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# Program

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## Syndromdiagnostikk

**Dato** Onsdag 26. og torsdag 27. august 2026 | **Sted** Bikuben kurs og konferansesenter, Haukeland sykehus

### Dag 1 – Onsdag 26. august

Tidspunkt	Tema	Underviser
08:30 – 09:00	Registrering	-
09:00 – 09:05	Velkommen	Siren Berland
09:05 – 09:35	Genetiske analyser; hvem, hva og hvordan	Cecilie Rustad
09:35 – 10:00	Imprintingsykdommer	Siren Berland
10:00 – 10:10	Coffee break	-
10:10 – 10:30	Nytt fra sjeldenfeltet	Stein Are Aksnes, Sjeldensenteret
10:40 – 11:00	Mitochondrial cases	Kristin Varhaug
11:00 – 11:30	The use of 3D facial capturing in dysmorphology	Jeroen Breckpot, Leuven
11:30 – 12:30	Lunsj	-
12:30 – 13:00	Unexpected extreme variability in monogenic syndromes complicating variant interpretation and counselling	Emilia Bijlsma, Leiden
13:00 – 14:00	Cases	-
14:00 – 14:30	Post-exome project	Sofia Douzgou Houge
14:30 – 14:45	Break with coffee and snack	-
14:45 – 15:15	Non-invasive PGT	Han Brunner, Nijmegen
15:15 – 17:00	Cases	-

**Dag 2 – Torsdag 27. august**

<b>Tidspunkt</b>	<b>Tema</b>	<b>Underviser</b>
08:00 – 08:20	Registration	-
08:20 – 08:30	Welcome	Charlotte von der Lippe
08:30 – 09:00	tba	Jeroen Breckpot, Leuven
09:00 – 09:30	Amniotic bands – still a mystery?	Dian Donnai, Manchester
09:30 – 09:45	Break with coffee & snack	-
09:45 – 12:00	Cases	-
12:00 – 13:00	Lunch	-
13:00 – 14:15	LIS award and cases	-
14:15 – 14:30	Break with coffee	-
14:30 – 15:00	Examples of gain of function variants and loss of function variants and loss of function variants in the same gene (and other instructive cases)	Emilia Bijlsma, Leiden
15:00 – 15:30	Can we improve our gene pool? And should we?	Han Brunner, Nijmegen
15:30 – 16:00	Cases and closure	-