

Case Report Form for Genetic and Immunological Analysis of Inflammatory Bowel Diseases in Children

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1. Contact information of attending physician

Physician's name:

Department:

Institution:

Postal address:

Phone:

FAX:

E-mail:

Date of last assessment:

d

d

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y

y

y

y

2. Patient's demographics

Patient's name:

Sex:

☐ female

☐ male

Date of birth:

d

d

/

m

m

/

y

y

y

y

☐ alive

☐ dead, date of death:

d

d

/

m

m

/

y

y

y

y

cause of death:

Gestational age:

Pregnancy complications:

Ethnicity:

☐ Caucasian

☐ First Nations

☐ Black (e. g. African, Haitian, Jamaican)

☐ Asian (e.g. Chinese, Japanese, Vietnamese, Cambodian, Filipino, Korean, Laotian)

☐ South Asian (e.g. East Indian, Pakistani, Sri Lankan, Punjabi, Bangladeshi)

☐ Arab/West Asian (e.g. Armenian, Egyptian, Iranian, Moroccan, Lebanese, Afghani)

☐ Latin American (e.g. Mexican, Cuban, Puerto Rican, Central/South American)

☐ Turkey, please specify:

☐ Other:

Country of birth:

Country of origin/ethnicity, mother:

Country of origin/ethnicity, father:

3. Clinical data

Date of first symptoms of IBD:

d d / m m / y y y y

Date of diagnosis:

d d / m m / y y y y

3.1. Gastrointestinal symptoms

(Please indicate: No; Yes; Unk, unknown)

[illegible]

3.2. Disease behavior (Levin et al., Pediatric modification of the Montreal classification for inflammatory bowel disease: the Paris classification. *Inflamm Bowel Dis.* 2011)

- **B1 - Nonstricturing, nonpenetrating disease:** uncomplicated inflammatory disease without evidence of stricturing or penetrating disease.
- **B2 - Stricturing disease:** the occurrence of constant luminal narrowing demonstrated by radiologic, endoscopic, or surgical examination combined with prestenotic dilation and/or obstructive signs or symptoms but without evidence of penetrating disease.
- **B3 - Penetrating disease:** the occurrence of bowel perforation, intraabdominal fistulas, inflammatory masses and/or abscesses at any time in the course of the disease, and not secondary postoperative intra-abdominal complication (excludes isolated perianal/rectovaginal fistulae).
- **B2B3 – Stricturing and penetrating disease:** the presence of both B2 and B3 phenotypes in the same patient, either at the same moment in time, or separately over a period of time.

3.3. Extra-Intestinal manifestations

[illegible]

3.4. Symptoms of Primary Immunodeficiency (PID)

	No	Yes	Unk	If yes, please specify:
Four or more new ear infections within 1 year:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Two or more serious sinus infections within 1 year:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Two or more pneumonias within 1 year:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Recurrent, deep skin or organ abscesses:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Persistent thrush in mouth or fungal infection on skin:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Two or more deep-seated infections including septicemia:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Severe viral infections requiring hospitalization:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Recurrent infections with atypical mycobacteria:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Other:				
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	

3.5. Autoimmune diseases

	No	Yes	Unk	If yes, please specify:
Autoimmune thyroiditis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Diabetes mellitus type I:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Hemolytic anemia:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Thrombocytopenia:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Autoimmunneutropenia:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Vasculitis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Glomerulonephritis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Arthritis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Celiac disease:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Primary sclerosing cholangitis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Autoimmune hepatitis/Overlap syndrome:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Other:				
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	

3.6. Allergic diseases

	No	Yes	Unk	If yes, please specify:
Food allergy:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Asthma:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Atopic dermatitis:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Other:				
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	

3.7. Anthropometry

Date (dd/mm/yyyy):	Height (cm):	Weight (kg):
Birth:		
Diagnosis of IBD:		
Date of last assessment:		

4. Diagnostics

4.1. Endoscopy/Histology (Gastrointestinal involvement/Disease location)

	First						Last							
Date of upper GI endoscopy:	m	m	/	y	y	y	y	m	m	/	y	y	y	y
Date of lower GI endoscopy:	m	m	/	y	y	y	y	m	m	/	y	y	y	y

Please indicate: nor, normal; abn, abnormal (consistent with IBD); nv, not visualized

	<u>Endoscopy</u>			<u>Histopathology</u>			
	nor	abn	nv	nor	abn	nv	If yes, please specify:
Mouth:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Esophagus:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Stomach:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Duodenum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Jejunum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Proximal ileum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Distal ileum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Terminal ileum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Cecum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Ascending Colon:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Transverse Colon:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Descending Colon:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Sigmoid:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Rectum:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
Perianal/perineal:	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	
				No	Yes	Unk	
Histological detection of granuloma?				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	Location?

