anaemia, transient abnormal myelopoesis, post-transplant lymphoproliferative disorder and Fanconi anaemia. Of the remaining 154 patients diagnosed with cancer, 88 (57.1%) were classified as ‘green’ according to the Royal Marsden Hospital ‘Stop-light’ system. 48 (31.2%) were classified as ‘orange’ and 18 (11.7%) were classified as red, qualifying for automatic referral to clinical genetics.

Conclusions A significant proportion of patients diagnosed with cancer in OLCHC may benefit from referral to clinical genetics and screening for underlying cancer predisposition syndromes.
like symptoms, movement disorders and altered state of consciousness. Neuron specific antibodies (LG1, CASPR2, AMPA1R1/2, NMDAR, GABABR) in CSF/serum, antibodies against GAD, thyroid peroxidase and paraneoplastic syndrome panel (anti-Hu, anti-Yo, anti-Ri, anti-Amphiphysian, anti-Tr, anti-PCA-2, anti-Ma, anti-CV-2, anti-ANNA-3) were detected in the serum.

Results The mean onset age of patients were 8.3 years (2–13.8 years). Female and male ratio was 8:7. In all patients admitted to the hospital the most common symptom was seizure (73%). The other symptoms were psychiatric (20%) and altered state of consciousness (7%). Seven (46%) patients with acute seizures present with status epilepticus. The mean hospitalization time of patients were 38.5 days (14–70 days) and 20 days (3–50) at Intensive Care Unit. Five patients were mechanically ventilated.

Significant serum levels of antibodies were detected in 10 out of 15 patients. (GAD n = 8, anti-TPO n = 5) CASPR2 and NMDA in CSF were detected in different two patients. One patient diagnosed with FIRES. Oligoclonal banding or elevated IgG index were detected in 6 of 15 patients. Lymphocytic pleocytosis (40%) were detected in cerebrospinal fluid. All patients had abnormal electroencephalogram findings but 35.7% of patients had abnormal brain magnetresonance imaging findings at the time admission of the hospital.

Immunotherapy, firstline therapy (IVG n = 14, pulse steroid n = 13, plasma exchange n = 7) and second line therapy (rituximab n = 1, cyclophosphamide n = 1) were contributed to treatment.

After the discharge of the hospital no more seizure were seen in 12 patients (80%) during follow-up period. At the last visit all of these patients had normal EEG findings. But in the other 3 patients epileptic activity was detected in their EEGs. Among them, only one patient was resistant to the antiepileptic treatment.

Conclusions In this study, we observed that seizures with IME were generally resistant to the treatment during the acute phase of the illness in pediatric population. But after proper and early immunotherapies, seizures remission was detected with good prognosis in most of them.

Etiology of microcephaly: 5 years experience

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Introduction Rett syndrome (RS) is an X-linked dominant neurodevelopmental disorder affecting girls, most commonly caused by MECP2 loss-of-function mutations. The typical phenotype of RS is characterised by a period of developmental stagnation in late infancy, followed by progressive dementia, developmental regression, stereotyped hand-movements, microcephaly and gait dyspraxia. There are well recognised variants such as Atypical Rett syndrome (Angelman-like), Zappella-variant (preserved speech variant), and Congenital Rett syndrome.

Objective We describe two cases of unrelated Irish females who presented towards the end of their first decade with unusual Rett phenotypes.

Results Both cases had normal early development. Mild/moderate developmental abnormalities were picked up at 2 years. Prior to onset of epilepsy, both cases were in mainstream school with a special needs assistant and had upright and independent gait. Case 1 developed severe progressive scoliosis from preschool age.

Case 2 and Case 2 presented with severe, refractory epilepsy aged 10 and 9 respectively; consisting of a mixture of atonic and complex-partial seizures evolving to include epileptic spasms with epileptic-encephalopathy in both. In both cases gait deterioration began as an increasingly crouched posture and shuffling gait progressing to wheelchair-dependence. Concurrently, there was a profound regression in motor skills, cognitive abilities, very poor appetite and weight loss. Neither had features of hand-wringing or microcephaly.

Case 1 was notable for excessive drowsiness and development of progressive neuromuscular muscle weakness leading to frequent pneumonias, life-threatening choking episodes and a progressive deterioration in her respiratory function. She died, aged 13 years, of respiratory failure. Case 2, aged 13, does not have features of progressive bulbar palsy described in Case 1.

Extensive workup was unrevealing except for de-novo heterozygous mutations in their MECP2 genes. In Case 1, a novel variant of MECP2 was detected: A duplication of the majority of the protein-coding portion of exon 4. Isolated duplication of large sections exon 4 has never, to our knowledge, been described before. In Case 2 there was a deletion and seven base pair insertion in exon 4: 1006_1275delinsBATGCC(p.Leu336Asps12), predicted to introduce a premature termination codon.

Conclusions Case 1 represents a unique case of MECP2 mutation, expanding our knowledge of the deleterious mutations leading to a Rett phenotype.

We suggest that young girls with late onset developmental regression, new onset refractory epilepsy and crouching gait be investigated for MECP2 gene mutations.

Early onset of scoliosis in an ambulatory child, and severe neuromuscular weakness are not previously known features of this condition.