Nasjonal kompetansetjeneste for SJELDNE DIAGNOSER

Haukeland University Hospital

SYNDROMDIAGNOSTIKK-MØTET 2024

Storstuen, Kurs- og konferansesenteret i Bikuben, Haukeland universitetssykehus
Tirsdag 27. og Onsdag 28. august 2024

Tuesday Aug 27th

08:00 – 08:15 Registrering

08:15 – 08:25 Velkommen v/ Siren Berland og Charlotte von der Lippe

08:25 – 08:35 *Nytt fra sjeldenfeltet*, NKSD

08:35 – 09:05 *Genetiske analyser; hvem, hva og hvordan*, Siren Berland, Bergen

09:05 – 09:35 *Hvordan forstå og håndtere genetisk analysesvar*, Cecilie Rustad, SSD og Charlotte von der Lippe, Skien

09:35 – 09:50 Break with coffee

09:50 - 10:15 *40 years of diagnosing Williams syndrome*, Emilia Bijlsma, Leiden

10:15 – 10:40 *Non-invasive prenatal testing. There is (much) more than the common trisomies,* Koen Devriendt, Leuven

10:40 – 11:00 Break

11:00 – 11:30 *What if we sequenced everybody all of the time?* Part 1 , Han Brunner, Nijmegen

11.30 – 12:00 Cases

12:00 – 13:00 Lunch

13:00 – 13:30 *Annerledesverden,* Hilde Trætteberg Serkland (No)

13:30 – 16:00 Cases, with break (coffee & snack)

Wednesday Aug 28th

08:00 – 08:20 Registration

08:20 – 08:30 Welcome, Siren Berland, Bergen and Trine Prescott, Skien

08:30 – 09:00 *What if we sequenced everybody all of the time?* Part 2 , Han Brunner, Nijmegen

09:00 – 09:30 *Mosaicism and genetic syndromes*, Koen Devriendt, Leuven

09:30 – 09:45 Break with coffee & snack

09:45 – 12:00 Cases

12:00 – 13:00 Lunch

13:10 – 13:40 *Can fetal teratogens mimic genetic disorders*? Dian Donnai, Manchester

13:40 – 14:10 *Instructive cases*, Emilia Bijlsma, Leiden

14:10 – 14:40 *The value of whole exome sequencing in prenatal diagnosis*, Koen Devriendt, Leuven

14:50 – 15:05 Break with coffee

15:05 – 15:30 *Munnhulefunn og syndromer*, TAKO senteret (No)

15:30 – 16:00 Cases with break (coffee/snacks)

Tentativt program. Kurset er gratis, men begrenset antall fordrer at programkomiteen forbeholder seg retten til prioritering av enkelte, samt bindende påmelding til mona.elisabeth.vetas@helse-bergen.no