

I am a PhD student of Bernd Neubauer and work in the field of genetic determined epilepsies within the Euroepinomics core CoGIE. My mayor focus, the benign epilepsy with centrotemporal spikes (BECTS) was first described by Martinus Rulandus in 1597 (Van Huffelen et al., 1989). The syndrome is now named rolandic epilepsy (RE) because of the characteristic features of partial seizures involving the region around the lower portion of the Rolandic fissure. RE is one of the most common epilepsy syndrome in children, accounting for about 15 % of epilepsies beginning before the age of 16 years (Freitag et al., 2001; Shinnar & Pellock, 2002). Rolandic epilepsy (RE) is has a complex genetic inheritance (Bali et al . 2005, Strug et al., 2009). The electroencephalography (EEG) endophenotype of centrotemporal sharp waves (CTS) is the hallmark of RE. The EEG is also present in seizure unaffected family members and is inherited in an autosomal-dominant manner (Heijbel et al., 1975, Bali et al . 2007). Despite the strong genetic contribution to the EEG trait, the etiology of epilepsies with centrotemporal spikes is largely unknown.

Motivated by recent findings for idiopathic generalized epilepsies (IGE) that rare recurrent microdeletions represent strong risk factors for IGE (Helbig et al., 2009; DeKovel et al., 2011) we have also performed a genome-wide copy number variation (CNV) analysis for our RE samples. It seems that we have made very interesting preliminary results. To validate and to analyze these the results in more detail we want to apply the "Array-comparative genomic hybridization" (array-CGH) method. Therefore, we want to collaborate with the Mefford group which are worldwide known specialist for this technology and a associated partner of the RES CRP. We don't just want to establish this collaboration by simply sending DNA samples to the group. Instead we would prefer a scientific exchange where I will have short term visit at the Mefford lab, to learn the technology they apply and to establish the new collaboration by meeting the lab members. The total duration will be 14-21 days.