ALLOGENEIC HEMATOPOIETIC STEM CELL TRANSPLANTATION FOR CHILDREN WITH SEVERE COMBINED IMMUNODEFICIENCY (SCID)

I.Tezcan, T.Turul, D. Uckan, M.Cetin, O.Sanal, F.Ersoy
Hacettepe University
Children's Hospital
Immunology Unit

SCID

- a rare congenital syndrome
- a clinical phenotype in which
- absence of antigen specific T and B lymphocyte immunity
- a variety of underlying genetic abnormalities
- fatal unless hematopoietic stem cell transplantation (HSCT) is performed

Patient Characteristics-1

- 46 patients with SCID enrolled in the study
- 48 transplantations have been performed totally
- during June 1994 - May 2005
- in Hacettepe University Ihsan Dogramaci Children’s Hospital

SCID phenotypes

• T-B+NK-	12	(26%)
• T-B+NK+	13	(28.2%)
• T-B-NK+	17	(37%)
• T-B-NK-	1	(2.2%)
• T+B-NK+	2	(4.4%)
• T+B+NK+	1	(2.2%)

Patient Characteristics-2

SCID phenotype	No. of patients
• B+ SCID	25
• B- SCID	18
• CID (combined immunodeficiency)	3
Total	46

	B+SCID	B-SCID	CID
Median age of diagnosis (months)	6.5 (1-72)	7.5 (1-12)	7 (4-11)
M/F ratio	18/7	12/6	2/1
Total number	25	18	3

No. of patients

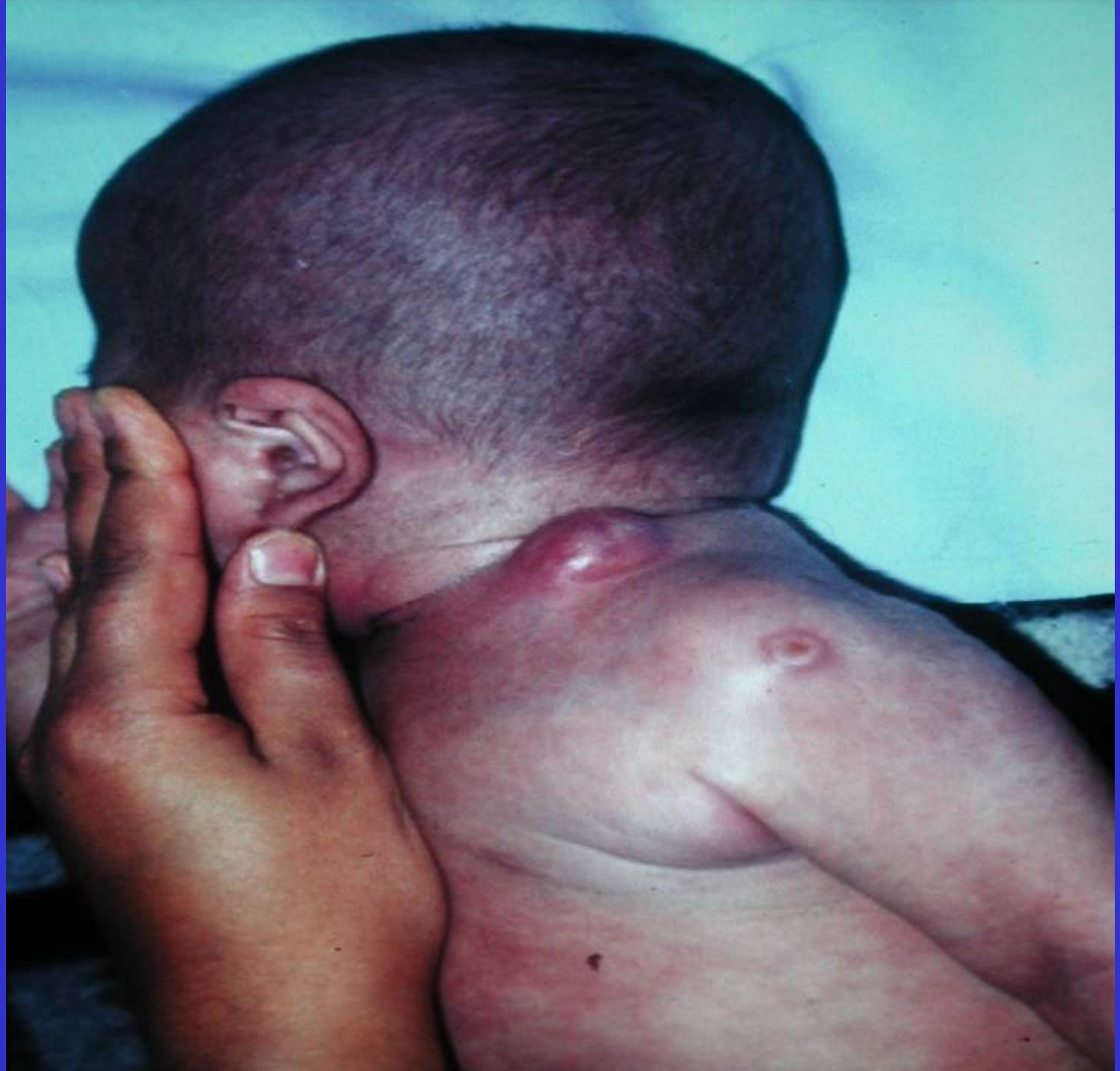
	Identical	Haplo - identical
B +	21	4
B -	10	8
CID	2	1
Total	33	13

