Diagnostic Journey, Seizure Burden, and Quality of Life Among Patients with CDKL5 Deficiency Disorder Using Real-World Data

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OBJECTIVES

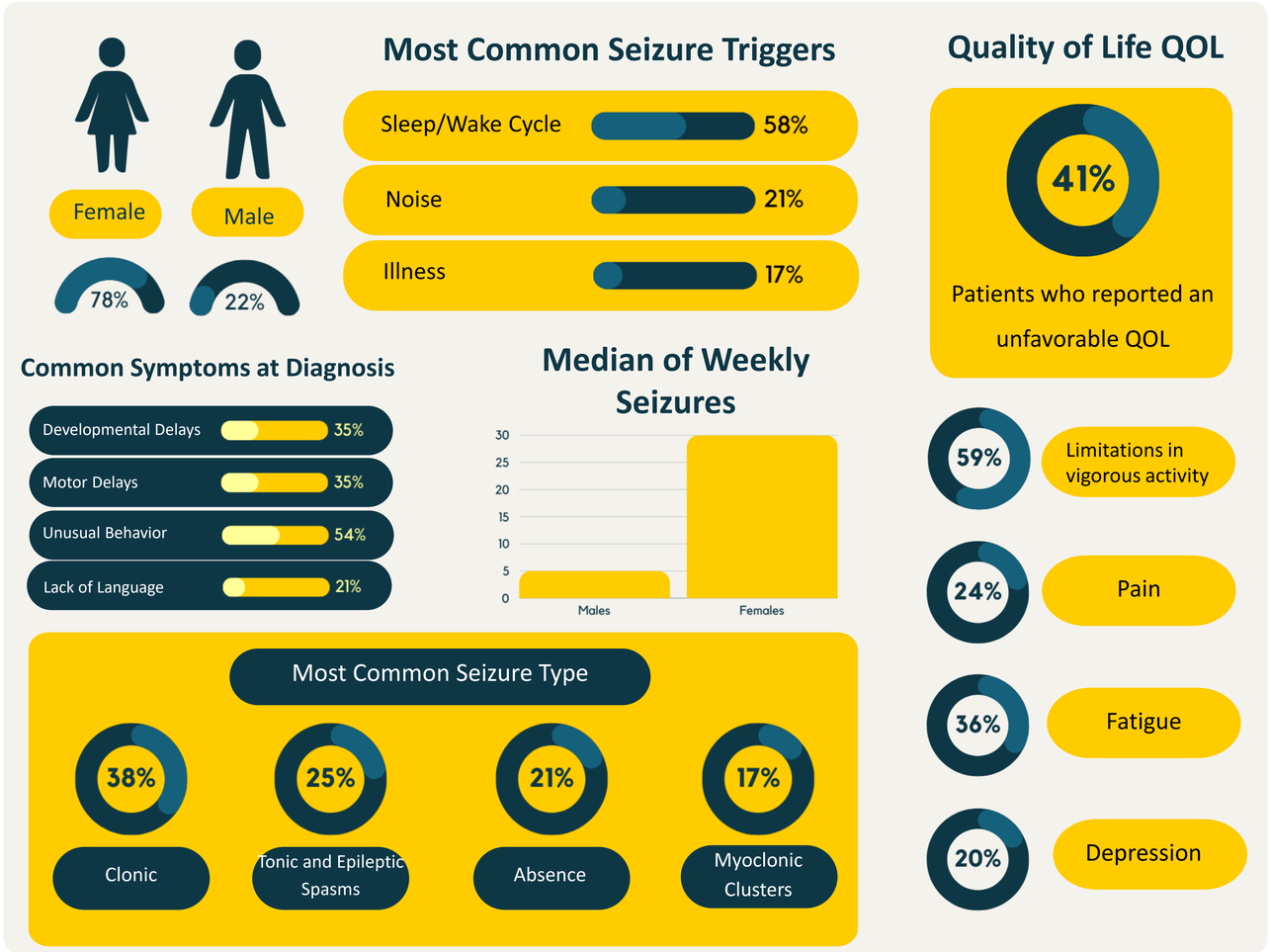
Using data collected from an international registry of patients with CDKL5 Deficiency Disorder (CDD), we aimed to describe the diagnostic journey, seizure burden, and quality of life of these patients

METHODS

The CDKL5 Deficiency Disorder International Patient Registry (CDKL5 Registry) is an international, prospective, observational web-based registry of patients with a diagnosis of CDD confirmed by a clinician or genetic test. We analysed questionnaires completed by patient caregivers at registry enrolment from December 2018 through November 2023. Patient demographics, clinical characteristics, seizure characteristics, and quality of life were descriptively summarized. Domains of quality of life were measured using single-items with 5-point Likert scales; unfavourable quality of life was defined by selecting the lowest 2 options.

RESULTS

46 participants (36 females and 10 males) with CDD were included. The median age at symptom onset was 1 month and at diagnosis was 8 months. Apart from seizures, the most frequently specified symptoms leading to diagnosis were developmental delays (n=16, 35%), motor delays (n=16, 35%), unusual behaviors (n=12, 26%), and lack of language (n=5, 11%).



RESULTS CONT.

Among those who completed seizure diaries (n=24), seizures were most frequently triggered by sleep/wake cycle (n=14, 58%), noise (n=5, 21%) and illness (n=4, 17%). Females (n=19) and males (n=5) reported having a median of 5 and 30 seizures per week, respectively. Seizures experienced included tonic clonic (n=9, 38%), tonic and epileptic spasms (n=6 each, 25%), absence (n=5, 21%), and myoclonic clusters (n=4, 17%). At enrollment (median age 5 years), unfavorable quality of life was reported for general health (41%), limitations in vigorous activity (59%), pain (24%), fatigue (36%), and depression (20%).

CONCLUSION

Patients with CDD experience delayed diagnoses, substantial seizure burden and low quality of life. As the CDKL5 Registry matures, future research can address objectives to decrease diagnostic delay and improve patient outcomes in CDD.