

X-linked chondrodysplasia punctata Clinical Questionnaire
Please complete and return with sample

Name: _____

DOB _____

Date _____

Features	Present:	Yes	No
Growth			
IUGR		<input type="checkbox"/>	<input type="checkbox"/>
Failure to Thrive		<input type="checkbox"/>	<input type="checkbox"/>
Gestational Age	_____ wks		
Birth weight	_____ gms		
Birth length	_____ cm		
OFC	_____ cm		
Age at Exam	_____ yrs & mos		
Height	_____ cm		
Weight	_____ kg		
OFC	_____ cm		

Ophthalmologic			
Cataracts		<input type="checkbox"/>	<input type="checkbox"/>
Other		<input type="checkbox"/>	<input type="checkbox"/>
Describe _____			

Craniofacial			
Cleft palate		<input type="checkbox"/>	<input type="checkbox"/>
Other		<input type="checkbox"/>	<input type="checkbox"/>
Describe _____			

Skeletal			
Scoliosis		<input type="checkbox"/>	<input type="checkbox"/>
Chondrodysplasia punctata in infancy		<input type="checkbox"/>	<input type="checkbox"/>
If yes, list major bone segments involved: _____ _____			
Other		<input type="checkbox"/>	<input type="checkbox"/>
Describe _____			

Cardiovascular		<input type="checkbox"/>	<input type="checkbox"/>
If so, what is the defect? _____			

Renal		<input type="checkbox"/>	<input type="checkbox"/>
If so, what is the defect? _____			

Features	Present:	Yes	No
Dermatologic			
Alopecia		<input type="checkbox"/>	<input type="checkbox"/>
Ichthyosis		<input type="checkbox"/>	<input type="checkbox"/>
Atrophoderma		<input type="checkbox"/>	<input type="checkbox"/>
Other		<input type="checkbox"/>	<input type="checkbox"/>
Describe _____			

Developmental Delay		<input type="checkbox"/>	<input type="checkbox"/>
Age Sat	_____		
Age Walked	_____	Verbal IQ	_____
Age Talked	_____	Performance IQ	_____

Biochemical			
Increased 8(9)-cholestenol		<input type="checkbox"/>	<input type="checkbox"/>
Other		<input type="checkbox"/>	<input type="checkbox"/>
Describe _____			

Additional findings: _____

Relevant family history: _____

LAB USE ONLY (Do not write in this box):
Mutation Detected: _____