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Dysmorphism And Developmental Delay – A Case Of Xyy Syndrome

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A one year three months old male child first live born to non consanguineously married parents brought by the mother , who is reliable presented with complaints of not attained age appropriate milestones . He is the product of a full tem preganancy delivered via Elective LSCS (Mother had undergone surgical correction for ASD at the age of 21 years) . He was spontaneously conceived one year after a previous spontaneous abortion at third month of amennorhoea , Antenatal period was uneventful . Baby was born with a birth weight of 3 kg and had no significant postnatal or neonatal history .

Baby had history of developmental delay first noticed at 5 months of age , evaluated at local clinic where he was found to be hypothyroid . was started on 12.5 microgram levothyroxine . compliance to the treatment is poor .

He was referred to Child Development Centre , Thiruvananthapuram for evaluation of the above complaints at the age of one year 3 months . At the time of presentation he had attained head control but had not attained self sitting or standing

He weighed around 9 kg (3-15th) had a length of 69 cm (less than 3rd centile) . His Head circumference was 47 cm (50-85th) . On evaluation his tone was normal , reflexes normal but he had an episode of severe temper tantrums which was difficult to be controlled .

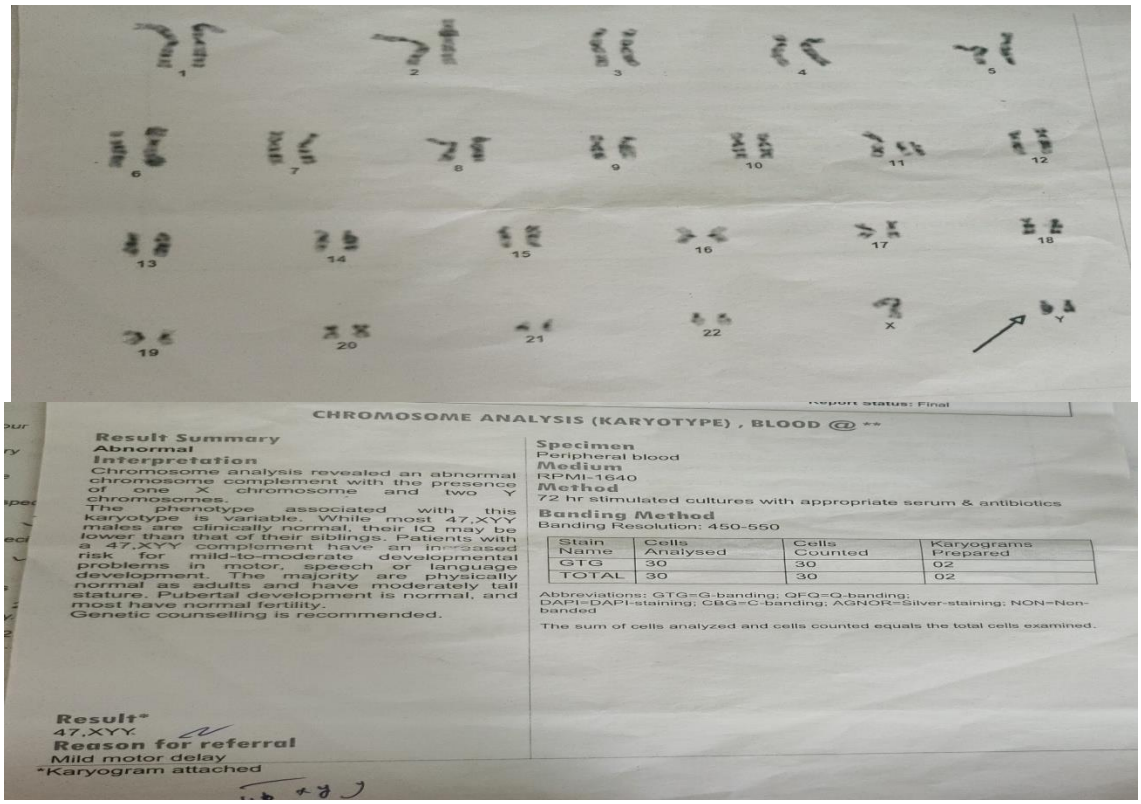
According to the mother, he is irritable and grumpy most of the time .He maintains good eye contact but he uses only one word Amma (mother) which is not intentional . He plays peek a boo with his mother but

does not clap on request . he listens intentionally when spoken to but does not point to objects on commands . Also he does not sustain interest in a picture book for more than 2 minutes .

