

# MULTI-OMICS ANALYSIS

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# Current and future work

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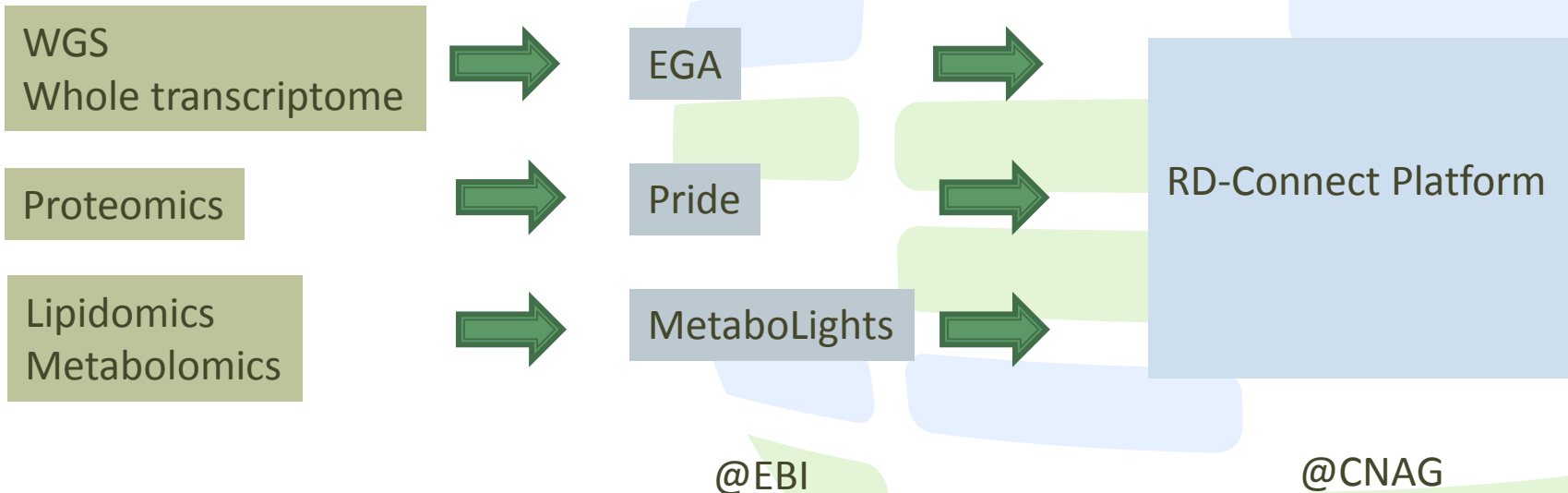
- Multi-omics data management
- Multi-omics tool development
  - RNA-Seq + WGS: diagnostic and disease modifiers
  - RNA-Seq + proteomics + metabolomics + proteomics: biomarkers and patient stratification
- Integration of tools into the RD-Connect platform



# Multi-omics data management

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- Individual-level data with phenotypes (patient-centered)
- Primary source @ EBI
- Integration between sources in RD-Connect platforms





# Multi-omics data management

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- Collection of relevant meta-data (from Neuromics) slow but ongoing
- Direct submission systems @ EBI in place
- Submission portal RD-Connect in development



# Use cases

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- Number of multi-omics use cases limited: only few human use cases with paired –omics data, mostly mouse
- Nature of use cases different and thus different requirements
  - Diagnostic
  - Disease modifiers
  - Patient Stratification
  - Biomarkers (disease progression and therapeutic response)



# RNA-seq

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- Analysis pipeline (nextpresso, Grana et al.)
- Expression level of genes and transcripts
- In progress: variant detection
- Whole genome and transcriptome integration use case:
  - Disease modifiers in Congenital Myasthenic Syndrome

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# Integration in RD-Connect platform

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- Not trivial: platform currently focused on diagnostics, not on biomarkers or disease modifiers
- Use cases heterogeneous
- From analysis of individuals (individual families) to groups of individuals
- Customizable workflows

