Screening for specific bone marrow failure/myelodysplastic syndrome genetic disorder in an aggregate identifies a significant subset of patients with inherited bone marrow failure/myelodysplastic syndrome.

- Children and young adults with aplastic anemia or myelodysplastic syndrome
- Patients transplanted in the Fred Hutchinson Cancer Research Center (1990-2012)

**Aplastic anemia**
- 45 young adults
- 53 pediatric

**Myelodysplastic syndrome**
- 64 young adults
- 46 pediatric

Targeted genetic screen analysis for presence of germline mutations in bone marrow failure/myelodysplastic syndrome genes

**Pathologic mutations**
- **Aplastic anemia**
  - 5.1% (5/98)
  - DKC1, MPL, and TP53

- **Myelodysplastic syndrome**
  - 13.6% (15/110)
  - ANCA, GATA2, MPL, RTEL1, RUNX1, SBDS, TERT, TINF2, and TP53
  - 1 patient had a somatically acquired RUNX1 mutation

Family history or physical examination failed to reliably predict the presence of germline mutations.

Keel et al., Haematologica, 2016