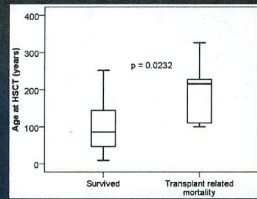
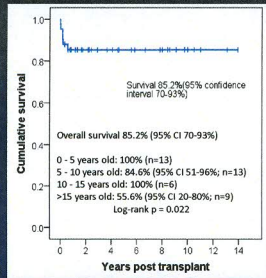


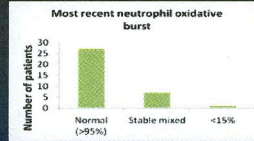
Transplant survival



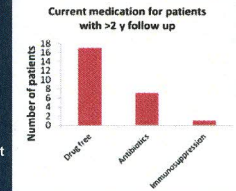
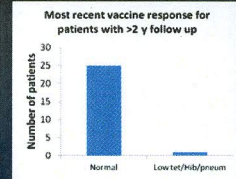
Pre-existing complications and TRM:

- Number of previous fungal infections (p=0.0136)
- Previous brain abscess (p=0.0183)
- Previous bone infection (p=0.0019)

Long term follow-up



- 1 severe chronic gut GvHD unresolved 22 months post HSCT
- 1 severe chronic gut GvHD resolved 5 years post HSCT
- 1 colitis (unknown aetiology) 14 years post HSCT
- 2 chronic lung disease
- 0.017 infections/patient-year beyond 2 years post HSCT (minor skin)
- 1 decreasing neutrophil oxidative burst undergoing further HSCT



Quality of Life Results

	UK norms	No HSCT N=26	Post HSCT N=21	P value: no HSCT vs post HSCT
Parent report	Mean (SD)	Mean (SD)	Mean (SD)	
Total	84.6 (11.2)	65.31 (20.43)	61.93 (18.74)	0.006
Psychosocial	82.2 (12.7)	66.74 (19.25)	61.17 (16.80)	0.010
Physical	89.1 (12.3)	60.35 (25.74)	65.27 (25.27)	0.002
Emotional	78.3 (15.5)	65.53 (20.08)	78.81 (18.57)	0.024
Social	86.8 (15.4)	71.86 (21.48)	88.33 (17.42)	0.007
School	81.5 (16.1)	55.86 (21.92)	79.13 (22.99)	0.001
Child report		N=18	N=17	
Total	83.9 (11.8)	75.39 (12.99)	61.79 (20.22)	0.276
Psychosocial	81.8 (13.2)	74.61 (13.27)	60.28 (19.10)	0.318
Physical	88.5 (11.6)	69.68 (17.69)	69.83 (17.20)	0.002
Emotional	78.5 (17.9)	72.64 (14.37)	65.63 (16.20)	0.016
Social	87.7 (16.5)	73.53 (17.12)	89.71 (19.16)	0.014
School	78.9 (15.9)	64.12 (15.64)	79.12 (18.73)	0.016

Features of IHES

Eczema	100%
Characteristic facies (>16y)	100%
Skin boils	87%
Pneumonias	87%
Lung cysts	77%
Mucocutaneous candidiasis	83%
Scoliosis (>16y)	76%
Delayed dental deciduation	72%
Brain T2 hyperintensities	70%
Pathologic fractures	57%
Chian: E malformation	18%

Antibody deficiency & PCP?

- CD40L deficiency
- ICF
- STAT 1
- Hyper IgE
- NEMO

EDA-ID (Anhydrotic ectodermal dysplasia & immunodeficiency)

NEMO - NF-κB essential modulator (XR)

Anhydrotic ectodermal dysplasia with bronchiectasis, but no ID EDA (XR)

