An unregistered TARDBP mutation in a case presenting with young-onset dementia

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Objective. This poster aims to report an unregistered mutation in Transactive Response DNA Binding Protein (TARDBP) gene in a patient presenting young-onset dementia.

Hypothesis: Novel heterozygous mutation in the TARDBP gene is linked to a case of with young-onset dementia.

Background. Pathogenic variants in TARDBP cause autosomal dominant fronto-temporal degeneration, characterized by TDP43-positive inclusions, dystonia, dyslexia, receptive dysphasia, and paraphrasic errors. In addition to the neurocognitive deficits, patients might suffer from cardiomyopathy and amyotrophic lateral sclerosis.

Case report. Molecular genetic analysis of whole-exome sequencing (WES) was carried out for a 45-year-old male patient presenting with cognitive decline and behavioural symptoms.

Discussion. WES Identified the heterozygous variant c.527A > T p.(Lys 176lle) in TARDBP gene. To the best of our knowledge the variant has not been described in the literature so far (HGMD 2019.3). No allele frequencies in the general population have been documented.

Conclusion. We believe that we have identified a novel mutation in the TARDBP gene. This mutation is likely to be linked to this patient presenting with young-onset dementia.

Off-licence use of clozapine in patients with emotionally unstable personality disorder: a case series analysis

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Objective. This retrospective case series followed emotionally unstable personality disorder (EUPD) patients following initiation of clozapine on an off-licence basis, aiming to examine tolerance by determining side effect prevalence and treatment cessation frequency, as well as examining efficacy, by investigating number of hospital re-admissions and symptom control.

Case report. This case series captured the experiences of 11 EUPD patients under the care of Mersey Care NHS Foundation Trust, all of whom had, at some time in the past five years, been initiated on clozapine. All patients were white British females, with a median age of 31. The median daily dose of clozapine was 300 mg. Most patients had significant psychiatric comorbidities, as well as illicit substance and / or alcohol misuse.

Whilst prescribed clozapine, patients were only admitted to hospital once on average and this was commonly for clozapine re-titration. Whilst in hospital, rates of self-harm were low, but ligaturing and suicide attempts showed higher prevalence. Patients still demonstrated self-harming behaviour out of hospital leading to A and E presentations. In the community, contacts with the police were minimal, with only two patients undergoing Section 136 assessments or arrests.

All patients reported side effects from clozapine - usually hypersalivation, over-sedation and constipation. All 11 patients experienced sinus tachycardia. Eight patients temporarily ceased taking clozapine at some point. In three patients, discontinuation of clozapine was as a result of intolerable side effects. Three patients experienced neutropenia, which subsequently resolved. Only two patients had a body mass index within healthy range.

Discussion. Despite patients reporting clozapine to provide symptomatic benefit for their EUPD, and improved their engagement with mental health services, prevalence of self-harm and of A and E presentations remained high, indicating the importance of community support and concomitant psychotherapeutic treatment. Patients with more robust community support showed greater adherence to clozapine.

High prevalence of side effects and obesity in these patients, in addition to risk of developing neutropenia, highlights the importance of rigorous monitoring after initiating clozapine. It is reassuring that, despite development of neutropenia in some patients, this recovered quickly, and clozapine treatment could resume.

Conclusion. Clozapine may be an effective pharmacological treatment for enabling EUPD patients to engage more therapeutically with services. Clozapine may be of greater benefit to those with more stable, less chaotic lives. Although diminished, patients still show self-harming behaviour and need for A and E admissions and re-hospitalisation. Side effects of clozapine are common and regular monitoring is required.

A haven for an extremely disturbed young person

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Objective. The reason to share this case is to high light the lack of resources in mental health services which can delay the provision of appropriate care and this can have negative impact on child health outcomes.

Background. A 10-year-old boy was referred to CAMHS. He presented with extremely challenging behaviours. After first appointment with CAMHS he attacked his father and nurses. He had to be restrained multiple times. He started to use wooden chair as a weapon, threatened to harm others and threatened to urinate on staff. He tried to kill him-self by ligature. Mental health act assessment was completed and when a decision was reached that detention under the mental health act was appropriate, no appropriate bed was available. He was admitted under Section II of MHA to paediatric ward where he remained for one week (with 2:1 CAMHS support). Then he was transferred to an inpatient CAMHS unit which was commissioned for children over 12 years of age. At a later date mental health tribunal panel upheld the section. After few days he was transferred to an age appropriate in-patient mental health bed. He stayed there for roughly 6 months and was discharged with a diagnosis of ADHD and Autistic Spectrum Disorder. There was a long delay in discharge, until appropriate specialist residential placement could be identified and he was transferred there. He is well settled now in the placement.

