

	MONDAY 9/01	TUESDAY 10/01	WEDNESDAY 11/01	THURSDAY 12/01	FRIDAY 13/01
	Syndromes			Rare pediatric cancers	Diagnosis, societal and economic issues (Part1)
9.30 am	Speaker introduction by students (5')				
Talk1(45')	S. Lyonnet (Institut Imagine) Introduction to Rare Disease	C. Boileau (Hopital Bichat) Marfan syndrome: from various clinical avatars to current knowledge and	B. Gardie (EPHE) Hereditary erythrocytosis and hypoxia-related disease	T. Mercher (Gustave Roussy) Pediatric Acute Megakaryoblastic Leukemia	Nicolas Garcelon (Institut Imagine) Artificial intelligence on hospital data for phenotyping, screening and diagnosis of rare diseases patients
	Presentation by a student of a paper proposed by the speaker (10')				
	Discussion (15')				
10h45 am Break 15min					
11.00 am	Speaker introduction by students (5')				
Talk2(45')	H. de Thé (Collège de France) PML, the unexpected effector of acute promyelocytic leukemia cure	G. Canaud (Institut Necker) Targeted therapy for patients with PIK3CA-related overgrowth spectrum	E. Pasmant (Institut Cochin) Neurofibromatosis type 1: from genotype to phenotype	O. Ayrault (Institut Curie) Signaling pathway alterations In Medulloblastoma, the most common malignant pediatric brain tumor.	D. Stoppa Lyonnet (Institut Curie)
	Presentation by a student of a paper proposed by the speaker (10')				
	Discussion (15')				
12h15am Lunch break					
	Diseases of the immune system	Tumor predisposition syndromes		Therapeutic issues	Diagnosis, societal and economic issues (Part2)
13.30 pm	Speaker introduction by students (5')		POSTERS	Speaker introduction by students (5')	
Talk3(45')	F. Rieux Laucat (Institut Imagine) (Immunogénétique des maladies auto-immunes pédiatriques)	J. Soulier (Hopital Saint-louis) Genetic predisposition to marrow failure and leukemia		A. Buj bello (genethon)	C. Bourgain (Cermes3) Beyond technology. The making of genetic prediction
	Presentation by a student of a paper proposed by the speaker (10')			Presentation by a student of a paper proposed by the speaker (10')	
	Discussion (15')			Discussion (15')	
14h45 pm Break 15min					
15.00 pm	Speaker introduction by students (5')			Speaker introduction by students (5')	
Talk4(45')	Anne Puel (Institut Imagine) Autoimmune phenocopies of inborn errors of immunity underlying infectious diseases: ex type I IFNs and viral diseases	C. Colas (Institut Curie) Constitutionnal Mismatch Repair Deficiency : clinical and molecular aspects		T.T. Maciel (Institut Imagine) Physiopathology of hemoglobinopathies (thalassemia and sickle cell disease)	The place of families and associations in the care, diagnosis and research of rare diseases
	Presentation by a student of a paper proposed by the speaker (10')			Presentation by a student of a paper proposed by the speaker (10')	
	Discussion (15')			Discussion (15')	
end 16h15pm					