

Number of patients with DNA samples available in AGORA (updated in March 2018)

Table 1. Number of DNA samples available

	5 390 children	4 015 children with birth defects	932 children with cancer	728 control children
Both parents	3 621	3 076	538	3
Only mother	1 161	310	128	722
Only father	103	68	34	
No parents	505	561	232	3
	13 896 samples	10 545 samples	2 170 samples	1 456 samples

Table 2. Diagnoses of the cancer patients

Category and phenotype	Number	Category and phenotype	Number
<i>Tumors^a</i>	932	Melanoma	2
ALL	272	Melanotic-neuroectodermal tumor of infancy	1
AML	17	Meningioma	8
Astrocytoma	50	Nasopharyngeal carcinoma	4
Atypical teratoidrhabdoid tumor	4	Neuroblastoma	43
Basal cell carcinoma	4	Neurofibromatosis	3
B-cell lymphoma	1	NHL-ACL	7
Chorioid plexus tumor	2	NHL B cell	52
Clear cell sarcoma kidney	1	NHL precursor B cell	1
CNS tumor	14	NHL T cell	22
Cong. mesoblastic nephroma	2	NHL other	4
Cortical adrenal carcinoma	2	Kidney tumor	1
Cranyopharyngoma	16	Non-rhabdo soft tissue sarcoma	5
Dysembryoplastic neuro-epithelial tumor	1	Oligodendroglioma	2
Ependymoma	17	Other tumors	2
Ewing sarcoma	30	Osteosarcoma	22
Extragenadal germ cell tumor	1	Ovarian germ cell tumor	7
Fibromatosis	2	Ovarian tumor benign	1
Ganglioglioma	2	Pilocytar asrocytoma	3
Ganglioneuroma	3	Plexus papilloma	1
Germinoma	5	PNET	3
Glioma optical	11	PNET atypical Ewing sarcoma	1
Hamartoma		Retinoblastoma	4
Hepatoblastoma	10	Rhabdoid tumor kidney	1
Brain tumor	2	Rhabdomyosarcoma	34
HLH	1	Squamous cell carcinoma	1
Hodgkin	69	Teratoma	9
Germ cell tumor	17	Testicular germ cell tumor	2
LCH	17	Wilms tumor	91
Malignant histiocytosis	1	Other [#]	9
Medulloblastoma	23	Unknown	14

^aTotal number of patients with 1 or more anomalies in a category; [#]Including 2 perineal lipomas and 7 other tumors

Table 3. Diagnoses of the patients with birth defects

Category and phenotype	Number	Category and phenotype	Number
<i>Digestive tract^a</i>	778	Ectopic kidney	13
Anorectal malformations	619	Polycystic kidney	3
Oesophageal atresia	81	Afunctional kidney	22
Tracheoesofagal fistel	18	Pelvic kidney	9
Duodenum/ileum/colon atresia	43	Horseshoe kidney	23
Cloacal extrophy	2	Ectopic ureter	15
Malrotation/position intestine	38		
Hirschsprung/dysganglionosis	27	<i>Other kidney problems^a</i>	81
Bile duct atresia	2	Renal insufficiency	13
Other [#]	28	Kidney stones	15
Unknown	14	Pyelonefritis	8
		Other [%]	46
<i>Abdominal tract</i>	94		
Diaphragmatic hernia	38	<i>Cardiovascular anomalies^a</i>	240
Omphalocele	23	Ventricular septal defect	87
Gastroschisis	16	Atrial septal defect	65
Other ^{&}	13	Aortic coarctation	12
Unknown	4	Open ductus botallicus	44
		Teratology Fallot	9
<i>Urogenital tract^a</i>	2,236	Mitralis valve insufficiency	3
Hypospadias	1,061	Truncus arteriosus	1
Meatus stenosis	136	Valvular pulmonal stenosis	4
Phimosis	95	Perifere pulmonal stenosis	16
Cryptorchidism	312	Transposition big vessels	2
Vesico-ureteral reflux (VUR)	491	Hypoplastic left heart syndrome	1
Mega-ureter	76	Patent ductus arteriosus (PDA)	11
Ectopic ureter	12	PAPVC	1
Neurogenic bladder	44	Double inlet left ventricle	1
Bladder extrophy	19	Bicuspide aorta valve (BAV)	8
Posterior urethral valves (PUV)	480	Aorta stenosis	5
Penoscrotal transposition	7	Right/left isomersism	1
Scrotum bifidum	29	Other ^{\$}	25
Ambiguous genitalia	3	Unknown	7
Uterus malformations	15		
Hydrocolpos	3	<i>Central nervous system anomalies^a</i>	81
Utriculus cyste	19	Spina bifida	21
Hernia umbilicalis	7	Encephalocele	2
Hernia inguinalis	87	Hydrocephalus	14
Ureterocele	65	Microcephalia	6
Hydrocele	74	Intraspinal lipoma	1
Varicocele	7	Tethered cord	26
Epispadias	20	Sirenomelie	3
Bladder divertikel	17	Caudal regression sequence	4
Buried pens	15	Other ⁺	25
<i>Other urogenital problems^a</i>	251	<i>Orofacial clefts^a</i>	361
Urolithiasis	16	Cheiloschisis	59
Incontinence	92	Cheilognatoschisis	50
Recidiving urinary tract infections	64	Cheilognatopalatoschisis	141
Dysfunctional voiding	24	Palatoschisis	109
Enuresis nocturna	29	Pierre Robin sequence	11
Hematurie	12	Lipomyeloschisis	2
Epididymitis	3	Bohring Opitz syndrome	1
Other [*]	78		

<i>Kidney anomalies^a</i>	705	<i>Skeletal anomalies^a</i>	265
Unilateral renal agenesis	64	Vertebral anomalies	145
Renal hypodysplasia	85	Extremity anomalies	77
Ureter-pelvic-junction obstruction	320	Rib anomalies	46
Hydronephrosis	245	Hip dysplasia	44
Duplex collection system	159	Sacral agenesis	22
Multicystic dysplastic kidney	63	Scoliosis	17
		Cranyosynostosis	3
		Other [@]	10

^a The total number of patients with one or more anomalies in a category; [#] Including 10 gastro-oesophageal reflux, 2 pylorus hypertrophy, and 16 other anomalies; [&] Including 8 pulmonary anomalies (pneumothorax, tracheomalacy, obstruction / malformation / dysplasia bronchus), and 5 other anomalies; ^{*} Including 3 adrenogenital syndrome, 8 torsio testis, 3 testis cyste, 7 other testis problems (vanishing, atrophy, retractile, retention, ascensus), 6 dilated urinary tract, 3 double vagina, 2 enuresis, 6 urethra stenosis, 4 trabecular bladder, and 36 other anomalies; [%] Including 25 dilatation, 6 hypoplastic or small kidneys, 3 cystic kidneys, 2 nephrotic syndrome, 2 reflux nephropathy, and 8 other anomalies; ^{\$} Including 12 open foramen ovale, and 13 other anomalies; [@] Including 2 M. Perthes, and 8 other anomalies; ⁺ Including 3 encephalopathy, 3 spina bifida occulta, and 19 other anomalies.