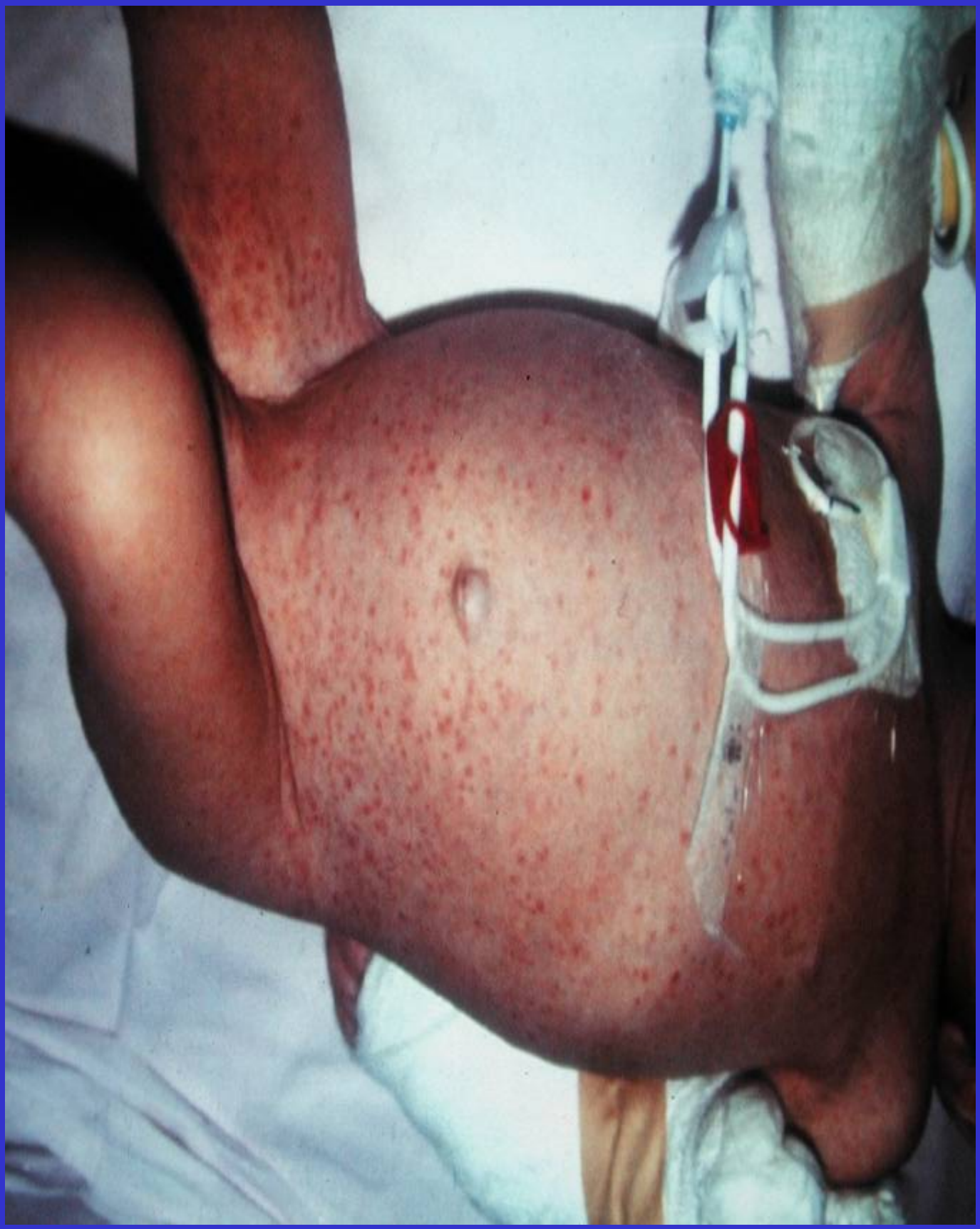
no.of patients

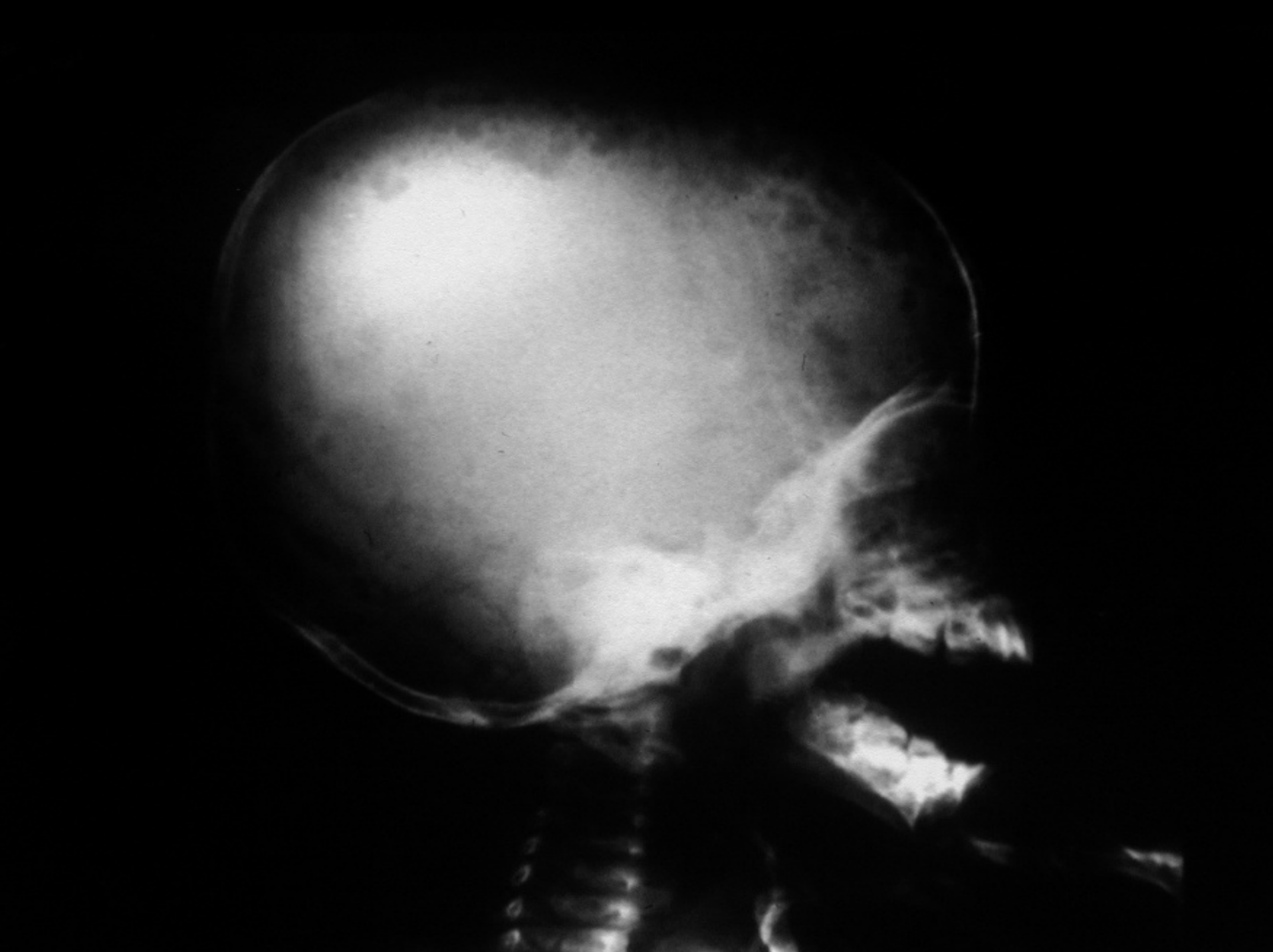
	Identical	Haplo-Identical
BM tx **PSCT(CD34)** **PSCT**	31 1 1	- 13
Median # **Nucl. cell (x10^8 /kg)** **CD34(x10^6/kg)**	7.5	9.2
>1 stem cell Tx	1	1
Median age at tx (months)	7.5 (1.5-90)	8.5 (3-25)
Median follow up(months)	21 (0.5-131)	7 (2-59)