Case report. Legal advice was later taken on this case. MHA 1983, Human Rights Act, Children Act 1989, Criminal Law Act 1967 and Code of Practice 2015 were considered and it was agreed that it was appropriate to use MHA 1983. There was discussion whether the Children Act could be relied on instead, but in

view of the fact that repeated restrain was required, he was in seclusion and possibility of need for rapid tranquilization post admission the decision was made to use the mental health act.
Conclusion. This case has highlighted a significant problem and calls for an urgent action to increase the number of inpatient age appropriate mental health beds and number of appropriate residential placements nationally. It has also been identified that application of legal frame work in children and adolescents can be a challenge and there is a need for targeted educational programmes for professionals on the use of legal frame work in children and adolescents.

Prolong psychosis preceding cognitive and motor symptoms; an unusual presentation in Huntington's disease

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Objective. To describe an unusual clinical presentation of Huntington's disease.
Case report. A 39-years-old married female, a homemaker, presented to the psychiatry clinic with her husband, with over a nine-year history of decreased sleep, suspiciousness, self-talking, agitation, anger outbursts, aggression, and social isolation. The patient was diagnosed with schizophrenia. Previously, she received various antipsychotics and Electroconvulsive therapy (ECT). The patient showed a partial response to treatment. Over the last 2-3 years, the patient had a progressive decline and later required supervision in her Activities of Daily Living (ADLs). She developed slurred speech limited to 1-2 worded answers, gait disturbance, falls, involuntary movements of the trunk and distal extremities, bowel and bladder incontinence, and severe weight loss. The patient's mother and older brother had a history of death in their early 40s due to an unknown cause.

At presentation, the patient was restless, irritable, self-talking incoherently, neither made nor maintained eye contact and tried hitting and biting upon approaching closely. She did not respond to any queries or followed commands. The patient showed poor personal hygiene. On examination, the patient was hemodynamically stable, had a loss of muscle bulk, broad-based gait, and choreiform movements of the trunk and distal extremities. We admitted the patient to the psychiatry ward and also consulted the neurology team. Her blood investigations showed ASMA antibodies positive, MRI brain was suggestive of Huntington's disease (HD), and her genetic test for Huntington gene confirmed the diagnosis of HD. We started the patient on Fluoxetine, Clonazepam, and Olanzapine. The patient showed a decrease in agitation, and her self-talking stopped.
Discussion. HD is a rare genetic disease that has well-characterized symptoms. However, as seen in our patient, these symptoms can evolve and progress unusually in the early and middle stages. Psychosis in HD patients is rare but known. Psychosis is rare in HD and usually presents after a clear clinical picture of HD is apparent. Our case discussed psychotic symptoms in the pre-choreic stage of HD which adds to the existing evidence on challenging presentations and management of HD. Further research can help increase confidence in these outcomes and treatment guidelines.
Conclusion. Our case highlights an unusual clinical presentation of HD, which can be challenging and lead to diagnostic delays. We recommend a thorough approach to history and revision of diagnosis in case of atypical presentations.

Psychosis in youth in Singapore: a case series

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Objective. In this report, we present a case series of children with psychotic symptoms referred to a child consultation liaison psychiatric service within a tertiary paediatric hospital in Singapore. The purpose of this case series is to identify common symptoms at presentation, review the current practices in our hospital for investigation and treatment of first episode psychosis and short-term outcomes.
Case report. We identified 9 cases over a 1 year period, for which 7 were seen whilst admitted to hospital and 2 in the outpatient clinic. There were 6 females and 5 males ranging in age from 11 to 16 years old. The commonest symptoms on presentation were perceptual disturbance (88%) most commonly auditory hallucinations and altered behaviour (55%). Of the 7 children admitted to hospital, all were seen by the neurology team prior to the request for a psychiatric opinion. All admitted patients had blood and radiological investigations carried out. Most of the children were started on a short course of antipsychotic medication with the majority continuing to attend follow-up outpatient.
Discussion. Only 9 cases were identified in this case series over a 1 year period highlighting that psychosis is not a common presentation in the paediatric population. From the history alone, it can be challenging to distinguish between primary and secondary causes of psychosis. Acute onset of symptoms and the presence of other neurological signs should raise the suspicion of an underlying organic cause. Out of 9 cases, only 1 case was treated for a presumed organic aetiology, which is consistent with findings from other authors who only found underlying organic factors in 12.5% of cases.

In this case series, we also noted that 45% of cases reported having symptoms for over 1 year before seeking help. This is also seen in the adult population in Singapore. Stigma, denial and lack of information about psychosis may all contribute to delay in seeking help. Although prolonged duration of untreated psychosis has been shown to be associated with poor long-term outcome, we found in our case series that even patients who reported a long duration of symptoms still responded well to medication.
Conclusion. There is room for collaboration with our neurology colleagues in the approach towards children with first presentation of psychosis, both in terms of investigations and management. Identifying reasons for disengagement from psychiatric care is an area for further investigations to improve outcomes in our patients.

Management of inappropriate sexual behaviour in frontotemporal dementia: a case study

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