to phenobarbital was not successful and lamotrigine worsened the myoclonic jerks. Different neuroleptic drugs failed to improve the psychiatric condition due to adverse effects or insufficient antipsychotic effect.
Conclusion: Our patient suffered from JME. Since JME cannot be linked to chromosome 22 so far and since there were no reports of myoclonic epilepsy in del(22)(q11.2) it seems rather unlikely that the chromosomal aberration accounts for JME in our patient.

P523
Epilepsy and Polymicrogyria in a Patient with 22q11 Deletion Syndrome
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Purpose: Chromosome 22q11.2 deletion syndrome is the most common micro deletion syndrome. It occurs at a frequency of 1/4000 live births and causes a wide spectrum of clinical disorders.
Method: We describe a case illustrating the variety of clinical associations with del 22q11, reporting the association with polymicrogiria.
Results: A 3 year old boy is the first child of healthy unrelated parents and the result of a normal pregnancy and delivery. At birth central cyanosis was noticed. He was submitted to a Blalock – Taussig procedure after the diagnosis of Fallot tetralogy. At 7 months total correction was carried out, with success. Other unusual findings included peculiar facies, mild retrognathia and bilateral preauricular appendices. At 4 months a mild left hemiparesis was described. He started walking at 2 years old, but unsteadily. Global delay was present with receptive and expressive language delay. By age 18 months he had seizures with fever, and at 2 years old focal apiretic seizures. His EEG was abnormal with a right focus and continuous spike-wave during sleep. He was started on sodium valproate. The seizures have been controlled and follow-up EEGs are improved. Cranial MRI showed right frontal and temporal polymicrogiria and deficient opercularisation of the right sylvian fissure. Routine chromosomal examination was normal (46,XY) but molecular cytogenetic testing by fluorescence in situ hybridisation (FISH) demonstrated a chromosomal 22q11.2 deletion. FISH studies performed on the parents were normal.
Conclusion: We stress the importance of looking for an underlying central nervous system anomaly in patients with 22q11.2 deletion. It will be advisable to include the screen of this deletion in the workup of neuronal migration disorders.

P524
Genetic Heterogeneity in Colombian Families with Generalised Epilepsy with Febrile Seizures Plus (GEFS+)
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Purpose: GEFS+ is a genetic and phenotypic heterogeneous disorder characterised by nonfebrile seizures and febrile seizures (FS) persisting beyond age 6 years. Mode of inheritance is autosomal dominant.
Method: Two extended Colombian pedigrees with GEFS+ were characterised phenotypically and linkage analysis was done.
Results: The first family showed a clear dominant mode of inheritance and the second family presented a more likely recessive mode of inheritance. FS loci were ruled out in these families (FEB1, FEB2, FEB3, FEB4). The first family showed a linkage to the SCN1A gene and currently the sequencing of the 26 exons of SCN1A is underway. The second pedigree had only a weak positive lod score for marker locus D2S129 and a new tightly linked marker (D2S127) is also currently being evaluated. A genome scan will be run in this family if the last candidate locus is excluded. Clinical symptoms involve onset of FS in a range of 3 months to 7 years. Age of onset for afebrile seizures presented a range of 1 year to 33 years. 7 individuals presented focal seizures instead of generalised tonic clonic seizures. Some individuals with mental retardation were present in one of the pedigrees.
Conclusion: Mutations in different genes are causing GEFS+ in Colombian families. In one family linkage to SCN1A could be found and a recessive mode of inheritance could be possible in the second family. Phenotypic heterogeneity is also present in these families.

P525
Phenotype and Genetic Simulation Analysis of Two Colombian Families with Generalised Epilepsy with Febrile Seizures plus (GEFS+)
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Purpose: GEFS+ is a phenotypic and genotypic heterogeneous disorder with febrile seizures (FS) persisting beyond 6 years of age and afebrile seizures. Phenotypic description and genetic simulation analysis of 2 affected families were completed.
Method: Extended pedigrees and individual anamnesis of nearly all family members, so that power simulation analysis could be undertaken.
Results: 3 of 9 members with seizures from the first family had GEFS+:FS and generalised tonic clonic seizures (GTCS) with frequency ranging from single to countless, starting at a mean age of 12 months and ending at 1, 8 and 19 years of age. Afebrile seizures were GTCS starting at 1, 14 and 19 years with frequency of 2 to countless. 2 members had only FS and 4 had afebrile GTCS. 5 of 9 members of the second family had the GEFS+ phenotype: FS started at a mean age of 3,6 years of age and ended beyond 6 years of age in three individuals, afebrile seizures started at an age range from 10 months to 14 years and seizure frequency was wide-ranging from 3 to countless. The remaining 4 family members had afebrile seizures. The simulation analysis for 2 families showed the following scores: families 1 and 2, with Z max of 2.78, 2.95, and an average of 1.98 and 1.82 respectively. These families together showed a Z max of 5.73 and an average of 3.78. The probability to find a lod score of 3 when homogeneity was assumed was 75%.
Conclusion: Heterogeneous phenotype was observed in these families and simulation analysis encourages mutation studies.

P526
Mild Epilepsy and Frequent Spontaneous Remission in a Large Kindred with Tuberous Sclerosis and a Missense Mutation in TSC2
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Purpose: Tuberous sclerosis (TSC) is an autosomal dominant disorder caused by mutations in the TSC1(9q34) or TSC2(16p13) genes. We analysed the genotype/phenotype correlations in an extended kinship presenting with familial epilepsy and found to have exceptionally mild TSC2.
Method: We studied a 3-generation family comprising 202 individuals. Available individuals underwent multisystem evaluations. 58 family members were genotyped for TSC1 and TSC2 markers.