baseline MRI scans from the four mutation carrier were compared to a control group of 26 age-matched cognitively normal subjects in a Voxel-based morphometry analysis.

Results: Genetic analysis showed a heterozygous c.464G>A nucleotide substitution in exon 5 of the valosin containing protein gene (R155H) in all of the subjects studied. All of them had a progressive myopathy that affected the shoulder and pelvic girdles predominantly. Paget's disease was only found in two subjects (4 and 5). All subjects were cognitively and behaviourally asymptomatic when first seen. Only subject 1 developed behavioural changes typical of frontotemporal dementia (loss of empathy, preference for sweet food and delusions). At the initial assessment only subject 2 showed any abnormalities in neuropsychological testing with evidence of executive dysfunction. Subject 1 developed executive dysfunction on follow-up assessment. Visual assessment of brain MRI was normal for all the members of the family scanned without any atrophy of the frontal or temporal lobes. However, the voxel-based morphometry analysis showed evidence of grey matter atrophy in frontal (premotor, prefrontal and orbitofrontal) and temporal (particularly medial) lobes.

Discussion: Inclusion body myopathy (IBM) with Paget's disease of the bone (PDB) and Frontotemporal dementia (FTD) (IBMPFD; OMIM 167320) is a rare autosomal dominant disease caused by mutations in the valosin containing protein (VCP) gene on chromosome 9p13.3-12. According to the literature IBM is present in 90% of carriers, Paget's disease of the bone 50% and FTD only 30%. Our data reflect this distribution but we also showed the presence of presymptomatic executive dysfunction in one subject and evidence for premanifest brain atrophy.

Conclusion: Executive dysfunction and brain atrophy can occur presymptomatically in VCP mutation carriers.

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ORAL HEALTH STATUS AND PERIODONTITIS IN ALZHEIMER'S DISEASE: A POPULATION-BASED, CASE-CONTROL STUDY FROM THE ZABUT AGING PROJECT

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Aims: The aetiopathogenesis for Alzheimer's disease (AD) have not been defined, although inflammation within the brain is thought to play a role. Recent data suggest that peripheral infections contribute to the inflammatory state of central nervous system diseases, including cogn impairment and dementia (1). Periodontitis (PD) is a prevalent, ch infection involving the tissues supporting the teeth associated with negative, anaerobic bacteria capable of promoting local and system lease of inflammatory mediators. Aim of the present study was to a the oral health status and the prevalence/severity of PD in inciden subjects evaluated during a population-based, 10-year follow-up st

Materials and Methods: 10-year follow-up data from the Aging Project, a population-based study on dementia and cogn impairment conducted in a rural sicilian population were used fo present case-control study. From the >1030 subjects evaluated a low-up, 130 were subjects with incident dementia according to D IV criteria, and 38 were diagnosed with probable AD accordi NINCDS-ADRDA criteria. Seventeen (44%) of the latter were t edentulous and were excluded from this study. Accordingly, the sample included 21 cases (M: 9; F: 12, age-range 64-93 years 21 age- and sex- matched controls (M: 9; F:12; age-range years). To evaluate oral health status the Decayed Missed Filled (DMFT) scoring and measurement of the periodontal probing (using Community Periodontal Index - CPI and the Periodontal S ning and Recording Index -PSR) were recorded. The association b en AD and oral health indexes was assessed using the $\chi 2$ test or as appropriate. A p value ≤ 0.05 was considered statistically signifi

Results: DMFT average score was similar in AD (18.7±8. controls (19.2±7.8) (p=0.743). Contrarily, differences between and controls groups regarding periodontal status approached stati significance; indeed, high PSR and CPI scores (>3, indicating mo te-severe PD) were recorded in nearly 78% (n=16) of cases and (n=10) of controls (p= 0.06).

Discussion and Conclusions: In this rural Sicilian adult-to e community an overall poor oral health was frequent with a high D score. The current data moderately support the hypothesis of a prevalence and severity of PD among AD patients. To confirm thes liminary results we are currently recruiting a wider sample, incl proteomic salivary profiles and RT PCR-based microbiological in gation on sub-gingival plaque samples, in order to better clarify th of PD and periodontal pathogens in the AD natural history.

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CEFTRIAXONE FOR ALEXANDER'S DISEASE: A F YEARS FOLLOW-UP

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Introduction: Alexander disease (AD) is a rare, usually fatal neu generative disorder, involving primarily astroglial cells in the caused by dominant mutations in the gene encoding glial fibrillar dic protein. Here we studied the tolerability and therapeutic effe the chronic use of cycles of ceftriaxone, a beta-lactam antibioti neuroprotective effects, in a patient affected by adult AD with a r progressive clinical course. In 2010, we reported the successful c outcome related to a 20-month course of intravenous, cyclical ceft ne, in a patient with adult-onset AD.

Objectives: To evaluate the tolerability and therapeutic effe cycles of ceftriaxone in this patient at a 4-year follow-up.

Methods: Gait ataxia, dysarthria, palatal myoclonus, and e nystagmus/oscillopsia were evaluated every 6 months over a 6-ye riod. For the first 2 years, without therapy, and for the following 4 after intravenous ceftriaxone 2 g daily, for 3 weeks monthly duri initial 4 months, then for 15 days monthly. Gait ataxia and dys