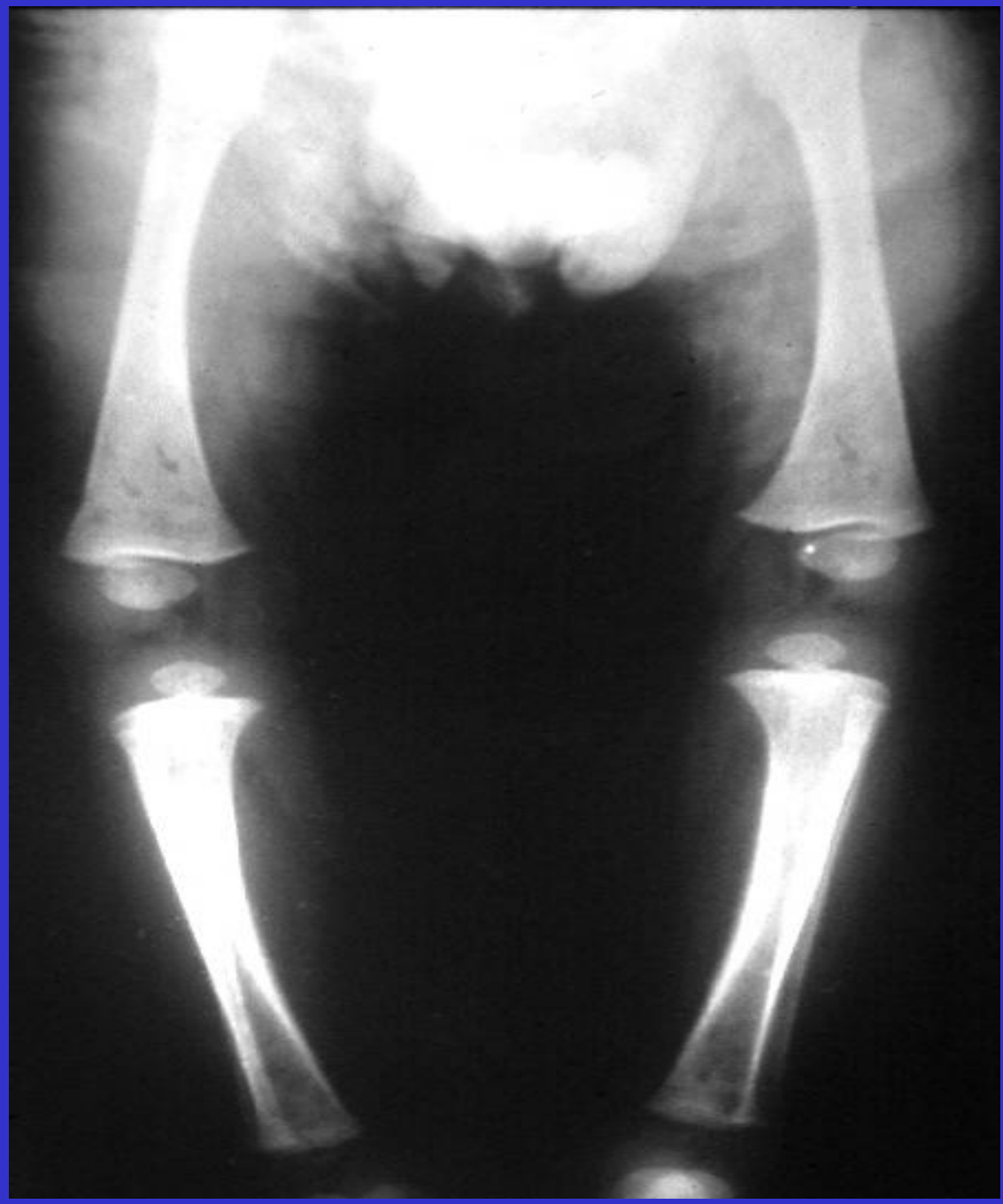
No of patients

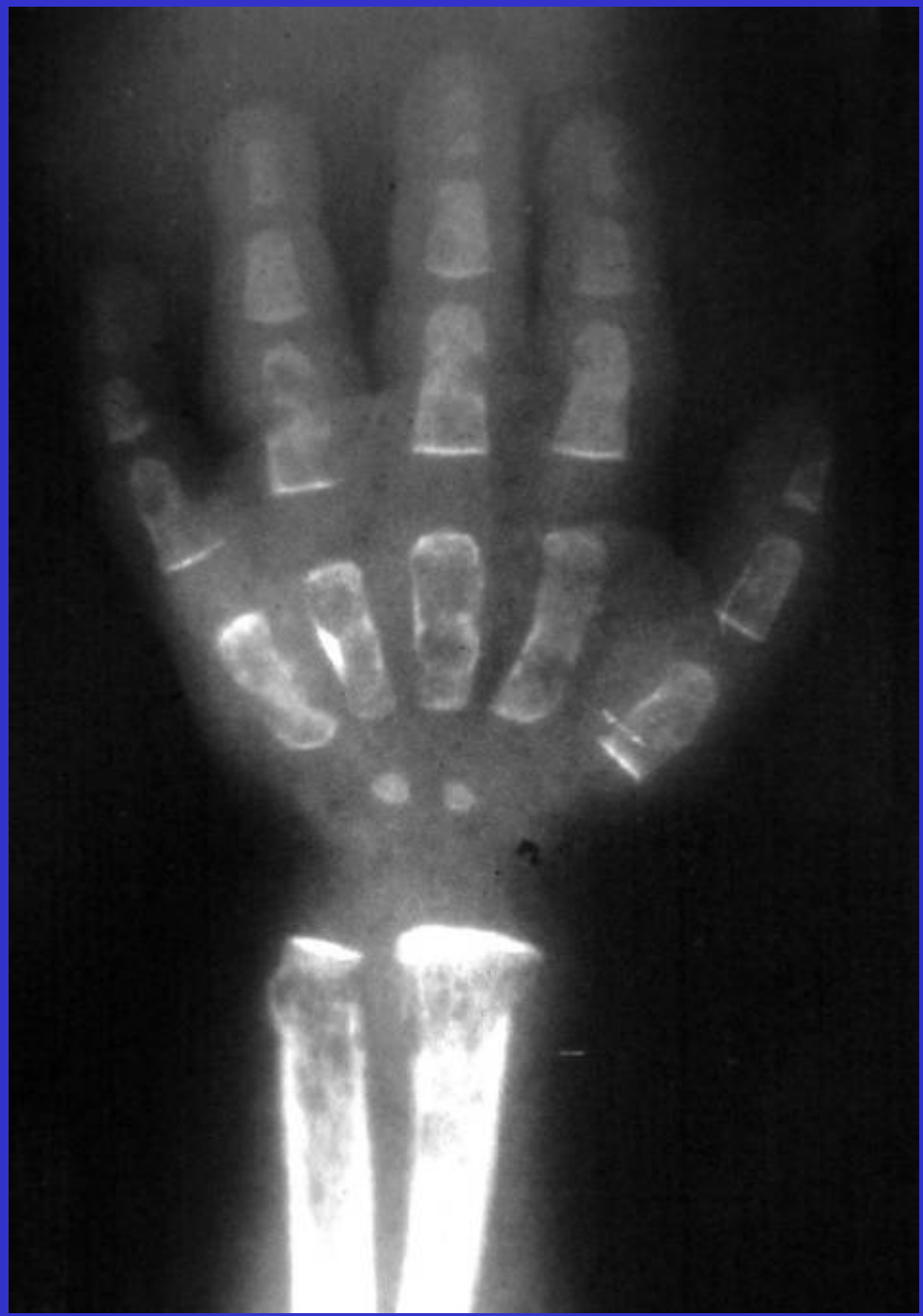
Tx type	GVHD	BO	BCG Inf.	Exitus	Alive
HLA Identic.	6	2	3	11 (33.3)	21 (63.6)
Haplo Identic.	4	1	1	8 (61.5)	5 (38.5)
Total # %	10 (21.7)	3 (6.5)	4 (8.7)	19 (41.3)	27 (58.7)











