

**DAY 1, Friday, September 23**

| <b>Talk</b>              | <b>Time</b> | <b>Presentation title</b> | <b>Speaker</b>  |              |
|--------------------------|-------------|---------------------------|---|--------------|
| <b>Chair: Dr. Canick</b> |             |                           |   |              |
| Talk #                   | 8:30-9:00   | 30                        | <b>REGISTRATION</b>                                     |              |
| 1                        | 9:00-9:30   | 30                        | Introduction and background                             | J. E. Haddow |
| 2                        | 9:30-9:45   | 15                        | What is medical screening?                              | N. Wald      |
| 3                        | 9:45-10:30  | 45                        | Principles of screening I                               | N. Wald      |
|                          | 10:30-10:50 |                           | <b>BREAK</b>  |              |
| 4                        | 10:50-11:35 | 45                        | Principles of screening II                              | N. Wald      |
| 5                        | 11:35-12:15 | 40                        | Reference data and distribution of markers              | G. Palomaki  |
|                          | 12:15-1:00  |                           | <b>LUNCH*</b>   |              |
| <b>Chair: Dr. Haddow</b> |             |                           |   |              |
| 6                        | 1:00-2:00   | 60                        | <b>WORKSHOP 1: computing univariate risks</b>           | Faculty      |
| 7                        | 2:00-2:30   | 30                        | Major defects identified by ONTD screening              | J. E. Haddow |
| 8                        | 2:30-3:30   | 60                        | Selected factors that affect screening                  | G. Palomaki  |
| 9                        | 3:30-3:45   | 15                        | NTD risk calculations                                   | G. Palomaki  |
|                          | 3:45-4:05   |                           | <b>BREAK</b>  |              |
| 10                       | 4:05-4:35   | 30                        | Interpreting amniotic fluid AFP and AChE results        | G. Knight    |
| 11                       | 4:35-5:20   | 45                        | Legal issues surrounding prenatal screening and patents | J.B. Haddow  |
|                          | 5:20        |                           | <b>QUESTIONS, ADJOURN</b>                               |              |

## DAY 2, Saturday, September 24

### Chair: Dr. Palomaki

|    |             |    |   |               |
|----|-------------|----|---|---------------|
| 12 | 8:00-8:25   | 25 | Ultrasound and neural tube defects  | A. Donnenfeld |
| 13 | 8:25-8:55   | 30 | Morbidity and mortality associated with Down syndrome and impact of screening on birth prevalence | J. E. Haddow  |
| 14 | 8:55-9:40   | 45 | Biology of serum markers as they relate to Down syndrome  | J. Canick     |
| 15 | 9:40-10:25  | 45 | Serum screening assays, part I  | G. Knight     |
|    | 10:25-10:45 |    | <b>BREAK</b>  |               |
| 16 | 10:45-11:15 | 30 | Serum screening assays part II  | G. Knight     |
| 17 | 11:15-11:45 | 30 | Relative performance of different combinations of markers in Down syndrome screening              | N. Wald       |
|    | 11:45-12:45 |    | <b>LUNCH*</b>   |               |

### Chair: Dr. Wald

|    |            |    |  |               |
|----|------------|----|--|---------------|
| 18 | 12:45-1:15 | 30 | Combining markers into a single risk estimate              | G. Palomaki   |
| 19 | 1:15-2:15  | 60 | Factors that affect multiple markers: an overview          | G. Palomaki   |
| 20 | 2:15-3:00  | 45 | <b>WORKSHOP 2: computing and using multivariate risks</b>  | Faculty       |
|    | 3:00-3:20  |    | <b>BREAK</b>   |               |
| 21 | 3:20-4:00  | 40 | <b>Concurrent sessions</b>                                 |               |
|    |            |    | (a) Ultrasound after a second trimester DS screen positive | A. Donnenfeld |
|    |            |    | (b) Epidemiologic monitoring of serum assays               | G. Knight     |
| 22 | 4:00-4:15  | 15 | Why first trimester screening?                             | J. Canick     |
| 23 | 4:15-4:45  | 30 | Measurement of nuchal translucency                         | A. Donnenfeld |
|    | 4:30       | 15 | <b>QUESTIONS, ADJOURN</b>                                  |               |

### Day 3, Sunday, September 25

Chair: Dr. Haddow

| Talk | Time        | Presentation title   | Speaker                    |
|------|-------------|--|----------------------------|
| 24   | 8:30-9:00   | 30 First trimester markers for Down syndrome   | G. Palomaki                |
| 25   | 9:00-9:30   | 30 Program implementation and protocols for first trimester  | G. Messerlian              |
| 26   | 9:30-10:00  | 20 The first trimester intervention trials   | G. Palomaki                |
| 27   | 10:00-10:35 | 35 Risk as a screening variable  | G. Knight                  |
|      | 10:35-10:55 | <b>BREAK</b>   |                            |
| 28   | 10:55-11:40 | 45 <b>CONCURRENT SESSIONS</b><br>(a) Proficiency testing for first trimester and integrated screening<br>(b) Pre-eclampsia | G. Knight<br>G. Messerlian |
| 29   | 11:40-12:10 | 30 The laboratory and NT measurement   | G. Palomaki                |
|      | 12:10-1:00  | <b>LUNCH</b>   |                            |
|      |             | <b>Chair: Dr. Canick</b>   |                            |
| 30   | 1:00-1:45   | 45 Integrated screening  | N. Wald                    |
| 31   | 1:45-2:00   | 15 Implementation of integrated screening  | G. Messerlian              |
| 32   | 2:00-2:30   | 30 <b>CONCURRENT SESSIONS</b><br>(a) Methods comparison<br>(b) Counseling implications                                     | G. Palomaki<br>E. Kloza    |
| 33   | 2:30-2:45   | 15 Serum integrated screening  | G. Knight                  |
|      | 2:45-3:05   | <b>BREAK</b>   |                            |
| 34   | 3:05-3:35   | 30 Screening policy and practical improvements   | N. Wald                    |
| 35   | 3:35-3:50   | 15 Sequential approaches using the integrated test   | N. Wald                    |
| 36   | 3:50-4:10   | 20 Implementing sequential screening   | G. Palomaki                |
| 37   | 4:10-4:40   | 30 Interpreting results for trisomy 18   | G. Palomaki                |
| 38   | 4:40-5:10   | 30 Undetectable uE3, SLOS, STSD: What do we do?  | J. E. Haddow               |

### Day 4, Monday, September 26

Chair: Dr. Messerlian

|    |             |   |             |
|----|-------------|---|-------------|
| 39 | 8:00-8:40   | 40 Introduction to molecular genetics                       | S. Richards |
| 40 | 8:40-9:20   | 40 Overview of massively parallel shotgun sequencing (MPSS) | J. Canick   |
| 41 | 9:20-10:00  | 40 Results of the INFANET study                             | G. Palomaki |
|    | 10:00-10:20 | <b>BREAK</b>  |             |
| 42 | 10:20-11:00 | 40 Considerations for implementation                        | G. Palomaki |
| 43 | 11:00-12:00 | 60 How might this change practice?                          | M. Mennuti  |
| 44 | 12:00-12:30 | 30 Round table discussion                                   | Faculty     |
|    | 12:30       | <b>ADJOURN</b>  |             |