Background: As abundance clinical resources, Peking Union Medical College Hospital (PUMCH) has undertook National Science and Technology Specific Projects since 2012, and established a large-scale clinical biobank and database specialized on Rare disease, Malignant tumor, cardiovascular, metabolic diseases and so on. The Biobank has had 8 categories of 89 diseases and Epidemiologic study as well as 50 thousand sets of specimens, that supported 63 items of research projects and more than high-quality 130 articles. The function of PUMCH Biobank has included Support/Service, Scientific Research, Education/Training, Partnership/Sharing.

Methods: As the diagnosis and guidance center which was appointed by Ministry of health, PUMCH focuses on rare diseases from diagnosis, treatment and research. With the low incidence, variety and difficult diagnosis, rare diseases have turned into an important medical problems for China and the world. Focusing on this issue, PUMCH take the led to establish the unified standard registration system for the national rare disease in China(NRDRS) to carry out largescale registration and cohort research. For solving the resource scarcity problems, the project would set up cohort of 59 rare diseases with medical record and biospecimen, probe into study of the accurate phenotype and improve the understanding of the pathogenic mechanism of rare diseases. In December 2016, project held the meeting for China national rare disease registration platform, and then launched International rare disease of China publicity week on February 2017, which with the theme of Research brings infinite possibilities. Subsequently, project set up the rare diseases unit at PUMCH outpatient firstly, which began the patient registration, diagnosis, treatment and prognosis assessment. Biobank undertake the sample and medical record collection, biospecimen process, storage and sequencing. As the sponsor, NRDRS project has held the 12th International Conference on rare diseases and orphan drugs (ICORD) in September 2017. All specialists, researchers and patient organizations of rare diseases joined the meeting and discussed the issues of drug development, research, policy and regulations, etc.

Results and Conclusion: As an important basis of translational medicine research, the value of Biobank for rare diseases is fully embodied. The NRDRS project, which combining Biobank and Rare disease would bring new hope to patients and makes rare diseases rare no longer.

Fig.1 The disease categories and biospecimen size and distribution of PUMCH Biobank.

Fig.2 The function and training of PUMCH Biobank.

Fig.3 The Web set of NRDRS project

Fig.4 The distribution and map of NRDRS project.

- 20 top medical institutions and hospitals in China
- 50,000 cases covering more than 50 diseases
- Multi-omics database and multi-center biological bank