Long term survival in severe combined immunodeficiency: The role of persistent maternal engraftment

- 8-year-old male (maternal engraftment): recurrent resp. tract inf., oral apht., bifid thumb, cafe au lait spots, skin manifestations 46XX karyotype analysis of blood
- %32 of monocytes are XX, %68 XY
- All fibroblasts are %100 XY
- 6 months old male (classical T-B+NK- SCID): persistant diarrhea
- JAK3 deficiency (kindly performed by Genevieve De Saint Basile, Hopital Necker)

- T-B-NK+ SCID
- HLA identical BMT from mother
- At +5 posttransplant month→ pancytopenia
- HSV PCR (+)
- Significant improvement – Acyclovir treatment

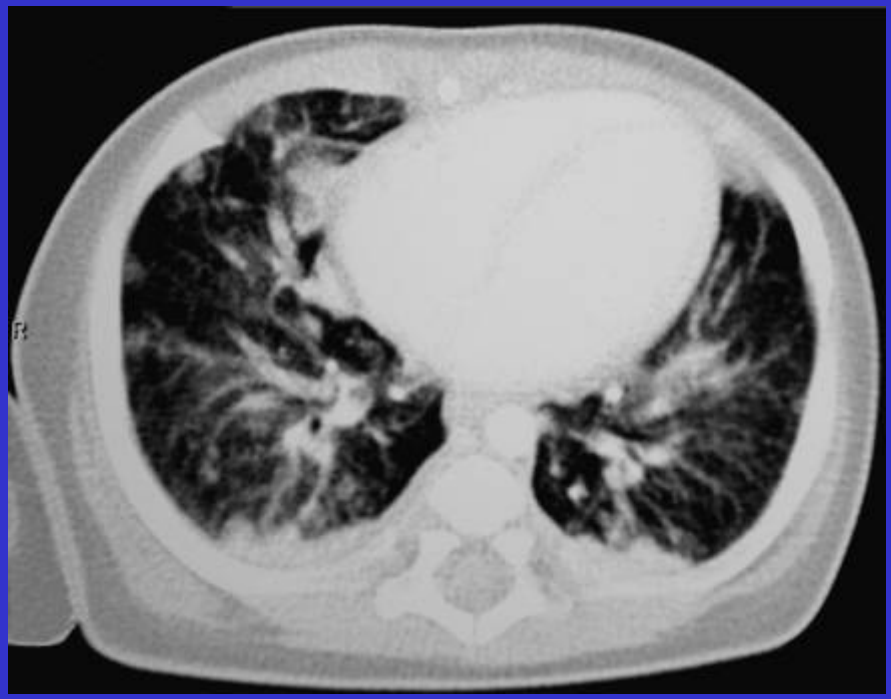
Immune reconstitution

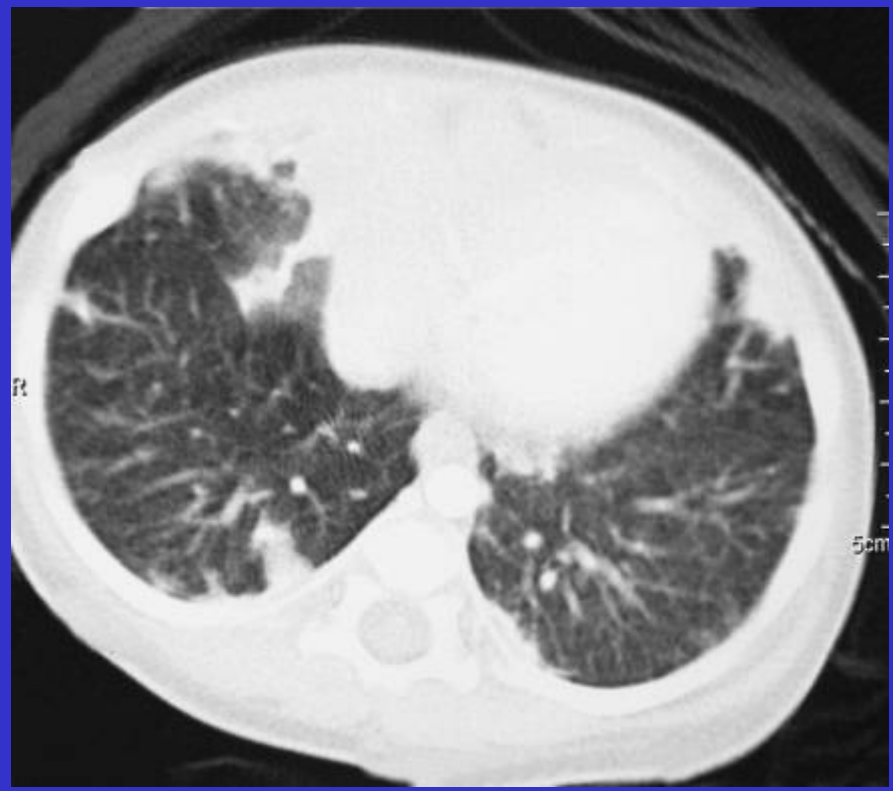
at posttx. 6 months

	B+	B-	CID
HLA Iden.	(n=14)	(n=6)	(n=2)
B cell fx.	9/14	4/6	1/2
T cell fx.	14/14	6/6	1/2
Haploiden.	(n=4)	(n=3)	-
B cell fx.	0/4	0/3	
T cell fx.	3/4	1/3	

Death analysis of HLA identical Tx

	patients	Alive (%)	Death (%)
Age at tx			
<6 months	12	10 (83.3)	2 (16.6)
>6 months	21	12 (57.2)	9 (42.8)
SCID pheno.			
B+	21	13 (61.9)	8 (38.1)
B-	10	7 (70)	3 (30)
CID	2	2 (100)	0 (0)
NK+	25	16 (64)	9 (36)
NK-	8	6 (75)	2 (25)
Pulmonary inf before tx.			
Yes	23	14 (35.7)	9 (64.3)
No	10	9 (90)	1 (10)
>3 months posttx			2 (18.1)
<3 months posttx			9 (81.9)

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5cm

In Conclusion;

- Prognosis in HLA identical tx was found to be assoc. with

✓ Age of tx (>6 months)

✓ Presence of pulmonary inf. before tx