



BUILDING THE FOUNDATION FOR GENOMIC MEDICINE FOR PATIENTS WITH RARE DISEASES

Organisé par la filière de santé AnDDI-Rares
Co-organisé avec l'Association Francophone de Génétique Clinique
Co-organisé avec l'Institut Imagine
Sixième séminaire international FHU – TRANSLAD

Jeudi 20 avril 2017

*Amphithéâtre 6ème étage
Institut Imagine
24, boulevard du Montparnasse
75015 PARIS*

Guests :

Pr Kym Boycott, Children's Hospital of Eastern Ontario (CHEO) (Canada)

Pr Heidi Rehm, Brigham and Women's Hospital and Harvard Medical School (Etats-Unis)

Pr Jill Clayton-Smith, Central Manchester University Hospitals (Angleterre)

PROGRAMME

MATINEE (modérateurs: Pr Laurence Faivre, Pr Benoit Arveiler)

- 10h00 – Introduction of the day, *Pr Stanislas Lyonnet*
- 10h15 – The evolving role of the Clinical Geneticist, *Pr Jill Clayton-Smith*
- 11h15 – Integrating Genomics into the Practice of Medicine, *Pr Heidi Rehm*
- 12h00 – The depth of rare genetic diseases: Strategies to identify the remaining genes and diseases, *Pr Kym Boycott*

- 12h45 – *Déjeuner buffet sur place*

APRES MIDI (modérateurs : Pr Damien Sanlaville, Pr Sylvie Odent)

- 14h00 – Présentation de ClinGen ressource, *Pr Heidi Rehm*



- Developing resources to support genomic medicine
- Variant interpretation guidelines, data sharing and resolving differences in variant classification
- Defining gene-disease validity and actionability
- 15h00 – IRDiRC and GA4GH: International efforts to advance rare disease discovery, *Prs Heidi Rehm & Kym Boycott*
 - The Matchmaker Exchange
- 15h30 – Therapeutic opportunities from genomic sequencing, *Pr Kym Boycott*
- 16h00 – Round table with the guests: Building the foundation for genomic medicine for patients with rare diseases, *Pr Jill Clayton-Smith, Pr Thierry Frebourg, Pr Jean-Louis Mandel, Pr Arnold Munnich, Jean-François Deleuze, Virginie Bros-Facer*
- 16h30- Conférence de clôture : Présentation of « plan Médecine Génomique », *Pr Yves Levy*

INSCRIPTION

Gratuite

Lien d'inscription : <https://goo.gl/forms/whnXi9wYcGN1UBNX2>

ACCES

Institut des Maladies Génétiques **Imagine** - 24, boulevard du Montparnasse 75015 Paris



Métro lignes 4, 6, 10, 12, 13, Stations : Duroc, Falguière, Montparnasse-Bienvenue