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PUMCH brings hope in rare diseases and Scoliosis patients

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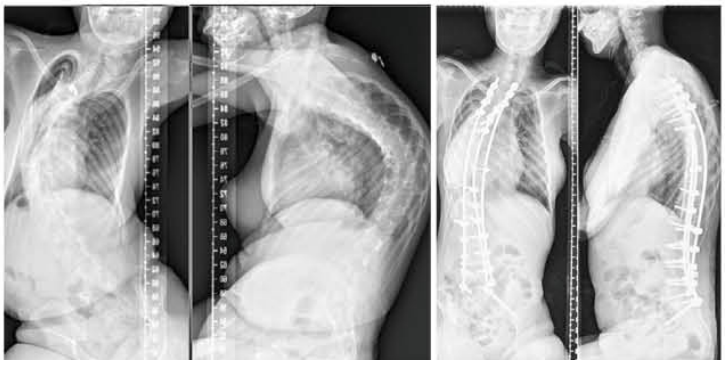
The 2018 PUMCH Medical Research award winner has achieved through surgery teams in the department of Orthopaedics by diagnosing 33 rare diseases involving 7 major systems in patients with scoliosis based on routine clinical diagnostic techniques, molecular biology, and genetic approaches



The Department of Orthopaedics of Peking Union Medical College Hospital (PUMCH), a leading scoliosis centre both in China and abroad, has successfully treated more than 200 patients with rare diseases accompanied by scoliosis in the past 15 years. This achievement has won the department the 2018 Peking Union Medical College Hospital Medical Research Achievement Award (second grade).

Rare diseases with accompanying scoliosis are extremely rare. The Department of Orthopaedics of PUMCH has been receiving such patients since 2000. Through multidisciplinary cooperation, the surgery teams in the Department of Orthopaedics have diagnosed 33 rare diseases involving 7 major systems in patients with scoliosis based on routine clinical diagnostic techniques, molecular biology, and genetic approaches. These diseases included dwarfism, congenital contractural arachnodactyly, mucolipidosis, mucopolysaccharidosis, glycogen storage disease, mitochondrial myopathy, neurodegenerative diseases, Ehlers-Danlos syndrome, Prader-Willi syndrome, Proteus syndrome, Jacro-Levin syndrome, Gilbert syndrome, Freeman-Sheldon syndrome, and spinal muscular atrophy.

All cases were carefully discussed and fully prepared before a multidisciplinary treatment protocol (including the spinal surgery) was performed, which improved the quality of life and laid the foundation for the subsequent systemic treatments. In addition, a Database of Patients with Clinically Rare and Complex Diseases has been successfully established in the Department of Orthopaedics.