

## Tandem Meeting on Pediatric Cancer Predispositions in Bern, Switzerland

**7<sup>th</sup> Workshop of the European Consortium Care for CMMRD  
(C4CMMRD), January 29<sup>th</sup> - 30<sup>th</sup> 2025**

**&**

**SIOPE Host Genome WG Meeting, January 30<sup>th</sup> - 31<sup>st</sup> 2025**

**at the University Hospital INSELSPITAL, Bern, Switzerland & online (hybrid)**



Registration: [Tandem Meeting on Pediatric Cancer Predispositions Registration](#)

With generous support from:



## 7<sup>th</sup> Workshop of the European Consortium Care for CMMRD (C4CMMRD), January 29<sup>th</sup> - 30<sup>th</sup> 2025

C4CMMRD Day 1: Wednesday January 29 <sup>th</sup> afternoon session	
<b>Location</b>	University Hospital INSELSPITAL, Bern, Switzerland; Anna Seiler Haus, Rooms 006_007, U1, Freiburgstrasse 20, Bern
<b>Start:</b> <b>14:00-14:30</b>	<p><b>Organization:</b> Administrative management of C4CMMRD (<i>Léa Guerrini-Rousseau/ Chrystelle Colas/ Katharina Wimmer – 10min</i>)</p> <p>Database C4CMMRD : Childhood cancer predisposition network in Nordic countries (<i>Pauline Hoarau / Anna Poluha and Sara Sjogren – 20 min</i>)</p>
<b>14:30-15:00</b>	<p><b>New guidelines:</b> Presentation How could they be evaluated? (<i>Katharina Wimmer – Diagnostics / Chrystelle Colas – Counselling, QoL, Surveillance / Léa Guerrini-Rousseau – clinical management</i>)</p>
<b>15:00-16:00</b>	<p><b>Germline <i>POLE/POLD1</i> variants and “CMMRD like” phenotypes</b> (<i>Moderation: Chrystelle Colas</i>) Clinical introduction (<i>Chrystelle Colas - 5 min</i>) Molecular specificities, functional tests (<i>Albain Chansavang, Nadim Hamzoul, Julien Masliah Planchon-25 min</i>) Novel genomic features of <i>POLE/POLD1</i>-mutant tumors: the utility for tumor classification and discovery of new driver alleles (<i>Polina Shcherbakova-25 min</i>) Discussion &amp; Proposal for collaboration</p>
<b>16:00-16:15</b>	<b>Coffee &amp; Snacks</b>
<b>16:15-17:45</b> Room 006_007 Room 223 Room 225	<p><b>Working Groups</b></p> <ol style="list-style-type: none"> <li>1. Haematological Malignancies/T-LBL (<i>Katharina Wimmer / Jan Loeffen</i>)</li> <li>2. ICI treatment experiences (<i>Léa Guerrini-Rousseau</i>)</li> <li>3. Polyposis (<i>Chrystelle Colas</i>)</li> </ol>
<b>19:30-21:30</b>	<p><b>Dinner: Restaurant in Old Town</b> Location: Restaurant Rosengarten <a href="https://maps.app.goo.gl/Wq764GoGtizqWHcL9">https://maps.app.goo.gl/Wq764GoGtizqWHcL9</a> Alter Aargauerstalden 31b, 3006 Bern</p>

