in physical and mental health; 7) Technology and Internet addiction. General lack of physical exercise was reported (3/10 reported weekly exercise). Younger age groups and those with severe ASD were more physically active. Parents of YP with severe ASD were less physically active than milder forms of ASD. Parents preferred group over individual sports for their children, whilst children preferred individual sports.

Conclusion: This highlighted the importance of sport in YP with ASD. YP preferred individual sports, reflecting ASD symptomatology. Parents felt burnt out and unsupported by society. Sport therapy services for ASD should be YP-centred and include public/parent psychoeducation. This opens new fields of research in aid of national service.

P6.04

Identifying barriers to antiepileptic treatment adherence in Malta

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Introduction: Adherence to treatment and lifestyle advice in persons with epilepsy is important to minimize the recurrence of breakthrough seizures and complications including injuries, hospitalization and loss of work. The aims of the study were thus to determine the rate of drug adherence amongst persons with epilepsy in Malta and to identify possible factors which may be affecting non-adherence rates.

Methods: Following literature review, an Adherence Assessment Tool was designed with the aim of calculating patient adherence rate (Medication Adherence Rating Scale and Proportion of Days Covered) and identifying factors that may be influencing the persons' adherence Relevant Permission was obtained from the Consultant Neurologists, Ethics Committee, and Mater Dei Hospital to approach persons with epilepsy and to gain access to patient files. Persons who agreed to take part in the study were asked to fill in the Adherence Assessment Tool. Responses were analysed using IBM® SPSS Version 22.

Results: A total of 200 Adherence Assessment Tools were distributed, of which 90 were returned filled in (response rate = 45%). This number was statistically representative of the patient population. The Proportion of Days covered was calculated for 47 respondents (52.22%). The Medication Adherence Rating Scale score showed that 75.56% of the respondents achieved either a high (9-10) or moderate (7-8) adherence rating score. Significantly higher adherence scores was obtained from respondents who did not take medications for other healthcare conditions, were supplied with written instructions by their neurologist or pharmacist, did not experience memory problems, did not mind taking their medications in public and had not experienced side effects. A higher quality of life in epilepsy score, shorter periods of time feeling low or down-hearted and person experiencing less bothersome work limitations and social limitations were also associated with better adherence.

Conclusion: The results demonstrate that the majority of the respondents were adherent to their antiepileptic treatment. Increasing patient and carer knowledge about epilepsy, treatment options and the importance to adhere to treatment regimens may encourage the patients to become more adherent. Teaching patients self-management skills, life skills and education society in general about epilepsy can help persons with epilepsy be self-sufficient, find employment and feel less stigmatized.

P6.05

Effect of an intronic variant within Zinc finger protein 384 gene on pre-mRNA splicing in a Maltese family with osteoporosis

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Introduction: Osteoporosis is a skeletal disease with a strong genetic basis. A study on an extended Maltese family with a highly penetrant form of osteoporosis, revealed the presence of the rs146089604 variant (c.686+32G>A) in intron 7 of the Zinc finger protein 384 (*ZNF384*) gene, predicted to affect pre-messenger RNA (mRNA) splicing. The aim of this study was to assess the functional effect of the variant using an exon-trapping vector transfected in three human cell types.

Methods: The target DNA region harbouring G or A allele was inserted in the p.SPL3 vector, creating mini-gene constructs that were transfected in human kidney-derived cells (HEK-293) and two human osteoblasts-derived cells (SaOS-2 and h-FOB). Extracted mRNA was converted into complementary DNA (cDNA), amplified by PCR and sequenced to determine the transcript size and identify any splicing variants.

Results: Mini-gene construct with the alternative A allele lead to exon 8 and part of intron 8 to be retained, both of which were spliced off in the presence of the G allele. These results were observed for constructs transfected in the osteoblasts-derived cell lines. In HEK-293 cells, no difference in transcript size was seen for the G or A allele, suggesting different splicing mechanisms.

Conclusion: Observations may indicate that the *ZNF384* rs146089604 could be a causal variant contributing to osteoporosis. ZNF384 transactivates type I collagen and matrix metalloproteinases, and suppresses bone morphogenic protein (BMP) and Wnt signalling resulting in reduced bone volume and strength. Thus, impaired *ZNF384* splicing could alter the protein's function affecting bone homeostasis.