

ALSTROM SYNDROME PATIENT QUESTIONNAIRE

Today's Date: _____

Name of patient: _____

Gender: _____

Date of birth: _____

Mailing address: _____

Email address: _____ Telephone: _____

Name of father: _____

Name of mother: _____

Siblings name/age: _____

General	
Ethnicity (Paternal/Maternal)	
Known Consanguinity?	
Height	
Weight	
Visual acuity	
Age of onset of first vision symptoms or nystagmus?	
Hearing impairment? <i>Age of onset of hearing loss</i>	
Chronic otitis media (glue ear)	
Balance disturbances?	
Cardiac abnormalities? <i>Age of onset of cardiac abnormalities?</i>	
Has the patient had an echocardiogram? <i>When? What were the results?</i>	
Hypertension?	
Hyperinsulinemia? Glucose tolerance? Age of onset? <i>Please provide test results, if possible</i>	

Diabetes? <i>Age of onset of diabetes?</i>	
Liver function (ALT, AST, GGT) <i>Please provide test results, if possible</i>	
Enlarged liver / spleen?	
Renal function? Renal abnormalities? <i>Please provide creatinine, BUN test results, if possible</i>	
Cholesterol or triglyceride levels elevated? <i>Please provide lipid test results, if possible</i>	
Thyroid function? Sub-clinical hypothyroidism?	
Gastrointestinal problems? (reflux, etc)	
Male Hypogonadism? Normal menstruation or are there menstrual irregularities?	
Digital abnormalities?	
Scoliosis? Skeletal abnormalities?	
Urological symptoms (UTI, retention, incontinence, other?)	
Pulmonary symptoms (chronic bronchitis, frequent pneumonia, COPD, etc)	
Neurological signs? (Absence seizures, autistic spectrum, tics, ataxia, etc?)	
Muscle or joint pain?	
Birth problems? (hypotonia, hypoxia, jaundice, etc)	

Developmental delay? (fine, gross motor skills, language)	
Dental abnormalities?	
Other unusual features? <i>Please explain</i>	

Please attach any other pertinent patient information.