4.2. Imaging (e. g. MRI, Video capsule endoscopy, Barium studies)

[illegible]

4.3. Blood cell counts

☐ Not done

☐ Done, date:

m

m

/

y

y

y

y

 (Initial)

m

m

/

y

y

y

y

 (Last)

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

CBC:

Initial

Last

Hb (g/dl):

MCV (fl):

WBC (cells/ μ l):

Plt (cells/ μ l):

Diff (%):

Initial

Last

Neutrophils:

Monocytes:

Lymphocytes:

Eosinophiles:

Basophiles:

4.4. Immunological work-up and genetic testing

4.4.1. Immunophenotyping (T cell subsets, B cell subsets, NK cells)

☐ Not done

☐ Done, date:

m

m

/

y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

4.4.2. Serum immunoglobulin levels

☐ Not done

☐ Done, date:

m

m

/

y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

IgA (Unit):

IgD (Unit):

IgE (Unit):

IgG (Unit):

IgM (Unit):

Below

Normal range

Above

☐

☐

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4.4.3. Antibody titers in response to vaccination

☐ Not done

☐ Done, date:

m

m

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y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

4.4.4. Functional immunological assays (T cell proliferation, NK cell assays)

☐ Not done

☐ Done, date:

m

m

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y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

4.4.5. Neutrophil NADPH-oxidase activity

☐ Not done

☐ Done, date:

m

m

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y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

4.4.6. Other immunological findings?

☐ Not done

☐ Done, date:

m

m

/

y

y

y

y

(under immunosuppressive therapy? ☐ No ☐ Yes, ☐ Unk)

Results:

4.5. Genetic diagnostics

☐ Not done

☐ Done, date:

m

m

/

y

y

y

y

☐ Unk

if done, please specify type of diagnostics:

Principal investigator:

Name:

Institution:

Country:

Results:

5. Treatment

5.1. Nutrition

Breastfeeding:

☐ No

☐ Yes

☐ Unk

Start of infant formula (months):

m

m

Elimination diet:

☐ No

☐ Yes

☐ Unk

If yes, please specify:

Formula:

Duration (start – end):

Good

Partial

None

ø tolerated

☐

☐

☐

☐

Exclusive enteral nutrition:

☐ No

☐ Yes

☐ Unk

If yes, please specify:

Formula:

Duration (start – end):

Good

Partial

None

ø tolerated

Total:

☐

☐

☐

☐

Partial:

☐

☐

☐

☐

Parenteral nutrition:

☐ No

☐ Yes

☐ Unk

If yes, please specify:

Duration (start – end):

Good

Partial

None

ø tolerated

Total:

☐

☐

☐

☐

Partial:

☐

☐

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5.2. Antibiotics

Medication:

Response to treatment:

Good

Partial

None

ø tolerated

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5.3. Anti-inflammatory and immunosuppressive drugs

Medication (dosage, mg/kg/d):

No

Yes

Unk

Duration (start – end):

Response to treatment:

Good

Partial

None

ø tolerated

Mesalazine (5-ASA):

☐

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Sulfasalazine:

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Steroids:

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Azathioprine:

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6-Mercaptopurine:

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Methotrexate:

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Infliximab:

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Other anti-TNF- antibodies:

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5.4. Surgical treatments

Treatment:

NoYesUnk

Percutaneous endoscopic gastrostomy:

m

m

/

y

y

y

y

Ileostomy:

m

m

/

y

y

y

y

Colostomy:

m

m

/

y

y

y

y

Bowel resection:

m

m

/

y

y

y

y

Strictureplasty:

m

m

/

y

y

y

y

Fistula/Abscess (Fistulotomy, Sedon, ...):

m

m

/

y

y

y

y

Other:

m

m

/

y

y

y

y

m

m

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y

y

y

y

m

m

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y

y

y

y

5.5. Hematopoietic stem cell transplantation (HSCT)

Not done

Done, date:

If HSCT was performed, please indicate:

Conditioning regimen:

Medication:

Dosage (mg/kg/d):

Duration (start – end):

GVHD prophylaxis:

Medication:

Dosage (mg/kg/d):

Duration (start – end):

Donor:

HLA-matching:

HSCT source (cells/kg):

Engraftment (d after HSCT):

Chimerism (Excel data preferred):

Complications:

- GVHD:

- Other:

Clinical outcome:

WBC (>1000/μl):

Granulocytes (>500/μl):

Plt (>50.000/μl):

6.1. Family history

If any family history of IBD, please indicate:

Other?

(In case of known consanguinity and a complex kindred, please provide a schematic pedigree)

[illegible]

Spontaneous abortions? ☐ No ☐ Yes, Please indicate number: ☐ Unk