PID patients in Motol

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PID patients in Motol

. Antibody deficiencies

. T cell or combined deficiencies

. Phagocytic disorders

. Complement deficiencies

. Other primary immunodeficiencies

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Antibody deficiencies

- XLA Bruton´s agammaglobulinemia
- IgA deficiency
- CVID (common variable immunodeficiency)
- selective IgG subclass deficiency

Antibody deficiencies

	Nr.	age
X-LA	7	1-31
IgA defic.	30	
CVID	23	5-63
selective subclass deficiency	5	11-13
Total	65	

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Combined deficiencies

Cellular and combined	Nr.	comment	
SCID	**10**	γc mutation	**5**
		RAG mutation	**2**
CID	**2**		
FHL	**4**	**Perforin deficinency**	**1**
WAS	**3**	WASP mutation	**2**
complete di George sy	**1**		
di George syndrome parc.	**37**		

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Phagocytic disorders

	Nr.	age
LAD I.	1	17
CGD	6	19-32
Total	7	

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Complement deficiencies

C1 esterase deficiency	5
C4 deficiency	4
C2 deficiency	5
MBL deficiency	1
Total	15

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Day stacionary IVIG therapy 2005			
	Nr.		age
CVID	23		5-63
X-LA	4		1-31
subclass deficiency	2		11-13
XLP	1		17
combined deficiency	1		23
Total	31		1 - 63

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Unexpected social challenge in XLA

case report

Patients - clinical picture

- Siblings - brothers 24 and 31 years
- repeated infections appeared around 2 years of age
- later frequent severe bronchopneumonias, sinusitis, otitis

- progressive postlingual sensorineural deafness
- psychomotorical retardation
- speaking difficulties →speech disorder
- behaviour disorders – agresivity, sexual harasment

Unexpected social challenge in XLA

case report

Laboratory investigation

undetectable levels of immuno-globulins and absence of B cells

BTK – mutation in btk gen (Xq21.3-22)

DDP – mutation in DDP gen

Diagnosis: XLA and Mohr Tranebjaerg sy

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Unexpected social challenge in XLA

case report

Family:

father died 1996 (cancer)

they lived together with mother - she died 2004 (stroke)

social status:

younger brother - irresponsible, lived with curator (mother)

curator now – Prague dictrict

Difficulties – communication (deafness and mutitas)

treatment observance

housekeeping

financial – the only income -disability allowance

Help for patients with PID

Patients organisation

We initiate creation of czech self help PID patients organisation

But:

Parents and families with PID patients are still low active

More information for interested persons

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Conclusions I.

Antibody deficiencies	(XLA, IgAdef., CVID,subclass defic.)	42	35,5 %
Increased chromosomal fragility	(AT)	2	2 %
Combined deficiencies	(SCID, perforin defic., XLP,WAS,diGeorge)	52	44 %
Phagocytic disorders	(LAD I., CGD)	4	3 %
Complement deficiencies		15	13 %
Defect in apopthosis	(ALPS)	3	2,5 %
Total		118	100 %

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Conclusions II.

- need for patients organisation
- need for cooperation with local social services and governemental structures

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