## C4CMMRD Day 2: Thursday January 30<sup>th</sup> morning session

<b>Location</b>	<b>University Hospital INSELSPITAL, Bern, Switzerland; Anna Seiler Haus, Rooms 006_007, U1, Freiburgstrasse 20, Bern</b>
<b>Start: 8:30-9:00</b>	Report from the 3 working groups and discussion ( <i>Katharina Wimmer / Jan Loeffen, Léa Guerrini-Rousseau, Chrystelle Colas – 10 mins each</i> )
<b>9:00-09:40</b>	<b>New data from C4CMMRD members</b> ( <i>Moderation: Amedeo Azizi</i> ) Medulloblastoma in CMMRD: pathology and clinical data ( <i>Amedeo Azizi and Christine Haberler - 20min</i> ) Report of an unusual CMMRD patient with breast and other cancers ( <i>Richard Gallon – 10min</i> ) Clinical and genetic characteristics of 14 paediatric patients with CMMRD syndrome from nine families ( <i>Maria Kurnikova – 10min</i> )
<b>9:40-10:10</b>	<b>Project updates and proposals</b> ( <i>Moderation: Marta Pineda</i> ) CMMRD screening program in Spain ( <i>Marta Pineda – 10 min</i> ) Classification of MMR variants found in CMMRD patients ( <i>Clara Ruiz-Ponte/Marta Pineda – 10 min</i> ) Collection of young adult patient with CRC ( <i>Clara Ruiz-Ponte – 10 min</i> )
<b>10:10-10:25</b>	<b>Coffee &amp; Snacks</b>
<b>10:25-10:55</b>	<b>Molecular aspects</b> ( <i>Moderation: Richard Gallon</i> ) Standardized NGS-based screening of CMMRD ( <i>Alex Duval -10min</i> ) Validation of a newly simplified multiplex PCR based MSI assay for the identification of CMMRD ( <i>Richard Gallon – 10 min</i> ) Organoid models ( <i>Marco Bruschi – 10min</i> )
<b>10:55-12:00</b>	<b>Prevention &amp; Treatment</b> ( <i>Moderation: Jacques Grill</i> ) Whole body MRI: the French experience ( <i>Léa Guerini-Rousseau – 10 min</i> ) Case report: incidental findings on whole body magnetic resonance imaging in CMMRD syndrome ( <i>Theodorus W. Kouwenberg – 10 min</i> ) Experience with MRI surveillance in CNS lesions in three children with CMMRD ( <i>Mette Jorgensen – 10 min</i> ) Discussion & Proposal for collaboration ( <i>10 min</i> )  Updates on immunotherapy risk stratification and immune combination for replication repair deficient hypermutant cancers. ( <i>Uri Tabori – 25 min</i> )
<b>12:00</b>	<b>Conclusion</b> ( <i>Katharina Wimmer</i> )
<b>12:00-12:45</b>	<b>Lunch break</b>

## SIOPE Host Genome WG Meeting, January 30<sup>th</sup> - 31<sup>st</sup> 2025

SIOPE HGWG Day 1: Thursday January 30 <sup>th</sup> afternoon session		
<b>Location</b>	University Hospital INSELSPITAL, Bern, Switzerland; Anna Seiler Haus, Rooms 006_007, U1, Freiburgstrasse 20, Bern	
<b>From 12:45</b>	Welcome coffee	
<b>Start: 13:00</b>	Welcome address	
<b>13:15-13:45</b>	Plenary session (talks with plenary discussion) Towards immune cancer interception using LNP RNA vaccines in cancer predisposition (talk by Uri Tabori from Toronto, Canada)	
<b>13:45-14:30</b>	Circulating tumor DNA for early cancer detection (talks by Kendra Maas from Heidelberg, Germany & Uri Tabori, Toronto, Canada)	
<b>14:30-15:15</b>	National and international multidisciplinary advisory boards (talks by Helène Antoine-Poirel from Brussels, Belgium & Franck Bourdeaut, Paris, France)	
<b>15:15-15:30</b>	Coffee & Snacks	
<b>15:30-15:45</b>	Introduction to workshops (15 mins)	
<b>15:45-19:00</b>	Workshops	
	<b>Location 1: Anna Seiler Haus Room 005</b>	<b>Location 2: Anna Seiler Haus Room 006_007</b>
<b>15:45-17:15</b>	Workshop 1: <b>Segmental PIK3CA/AKT related overgrowth syndromes</b> (screening, treatment & cancer surveillance) <i>Chair: Mark Davies; Guillaume Canaud</i>	Workshop 2: <b>Li Fraumeni Syndrome low risk variants</b> (variant interpretation and cancer surveillance) <i>Chair: Karin Wadt</i>
<b>17:15-17:30</b>	Coffee & Snacks	
	<b>Location 1: Room 005</b>	<b>Location 2: Room 006_007</b>
<b>17:30-19:00</b>	Workshop 3: <b>Bone sarcoma predispositions</b> (screening and cancer surveillance) <i>Chair: Franck Bourdeaut</i>	Workshop 4: <b>Rare non-syndromic predispositions to hematological malignancies</b> (e.g. ANKRD26, ERCC6L2: diagnosis, treatment & surveillance) <i>Chair: Nicolas Waespe</i>
<b>19:30-21:30</b>	Dinner: Restaurant in Old Town Location: Restaurant Volkshaus 1914 <a href="https://maps.app.goo.gl/ck9Yac51eRdSmzy87">https://maps.app.goo.gl/ck9Yac51eRdSmzy87</a> Zeughausgasse 9, 3011 Bern	

