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## PMBIO Module 07

### Clinical. Clinical Applications

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Introduction to bioinformatics for DNA and RNA sequence  
analysis (IBDR01)

29 October - 2 November, 2018

Glasgow



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# Learning objectives of module 07: Clinical

- **Key concepts:** Clinical interpretation of variants, variant interpretation types (predisposing, predictive, predisposing, diagnostic), variant interpretation guidelines, clinical applications of passenger somatic variants (early detection, minimal residual disease tracking, personalized vaccines).
- Prioritize germline variants to identify putative de novo and clinically relevant mutations
- Germline pathogenicity assessment (demonstrate concepts with some example variants using ClinGen Variant Curation Interface)
- Biological function or oncogenicity (demonstrate concepts by uploading a filtered VCF into CRAVAT)
- Somatic clinical actionability (demonstrate concepts with some example variants using CIViC)