CASE STUDY #2

TURNER SYNDROME

firststep Dx BUCCAL PLUS

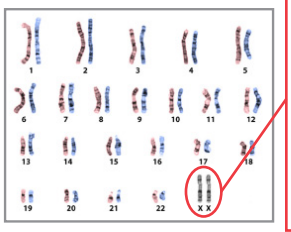
For individuals with autism and other disorders of childhood development

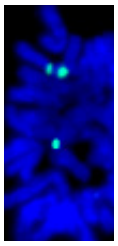
Patient and Clinical Presentation

Medical geneticist ordered FirstStep^{Dx} for 22-month-old female with failure to thrive

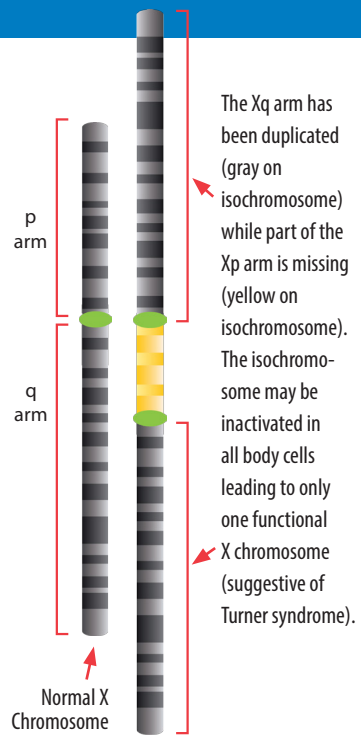
FirstStep^{Dx} Results

- Complex X chromosome rearrangement suggestive of Turner syndrome





Lineagen confirmatory testing shows one normal X (with one centromere) and one isochromosome X (with two centromeres).



Turner Syndrome Characteristics

- Short stature
- Normal intelligence
- Webbed neck
- Delayed puberty
- Possible infertility
- Difficulties with math and social skills
- Incidence of 1 in 2,500 girls
- AAP health care supervision guidelines published

AAP Management Guidance*

TREATMENT

- Growth hormone therapy
- Hypertension treatment
- Hearing loss
- Obesity and weight management
- Glucose intolerance
- Sex hormone therapy

LABORATORY EXAMS

- Blood glucose
- Ultrasonography for UTIs
- Thyroid levels
- Lipid profiles

CLINICAL EXAMS

- Pediatric cardiology evaluation
- Hypertension
- Orthopedic issues
- Ophthalmologic evaluation
- Hip exams for dysplasia
- Hearing screening
- Blood pressure and peripheral pulses
- Renal ultrasound
- Lymphedema
- Feeding problems
- Otitis media exam
- Annual scoliosis evaluation

* Frias JL, Davenport ML, AAP Committee on Genetics. 2003. Health Supervision for Children With Turner Syndrome. *Pediatrics* 111: 692-702.

Case Study Summary

- Early initiation of growth hormone therapy has been shown to be beneficial but most girls with Turner syndrome are diagnosed in late childhood
- Identification of genetic pathology led to significant changes in clinical management
- Genetic counseling sessions focused on understanding this complex genetic mechanism and the risk of recurrence

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Detailed information about FirstStep^{Dx} PLUS is available at www.firststepdx.com or call us at 888-888-6736 (OPEN).