The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

Please complete this questionnaire as best you can.

Most of this information is reported in your gestational medical records, if you don't remember the answer to a question, would you be willing to release prenatal records from the pregnancy to participate in this study?

If Yes, please fill out a medical release form for the OB medical records.
If No, just answer "unknown".

Your answers will help us to better understand the prenatal profile of Pallister-Killian Syndrome (PKS) and to define an approach to prenatal diagnosis of PKS for families with or without a prior PKS pregnancy. Thank you!

Today's Date: _____

Child's Name: _____

Date of Birth: _____

Sex: Male ____ Female ___

Parent/Guardian Names: Mother:_____Father: _____

Address: _____

Phone: _____

Email: _____

The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

PART A
PREGNANCY HISTORY

Mother Age at birth:_____

Month gestation when you first felt fetal movement: _____

How would you define fetal movement? Increased _____ Normal _____Decreased _____

During the pregnancy

- **Did you have vaginal bleeding?** YES_____NO_____
- **Did you have any severe nausea or vomiting that required a visit to the Emergency Department?** YES_____NO_____
- **Did you have any infectious diseases or infections? (including urinary tract infections)** YES_____NO_____
  If Yes, LIST
  _____
  _____

- **Did you have diabetes (including gestational diabetes) ?** YES _____ NO_____
- **Did you have hypertension (with or without proteinuria and/or seizures)?** YES _____ NO_____
- **Did you have any other complication?** YES _____ NO_____

  If Yes, LIST
  _____
  _____

- **Did you smoke cigarettes?** NO ___ YES ___ If so, How many per day? _____
- **Did you have any alcoholic beverages?** NO ___ YES ___
- **Did you take any medications or drugs (including vitamins)?** YES __ NO __

  If Yes, LIST:
  _____
  _____

The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

- **Were you exposed to radiation (X-rays)?** YES_____NO_____. If Yes, at what gestational age? _____

- **Did conception occur spontaneously?** YES_____ NO_____

If no, which reproductive technology or assistance (IUI, IVF, ICSI, PGD, donor) did you use?
_____

- **Did you have an ultrasound(s) completed?** YES ____ NO ____

If so, how many (approximately) _____ and at what gestational ages? _____

Were any of the following findings detected by prenatal ultrasound?

☐ Increased fluid (polyhydramnios)
☐ Decreased fluid (oligohydramnios)
☐ Microcephaly (small head circumference)
☐ Cleft palate
☐ Micrognathia (small chin)
☐ Diaphragmatic hernia
☐ Intrauterine growth retardation (IUGR – or small size)
☐ Large size
☐ Extra digits
☐ Accessory (extra) Nipples
☐ Shortening of the long bones
☐ Ventriculomegaly (enlarged spaces in the brain)
☐ Brain difference (please list) _____
☐ Kidney difference (please list) _____
☐ Cardiac difference (please list) _____
☐ Gastrointestinal difference(please list) _____
☐ Other differences (please list):
_____
_____
_____

Was it a singleton pregnancy or a twin/multiple pregnancy? _____

Was amniotic fluid volume normal?

The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

YES ____ NO ____ If no, was it increased ____ or decreased____ ?

Did you have the first trimester screen blood test (usually done around 10 weeks gestation) ?

YES ____ NO ____ If so, was it normal? YES ____ NO ____ If No, please fill in the following questions as best you can:

- Pregnancy-associated plasma protein-A (PAPP-A) serum level Increased____ Decreased____
- free Chorionic gonadotropin (bhCG) serum level Increased____ Decreased____
- Nuchal Translucency scan measurement Increased____ Decreased____
- Absence or Hypoplasia of fetal nasal bone YES ____ NO ____

Did you have the multiple marker screen blood test (usually done around 16 weeks gestation and known as triple or quadruple screen)?

YES ____ NO ____ If so, was it normal? YES ____ NO ____ If NO please fill in the following questions:

- Alpha-fetoprotein (AFP) serum level Increased____ Decreased____
- Estriol serum level Increased____ Decreased____
- Chorionic gonadotropin (b-hCG) serum level Increased____ Decreased____
- Inhibin-A serum level Increased____ Decreased____
- Nuchal fold measurement Increased____ Decreased____

Did you have NonInvasive Prenatal Testing - NIPT - (usually done by a simple blood test that analyzes DNA called cell-free DNA to determine baby's risk for genetic chromosomal abnormalities)?

YES ____ NO ____ .

If so, what was the result? : _____

Did you have amniocentesis (a diagnostic test for chromosomal abnormalities usually done between 15-18 weeks gestation through the sampling of a small amount of amniotic fluid by a thin needle through your belly)?

YES ____ NO ____ .

