Tuesday
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11:00 s.t.

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Spliceosomal factor mutations in human diseases

Recent advances in high throughput sequencing have revealed many new mutations underlying both inherited diseases and acquired conditions such as cancer. A surprising number of these mutations are in genes for components of the spliceosomes. I will focus on two diseases. First, autosomal recessive mutations in the minor spliceosomal snRNA U4atac lead to a spectrum of conditions affecting the body size, brain, skeletal and immune systems. Biochemical studies and mouse models will be discussed. Second, somatic mutations of several spliceosomal factors have been frequently found in bone marrow neoplasms, particularly myelodysplastic syndromes. Biochemical and gene expression alterations due to these mutations will be described.

Host: Reinhard Lührmann

Large Seminar Room, Administration Building
Max Planck Institute for Biophysical Chemistry, Am Fassberg 11, 37079 Göttingen