mutation and gives rise to considering the KIF1β CMTD subtype as an independent entity, suggesting the need for reclassification. We recommend the use of exome sequencing for mutations difficult to identify using previously available tools.

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118265

Genetic markers of catamenial epilepsy in women

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Background and aims

To study the role of the sodium channel gene (SCN1A) in the development of catamenial epilepsy.

Methods

Molecular genetic studies with determination of the frequency of alleles of the SCN1A gene were performed in 100 women examined by us. Of these, 60 patients with catamenial epilepsy, 20 with symptomatic epilepsy. The control group consisted of 20 healthy women of the same age. The alleles of the SCN1A gene of the polymorphic locus D2S2330 and D2S124 were studied.

Results

The frequency of occurrence of the alleles D2S2330*9, D2S2330*12 and D2S2330*11 was greater and reached from 10 to 15%,respectively.And we found the D2S2330*10 allele in only one patient with symptomatic epilepsy.In the control group,the alleles D2S2330*10 and D2S2330*12 were more common,which makes it possible to consider their presence as a criterion of reduced risk for catamenial epilepsy.The most common in the main group of patients were the D2S124*4 loci (28.3%) and D2S124*5 (21.6%),while the D2S124*5 locus in the control group was found in 10% of cases in the control group,patients with DNA loci D2S214*2 (25%) and D2S214*3 (45%) were more common,while in the group of patients with catamenial epilepsy they were less common,locus D2S214*2–15% and locus D2S214*3–21.6%.

Conclusions

The relationship between the development of catamenial epilepsy and the SCN1A gene polymorphism at the D2S2330 and D2S214 loci was revealed,which does not exclude canalopathy in the pathogenesis of this type of epilepsy.This,in turn,plays an important role in the development of methods for predicting and differentiating therapy for catamenial epilepsy.

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Sex-specific differences in transcriptomic profiles and cellular characteristics of oligodendrocyte precursor cells

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Background and aims

Oligodendrocyte precursor cells (OPCs) regulate the neuronal system in various ways and play crucial roles in brain homeostasis. Although recent studies have shown regional diversity and heterogeneity of OPCs, sex-related differences in OPCs are largely unknown. The purpose of present study was to investigate sex-specific differences in transcriptomic profiles and cellular characteristics of OPCs.

Methods

For in vitro experiments, we prepared primary OPC culture obtained from neonatal rats. Sex identification was performed by PCR using sex-determining region Y gene. First, we evaluated the capacity of proliferation, migration, and differentiation in OPCs. Second, we examined the ischemic tolerance in OPCs after oxygen glucose deprivation. Third, we assessed the effects of OPC-derived factors on the integrity of blood brain barrier (BBB). For in vivo experiments, we conducted BrdU incorporation assay to assess the proliferation and differentiation of OPCs in adult mice. Finally, we performed RNA-seq analysis to investigate the sexual dimorphism in transcriptome profiles of cultured OPCs.

Results

Female OPCs have a higher capacity for proliferation and migration. In addition, female OPCs are more resistant to ischemic stress, and can enhance the BBB integrity. Meanwhile, male OPCs have a higher capacity for differentiation and myelination. RNA-seq analysis revealed substantial transcriptomic differences in OPCs.

Conclusions

The present study demonstrates sex-related differences in the cellular characteristics and transcriptional profiles of OPCs. Our findings may help to better understand the pathomechanisms of neurological and psychiatric diseases with sexual dimorphism.

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Expanding the disease spectrum of recessive ECEL1 mutations beyond distal arthrogryposis phenotype

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Background and aims

Distal arthrogryposis type 5D (DA5D) is a rare autosomal recessive disorder with wide phenotypic spectrum. DA5D is caused by mutations in ECEL1.

Methods

We describe two families (3 patients) with novel ECEL1 gene mutations detected by Next generation sequencing (NGS).

Results

Patient 1:12-year-old boy, born to consanguineous parents. He presented with birth asphyxia, motor developmental delay, severe contractures of fingers with webbing, pes planus, kyphoscoliosis, high arched palate, micrognatia, undescended testis, hypophonic speech with nasal twang, asymmetric ptosis, absent adductor digiti minimi, bifacial and distal LL weakness. Muscle MRI revealed asymmetric fatty infiltration in gluteus maximus, rectus femoris, hamstrings and gastrocnemius. Patients 2,3: 17-year-old monozygotic twins born to consanguineous parents and presented with motor development delay, white hairlock, small low set ears, elbow and wrist contractures, hypotonia, thinning and weakness of