IRUD

Initiative on Rare and Undiagnosed Diseases:

Japanese national UDP

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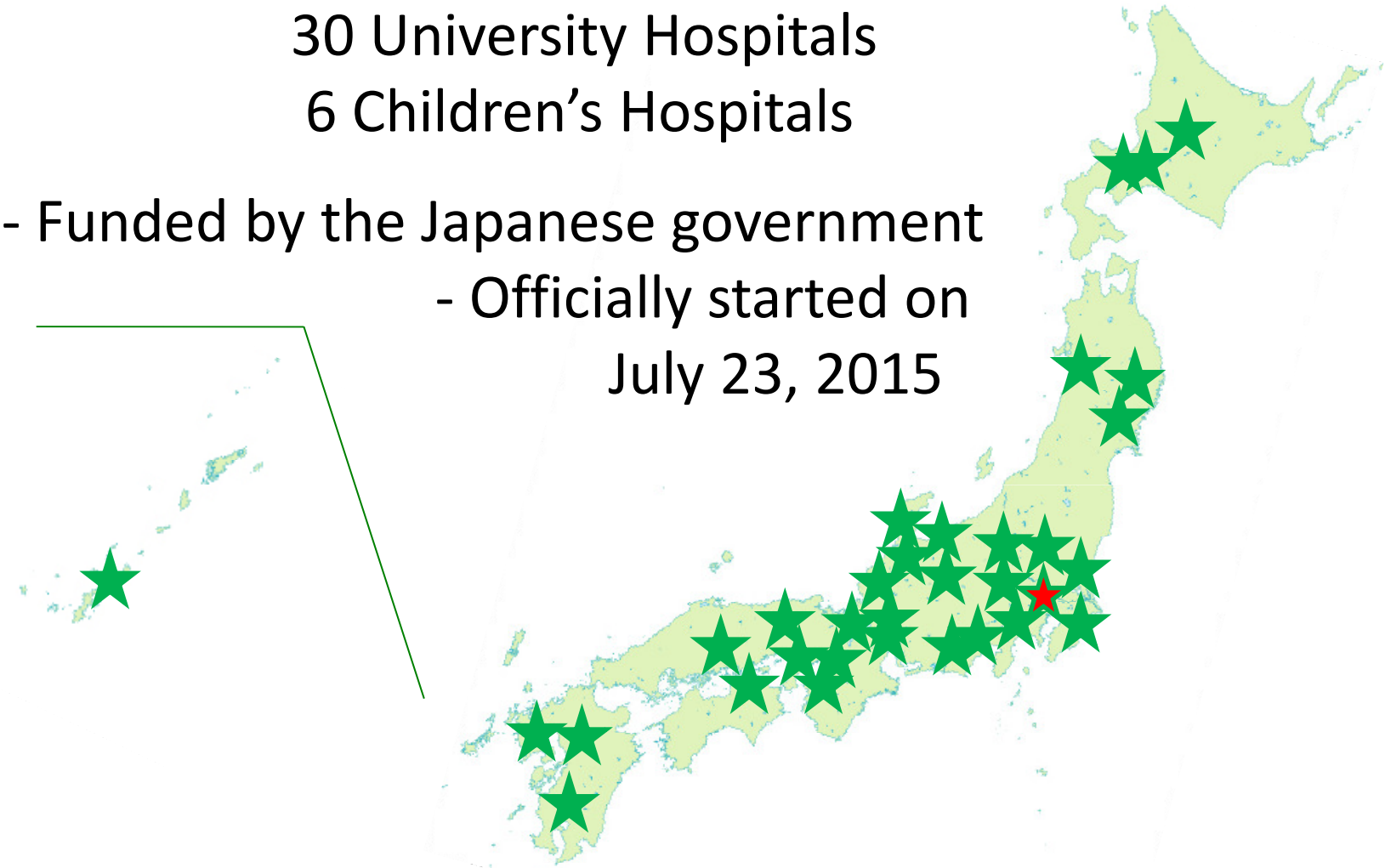
IRUD National UDP network

30 University Hospitals
6 Children's Hospitals

- Funded by the Japanese government
  - Officially started on July 23, 2015



~500 families recruited in 6 months
- Mostly outpatient-clinic basis
  - Exome analysis at 4 centers including Keio Univ

Identification of a new mendelian disease

Two unrelated patients carrying the same *CDC42* mutation

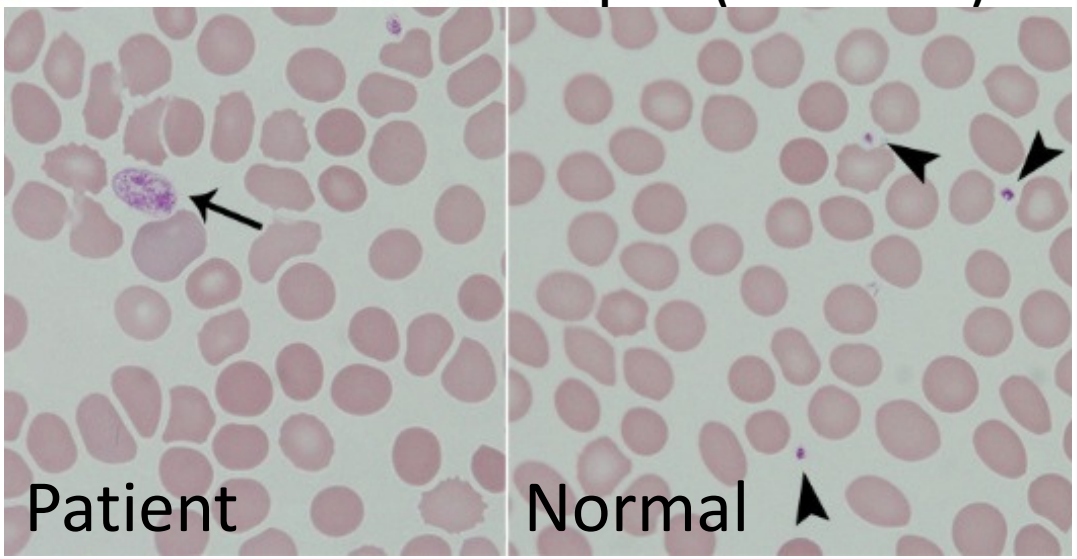
Thrombocytopenia

Macro-thrombocytes

Takenouchi T, Kosaki R, Niizuma T, Hata K, Kosal Am J Med Genet 167: 2822-5. 2015 Nov;

Takenouchi T, Okamoto N, Ida S, Uehara T, Kosaki K. Am J Med Genet, in press (2015 Dec 28)

Recognized and registered as a new mendelian disease (MIM 616737) in OMIM on Jan 13,2016



International collaboration

- 2015/6 Budapest meeting
- 2015/7 Start of national UDP
- 2015/10 UDP symposium in Tokyo with Dr Gahl, Dr Adams and Dr Wise from NIH
- 2015/11 Transpacific UDP workshop in Tokyo with Dr Tift, Dr Baynam and Dr Groza