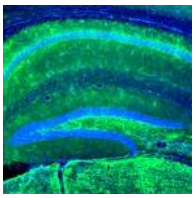


Functional genomic variation in the epilepsies (EuroEPINOMICS)

Genetics of the Rare Epilepsy Syndromes

EUROPEAN SCIENCE FOUNDATION

EUROCORES Networking Activity

Final Report: EuroEPINOMICS data analysis meeting

A) Summary

The EuroEPINOMICS data analysis meeting successfully lead to (1) update the members on the quality and scope of sequencing data, which was generated in different centres across the CRPs, (2) compare whole genome approaches with exome sequencing strategies, (3) compare pilot studies on genomes sequenced across different centers, (4) find best practices for separating *bona fide* variations from sequencing artifacts, (5) find reliable data sources for established variations relevant to epilepsies, and (6) discuss further sequencing strategies. Also, this meeting enabled the consortium to find a central data repository for all sequencing projects within EuroEPINOMICS.

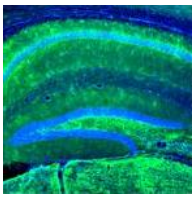
B) Final Programm

05.07.2012

12:00	Get Together - Lunch
14:00	Start of the meeting - Welcome & Introduction (Rudi Balling)
14:15	Overview of CRP – COGIE/State of the project (Holger Lerche)
14.30	Sequencing status COGIE at Cologne Center for Gen. (Holger Thiele)
15.10	Sequencing status RES at Sanger (Aarno Palotie)
15.50	Preliminary Results of RES (Arvid Suls)
16:50	Whole Genome Analysis – ISB/LSCB (Patrick May)
17:30	Exomes, X-omes, and targeted arrays for homozygosity centered recessive mutation screening in Dutch patients with Epileptic Encephalopathies. (Bobby Koeleman)
18:20	Concluding discussion (led by Holger Lerche)
19:00	End of first session
20:00	Dinner

06.07.2012

9:00	Epilepsiome data set collection
9:30	Exome repository
10:00	Phenoytping in RES (Sarah Weckhuysen/Johanna Jähn)
10:40	EpiPGX Case Record Form-implemetation in Filemaker for phenotype collection (Roland Krause)
11:20	SNAP2, a program for the screening for non-acceptable polymorphisms (Reinhard Schneider)
12:00	Structural biology and network analysis of identified variants (Antonio del Sol)
13:00	Lunch break
14:00	Social media activities (Roland Krause)
15:00	Closing discussion (led by Holger Lerche)



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C) Description of the scientific content of the event

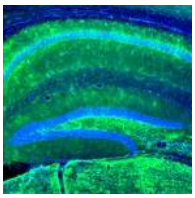
(1) Update of members on the quality and scope of sequencing data: Presentation of first sequencing data of the COGIe and RES CRPs.
(2) Comparison of whole genome approaches with exome approaches: The WGS results of COGIE were compared to the first WES results of the RES consortium.
(3) Comparison of pilot studies on genomes sequenced across different centers: The data acquired by the EuroEPINOMICS consortium were compared to Genome data achieved by different centers.
(4) Achievement of best practices for separating *bona fide* variations from sequencing artifacts: Strategies for distinguishing artifacts from true variations were discussed.
(5) Reliable data sources for established variations relevant to epilepsies: To filter variations for variants relevant to epilepsies, different data sources were discussed and interchanged between the different partners.
(6) Discussion of further sequencing strategies: The further process of sequencing within the different CRPs was discussed.

D) Assessment of the results and impact of the event on the EUROCORES Programm

The EuroEPINOMICS data analysis meeting resulted in a successful update of the consortium members on the quality and scope of sequencing data, which was generated in different centres across the CRPs. The two approaches whole genome sequencing and exome sequencing were compared as well as the results of different pilot studies on genomes sequenced across different centers. Reliable data sources for established variations relevant to epilepsies were set up, and further sequencing strategies were discussed. Also, this meeting enabled the consortium to find a central data repository for all sequencing projects within EuroEPINOMICS.

E) List of speakers and participants

1. Aarno Palotie
2. Anna-Elina Lehesjoki
3. Antonio del Sol
4. Arvid Suls
5. Bernd Neubauer
6. Bobby Koeleman
7. Enrico Glaab
8. Eva Reinthaler
9. Fritz Zimprich
10. Holger Lerche
11. Holger Thiele
12. Ingo Helbig
13. Jan Larsen
14. Johanna Jähn
15. José Serratosa
16. Kamel Jabbari
17. Kirsten Roomp
18. Patrick May
19. Peter de Jonghe
20. Peter Nürnberg
21. Reinhard Schneider
22. Roland Krause
23. Rudi Balling
24. S. Hande Çağlayan
25. Sarah Weckhuysen
26. Snezana Maljevic
27. Katharina Pernhorst
28. Ann-Kathrin Kriegel
29. Konrad Dębski
30. Philip Holmgren
31. Sunay Usluer
32. Julian Schubert
33. Dennis Lal
34. Abhimanyu Krishna
35. Gökhan Ertaylan
36. Chris Cotsapas
37. Sarah Killcoyne
38. Padhraig Gormly
39. Nikos Vlassis



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