On developmental evaluation , He earned a grade of V in head control , Grade III in sitting and Grade 0 in standing in CDC Grading of motor milestones ,

On applying DDST II , he had delay in all 4 domains with more delay noted in speech and language . On evaluation of social skills , using VSMS , Social age : was 5.6 months with a social quotient of 33%
Also during the general physical examination , he had short neck , clinodactyly and simian crease .

In view of dysmorphism and global developmental delay , karyotyping was advised and results came out to be XYY syndrome .



CHROMOSOME ANALYSIS (KARYOTYPE) , BLOOD @ **

Report status: Final

Result Summary
Abnormal
Interpretation
Chromosome analysis revealed an abnormal chromosome complement with the presence of one X chromosome and two Y chromosomes.
The phenotype associated with this karyotype is variable. While most 47,XYY males are clinically normal, their IQ may be lower than that of their siblings. Patients with a 47,XYY complement have an increased risk for mild-to-moderate developmental problems in motor, speech or language development. The majority are physically normal as adults and have moderately tall stature. Pubertal development is normal, and most have normal fertility.
Genetic counselling is recommended.

Specimen
Peripheral blood
Medium
RPMI-1640
Method
72 hr stimulated cultures with appropriate serum & antibiotics
Banding Method
Banding Resolution: 450-550

Stain	Cells	Cells	Karyograms
Name	Analysed	Counted	Prepared
GTG	30	30	02
TOTAL	30	30	02

Abbreviations: GTG=G-banding; QFQ=Q-banding; DAPI=DAPI-staining; CBG=C-banding; AGNOR=Silver-staining; NON=Non-banded
The sum of cells analyzed and cells counted equals the total cells examined.

Result*
47,XYY
Reason for referral
Mild motor delay
*Karyogram attached

XYY syndrome also known as Jacob Syndrome that is a rare chromosomal disorder that affects males , marked by the presence of an extra chromosome

Children born with 47 XYY karyotype have an increased growth rate from early childhood . This is attributed to increased gene dosage of three X/Y chromosome, pseudo autosomal region (PAR1) SHOX genes has been postulated as a caise of increased stature in them . Severe acne was noted in a very few early case reports ; but this relationship has been disapproved by current dermatologists . Average IQ was slightly lower than the normal counterparts

Genetics/Basic Defects

1. Caused by an additional Y chromosome in a male
2. Mechanisms of the extra Y chromosome in 47,XYY males

a. Paternal nondisjunction at meiosis II after a normal chiasmate meiosis I (84%)
b. Postzygotic mitotic error or nondisjunction at meiosis II after a nullichiasmate meiosis I (16%)

3. Spermatogenesis

a. Normal spermatogenesis in majority of XYY males. The supernumerary Y chromosome probably lost at the early stage of spermatogenesis ii. A large proportion of primary spermatocytes containing only one Y chromosome
b. Altered spermatogenesis in a proportion of XYY males: Persistence of the supernumerary Y chromosome through meiotic prophase increases spermatocyte degeneration. Though recent studies have disproved it

Clinical Features

No consistent physical stigmata or medical disorders
3. Growth and development
a. Tall stature
b. Larger tooth size
c. At risk for mild speech/language and motor delays and learning disabilities
4. Intelligence a. Normal range b. IQ: 10–15 points lower than siblings
5. Behavioral profile a. Childhood temper tantrums b. No increased incidence of aggression c. Heterosexual
6. Normal reproduction
7. Normal adult adaptation
8. Low fertility

Genetic Counseling

1. Recurrence risk a. Patient's sib: not increased
b. Patient's offspring: Patient generally has chromosomally normal children, despite the high theoretical risk of aneuploidy.
2. Prenatal diagnosis: fetal karyotyping from amniocytes or CVS
3. Management a. The extra Y chromosome represents a risk factor for motor and language development, but the environment remains a primary force in shaping child's development b. The increased frequency of prenatal detection of 47,XYY by amniocentesis necessitates the importance of making accurate information about the developmental prognosis of these individuals. c. Infancy and toddler: assess developmental milestones d. Childhood: assess school performance and provide intervention if needed e. Adolescence: no intervention needed
f. Adulthood annual physical examination

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