If so, what was the result? : _____

The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

Did you have Chorionic Villus Sampling – CVS (a diagnostic test for chromosomal abnormalities usually done between 10-12 weeks gestation, in which a sample of chorionic villi is removed from placenta through cervix or abdominal wall)?

YES ____ NO ____ .

If so, what was the result? : _____

PART B
BIRTH HISTORY:

- **Did you have a: vaginal delivery _____ or C-section _____? If c-section, what was the reason? _____**
- **Birth weight:**
- **Birth Length:**
- **Gestational age at delivery:**
- **Head circumference:**
- **Apgar Scores: _____**
- **After birth, did your child go to the NICU? Yes ____ No _____If yes, why?**
  _____
- **Duration in the hospital after birth: _____**
- **Other issues of note relating to the birth history:**
  _____
  _____
  _____
  _____
- **At the time of the child's birth, please describe any problems that occurred, and what test results (such as laboratory, radiology and other imaging studies) showed:**
  _____
  _____
  _____
- **Please describe complications, if any, of neonatal period:**
  _____
  _____
  _____
- **When/how was the diagnosis of PKS made for child?**

The Children's Hospital of Philadelphia
1002 Abramson Research Center, 34th Street and Civic Center Boulevard Philadelphia, PA 19104
Tel: 267-426-5388 Fax: 215-590-3850

Pallister-Killian Syndrome (PKS) – Prenatal Findings Study

- **Please, fill the Table 1 below with any issue detected in your child**

Table1

Postnatal Karyotype	From skin sample:			
	From blood sample:			
Congenital Diaphragmatic Hernia		YES	NO	
Heart Differences	Atrial Septal defects	Ventricular septal defects	other (explain)	NO
Respiratory Concerns	(explain)			
Gastrointestinal Differences	Intestinal Malrotation	YES	NO	
	Pyloric stenosis	YES	NO	
	Esophageal atresia	YES	NO	
	Feeding problem	YES	NO	
	Gatroesophageal reflux	YES	NO	
	Other (explain):			
Genito-urinary Differences	Dysplastic kidney(s):	YES	NO	
	Small kidney(s):	YES	NO	
	Hypospadias (males only):	YES	NO	
	Undescended testes (males only):	YES	NO	
	Other (explain):			
Brain Differences	Ventriculomegaly	YES	NO	
	Other (explain):			
Cleft Palate		YES	NO	
Skeletal/Limbs Differences	Polydactyly (extra-digits):			
	Join Contractures:	YES	NO	
	Scoliosis:	YES	NO	
	Other (explain):			
Umbilical Hernia		YES	NO	
Skin Findings	Eczema	YES	NO	
	Hemangiomas	YES	NO	
	Skin discoloration	YES	NO	
	Birthmarks	YES	NO	
	Hypopigmentation	YES	NO	
	Other (explain)			
Otolaryngology Concerns	Earing loss	YES	NO	
	Recurrent ear infections	YES	NO	
	Other (explain):			
Ophthalmologic Concerns	(explain)			
Dental Concerns	(explain)			
Growth delay	YES	weight:	height:	head circumference:
	NO			
Growth hormone blood level	(Report if available)			
Low blood sugar		YES	NO	
Muscle tone difference (low or increased)	(explain)			
Seizures		YES	age of onset	NO
Behavioural concern (autism, ADHD, other)	(explain)			
Development	Rolled	YES	at what age	NO
	Sat	YES	at what age	NO
	Crawled	YES	at what age	NO
	Walked	YES	at what age	NO
	Talked	YES	at what age	NO
	Toiled trained	YES	at what age	NO
	Dressed self	YES	at what age	NO
OTHER				

Name:

MR#

Age/Date of Birth:

Family History

	Name	Age	Highest grade completed	Occupation
Child's Biological Father				
Child's Biological Mother				

Please list all of mother's biological children and any miscarriages

Year	Outcome	Name	Sex	Present Age	Any developmental concerns
	Living Miscarriage				Yes No If yes, what?
	Living Miscarriage				Yes No If yes, what?
	Living Miscarriage				Yes No If yes, what?
	Living Miscarriage				Yes No If yes, what?

Is there anyone in the family with any of the following? (tell us who in relation to the child has these conditions)

Condition	**Father's Side**	**Mother's Side**	**Sibling**	**Detail/Treatment**
ADD				
ADHD				
Learning Disabilities				
Delayed Speech				
Need for Special Education				
Autism/PDD/Aspergers				
Birth Defects/genetic disorders				
Cerebral Palsy				
Delayed speech				
Mental Retardation or slow learner				
Seizures				
Behavior problems				
Depression				
Bipolar/Manic depressive				
Suicide				
Obsessive compulsive disorder				
Tics/Tourettes				
Excessive anxiety				
Medication for mental health				
Other mental health problems				
Thyroid disorders				
Muscular Dystrophy				
Substance abuse				
Other				