Program

Syndromdiagnostikk

Dato: Onsdag 27. og torsdag 28. august 2025 | **Sted:** Auditoriet Ulefoss, Sykehuset Telemark

Dag 1 – Onsdag 27. august

Tidspunkt	Tema	Underviser
09:00 – 09:30	Registrering	-
09:30 – 09:35	Velkommen	Cecilie F. Rustad og Charlotte von der Lippe, Skien
09:35 – 09:45	Nytt fra sjeldenfeltet	Stein Are Aksnes, NSSD
09:45 – 10:15	Genetiske analyser; hvem, hva og hvordan	Siren Berland, Bergen
10:15 – 10:40	Utilsiktede funn	Kristian Tveten, Skien
10:40 – 11:10	Brukermedvirkning (Norsk)	Charlotte von der Lippe, Skien
11:10 – 11:30	Variant med usikker signifikans (VUS)	Kristian Tveten, Skien
11:30 – 12:30	Lunsj	-
12:30 – 13:00	Mutations in non-coding genes	Emilia Bijlsma, Leiden
13:00 – 14:00	Cases	Participants
14:00 – 14:30	Mechanisms underlying phenotypic variability in 22q11.2 deletion syndrome and guidelines for follow-up	Jeroen Breckpot, Leuven
14:30 – 14:45	Break with coffee and snack	-
14:45 – 15:15	Why and how genetic therapies work (but not always very well)	Han Brunner, Nijmegen
15:15 – 17:00	Cases	Participants

Dag 2 – Torsdag 28. august

Tidspunkt	Tema	Underviser
08:00 – 08:20	Registration	-
08:20 – 08:30	Welcome	Siren Berland, Bergen
08:30 – 09:00	Differences in genetic background in syndromic and non-syndromic congenital heart defects: who should we test?	Jeroen Breckpot, Leuven
09:00 – 09:30	Monozygotic twins are not simple	Dian Donnai, Manchester
09:30 – 09:45	Break with coffee & snack	-
09:45 – 12:00	Cases	Participants
12:00 – 13:00	Lunch	-
13:00 – 14:15	Cases	Participants
14:15 – 14:30	Break with coffee	-
14:30 – 15:00	Instructive cases	Emilia Bijlsma, Leiden
15:00 – 15:30	Why are some syndromes more common than others?	Han Brunner, Nijmegen
15:30 – 16:00	Cases and closure	Participants and meeting organizers