## SIOPE HGWG Day 2: Friday January 31<sup>st</sup> full day session

<b>Location</b>	<b>University Hospital INSELSPITAL, Bern, Switzerland; Anna Seiler Haus, Rooms 006_007, U1, Freiburgstrasse 20, Bern</b>
<b>8.30-10:00</b>	<b>Overview on current projects Part I</b> (6 talks, 15 mins each, 12+3) <ol style="list-style-type: none"> <li>1. KiTDS-Care: Assessing Care and Support Needs in CPS Families, Michaela Kühlen</li> <li>2. Survey on Cancer Predisposition Identification &amp; Observation (SCOPE), Jakica Čavar</li> <li>3. Close follow-up of NF-1 patients matters, Dries Ruttens</li> <li>4. How we care for individuals with CPS, Valentina Härter</li> <li>5. Genotype-phenotype correlation in Fanconi anemia group D1, Agata Pastorczak</li> <li>6. Incidental findings in pediatric Li Fraumeni, Madeleine Adams</li> </ol>
<b>10:00-10:15</b>	<b>Coffee &amp; Snacks</b>
<b>10:15-11:45</b>	<b>Workshop summaries and discussion</b> (4 summaries, 15 mins each, 15 mins at end for overall summary)
<b>11:45-12:45</b>	<b>Difficult cases</b> (4-6 presentations, 10-15 mins each with discussion) Robin de Putter: JMML/ NRAS ; Vita Ridola : SDHA & SMAD4; Tüller: Melanoma/LFS
<b>12:45-13:45</b>	<b>Lunch break</b>
<b>13:45-15:15</b>	<b>Overview on current projects Part II</b> (6 talks, 15 mins each, 12+3) <ol style="list-style-type: none"> <li>7. CYPHER - Children &amp; Young People Hereditary CPS, Emma Woodward</li> <li>8. CanCYP: cancer risk prediction in CPS gene alteration, Emma Woodward</li> <li>9. Telomere related disease in childhood cancer (TELO-STAGE), Anna Byrjalsen</li> <li>10. Germline mutational landscape in pediatric cancers, Edith Sepulchre</li> <li>11. RAPADILINO and other RECQL4-related Syndromes, Sonja Strang-Karlsson</li> <li>12. Genetic and epigenetic predisposition in familial and bilateral Wilms, Jenny Wegert</li> <li>13. (BRCA/PALB2 project, TBD)</li> </ol>
<b>15:15- 15:30</b>	<b>“My cancer predispositions clinic”: 2<sup>nd</sup> edition: Denmark</b> (Ulrik Stoltze/ Karin Wadt, Denmark; 15 mins)
<b>15:30- 16:00</b>	<b>Wrap-up and any other business</b> -SIOPE meeting in May 2025 (Budapest, Hungary) -Information from the board members of the WG
<b>16:00</b>	<b>END of meeting</b>
<b>16:00-16:30</b>	<b>Coffee &amp; Snacks</b>



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