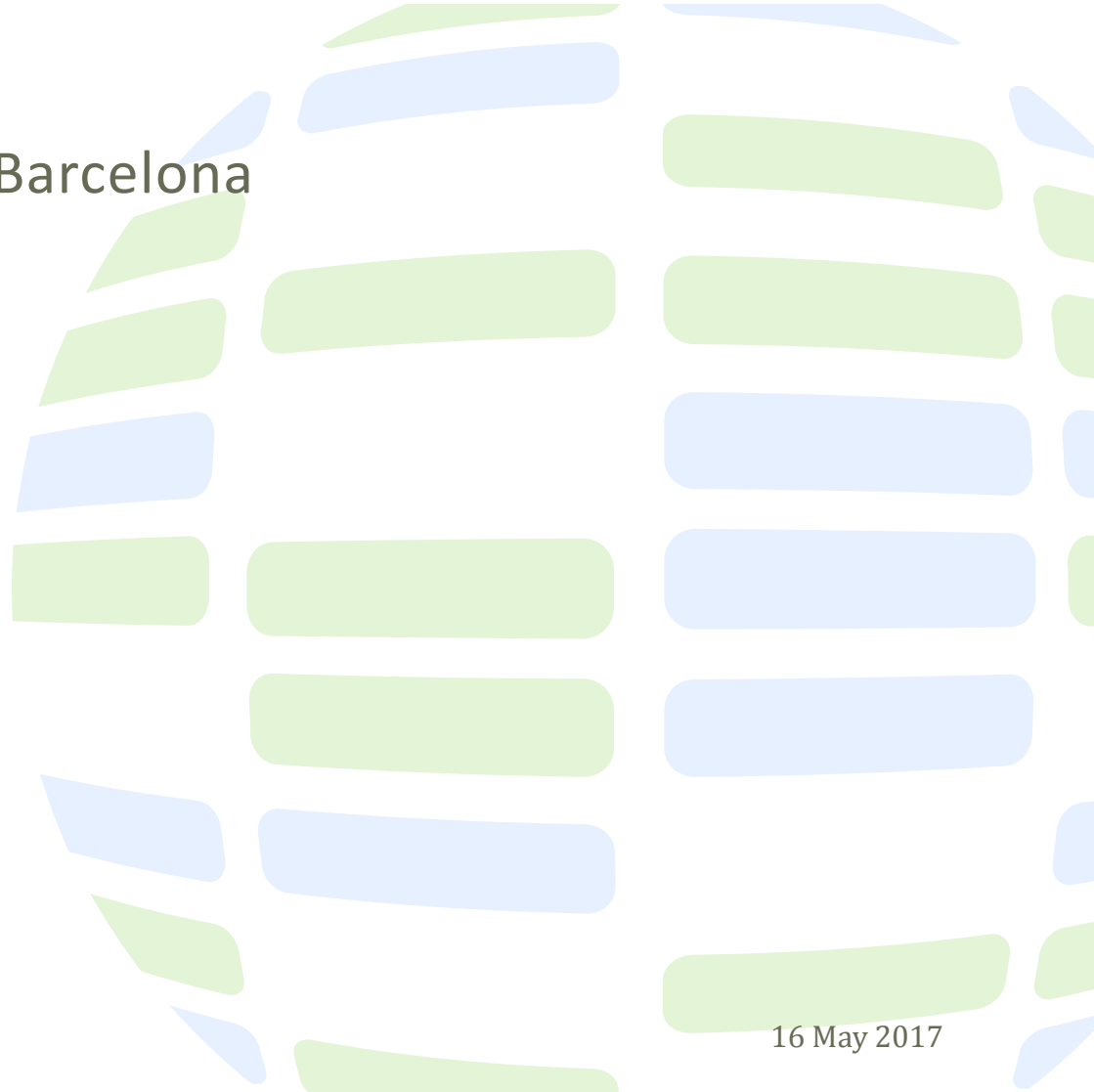




# Further strategy definition

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- Multi-omics task force
- Jamboree (July 10-12), Barcelona







# Acknowledgements

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EURen  Omics

Neur  Omics

RD  Connect



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