



**15th International Symposium on Variants in the Genome: detection,
sequencing & interpretation
25 - 27 May 2020
Saint Bernadin Conference Centre and Resort
Portoroz, Slovenia**

**DRAFT PROGRAM at 17 Feb. 2020
Program may be rearranged**

TBA = to be announced, TBC = to be confirmed

All Plenary Sessions will be held in the Emerald Room

Monday 25th May

8.30 - 14.00 Exhibitor Bump In

8.00 - 11.00 **REGISTRATION - Emerald Room Foyer**

9.00 - 10.30 **QIAGEN BIOINFORMATICS
HANDS - ON WORKSHOP**

Room: Emerald Room 1
(Included in registration)

**Introduction to variant interpretation
and reporting using an integrated
analysis pipeline**

Dr. Ruth Burton
QIAGEN Advanced Genomics, Manchester,
UK

PRE-MEETING WORKSHOP 2
TBA

Room: Emerald Room 2
(Included in registration)

TBA

10.30 - 11.00 **Coffee Break**

11.00 - 11.05 **INTRODUCTION & WELCOME**

Prof. Johan T. den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands

11.05 - 13.00 **PLENARY SESSION 1**
New and Emerging Technologies

11.05 - 11.35 **Experiences with Long-Read Sequencing**

A/Prof. Alexander Hoischen
Radboud Univ. Medical Centre, Nijmegen, Netherlands

11.35 - 12.05 **TBA**

12.05 - 12.40 **Presentations from Selected Abstracts**

12.40 - 13.00 **SPONSORED PRESENTATION**

TBA

TBA

13.00 - 14.00 **Lunch**
Room: TBA

14.00 - 15.30 **PLENARY SESSION 2**
Clinical Applications

14.00 - 14.30 **TBA**

14.30 - 15.00 **TBA**

15.00 - 15.30 **Presentations from Selected Abstracts**

15.30 - 16.10 **Poster Session 1**
Room: Emerald Room Foyer
Odd numbers to present: POS01, POS03 etc.

16.10 - 18.10 **PLENARY SESSION 3**
Theme TBA

TBA

18.10 - 18.20 **Assemble for Group Photograph**

18.30 - 20.30 **NETWORKING RECEPTION**
Grand Hotel St. Bernadin

Tuesday 26th May

8.30 - 10.30 **PLENARY SESSION 4**
Frontiers of Applications

8.30 – 9.00 **Editing (CRISPR/Cas9): Application for functional tests**

Dr. Rocia Acunia Hidalgo
Co-Founder & CTO, Nostos Genomics, Berlin, Germany

9.00 - 9.30 **TBA**

9.30 - 10.00 **Presentations from Selected Abstracts**

10.00 - 10.30 **SPONSORED PRESENTATION**

TBA

10.30 - 11.00 **Coffee Break**

11.00 - 13.00 **PLENARY SESSION 5**
Pipelines, bottlenecks and other challenges

11.00 - 11.30 **When the NGS pipeline fails: solving exceptional cases**

Prof. Johan T. den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands

11.30 - 12.00 **TBA**

12.00 - 12.30 **Presentations from Selected Abstracts**

12.30 - 13.00 **Genomic Technologies: validity, utility and responsibility to the public**

Genomic technologies provide a spectrum of information, from whimsical recreation genetics to life-changing clinical diagnosis. The analytical validity, and clinical or non-clinical validity and utility, varies widely in this spectrum. What responsibility do we have as professionals to educate the public about these issues? How should we do so?

Moderator: TBA

Panelists: TBA

13.00 - 14.00 **Lunch Break**
Room: TBA

14.00 - 15.30 **THINK TANK**

Calling all symposium attendees! Please send any topics you wish discussed in an open floor format to the Event Manager before the event. We will also send an email to all registered delegates before the event asking for topics. The scientific program committee will prepare the best topics in advance and crowdsource via *live polling* the topics to discuss at the beginning of this session. **YOU, vote the topics to discuss.** This will be your opportunity to discuss those burning issues, hear solutions from your peers, be exposed to a range of perspectives and experience for yourself a **fully immersive conference session format.** So make sure you come with your innovation and problem-solvers hat on!

15.30 - 16.10 **Poster Session 2**
Room: Emerald Room Lobby
Even numbers to present: POS02, POS04 etc.

16.10 - 18.00 **PLENARY SESSION 6**
Variant Interpretation in Practice

16.10 - 17.30 **ACMG Variant Classification/Interpretation Workshop**

Dr. Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

17.30 - 18.00 **Update on the ClinGen SVI recommendations to the ACMG-AMP system**
(how to use/modify existing ACMG-AMP criteria)

Speaker TBA

18.00 **DAY END - Evening at leisure**

Wednesday 27th May

8.30 - 10.30 **PLENARY SESSION 7**
Theme TBA

8.30 - 9.00 **TBA**

9.00 - 10.00 **TBA**

10.00 - 10.30 **SPONSORED PRESENTATION**

TBA

10.30 - 11.00 **Coffee Break**

11.00 - 14.00 **PLENARY SESSION 8**
The Power of Software, Databases and Bioinformatics

11.00 - 11.30 **The MANE project: Identifying reference transcripts in human protein coding genes**

Dr. Jane Loveland
Annotation Project Leader, EMBL-EBI, Hinton, Cambridge UK

11.30 - 12.00	Ensembl Dr. Astrid Gall <i>Ensembl Outreach Officer, EBI, Hinton, Cambridge UK</i>
12.00 - 12.30	UCSC Genome Browser Dr. Robert Kuhn <i>Associate Director, UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA</i>
12.30 - 13.00	CAMPFIRE SESSION Bioinformatics Tools: deficiencies and aspirations A discussion of the shortcomings of current bioinformatics tools. Open floor format. Facilitator TBA
13.00 - 14.00	Lunch Break Room: TBA
13.50 - 15.20	PLENARY SESSION 9 Genomic Data Sharing
13.50 - 14.30	GA4GH Variation Representation Schema Dr Reece Hart
14.30 - 14.50	TBA
14.50 - 15.20	SPONSORED PRESENTATION TBA
15.20 - 15.50	Coffee Break
15.50 - 17.00	PLENARY SESSION 10 Theme TBA
15.50 - 16.20	Presentations from Selected Abstracts
16.20 - 16.50	Is it time to re-imagine genomics? Speaker TBA
16.50- 17.00	SUMMARY - CLOSING Johan T. den Dunnen
17.00	SYMPOSIUM END