

## Turner syndrome in childhood period – A case Report

**Authors:** B. Tuyishimire<sup>1</sup>; O. Karangwa<sup>2</sup>; F. Rutagarama<sup>2</sup>; A. Ndatinya<sup>2</sup> C. Nsanzabaganwa<sup>2</sup>; L. Mutesa<sup>1,\*</sup>

**Affiliations:** <sup>1</sup>Center for Human Genetics, College of Medicine and Health Sciences, The University of Rwanda; Kigali, Rwanda; <sup>2</sup>Department of Pediatrics, Rwanda Military Hospital, Kigali, Rwanda

### ABSTRACT

**INTRODUCTION:** Turner syndrome is a frequent chromosome disorder characterized by short stature, gonadal dysgenesis and multisystem diseases associated with high morbidity and reduced life expectancy

**CASE:** We reviewed an 18 month old patient who presented to the genetics unit at Rwanda Military Hospital (RMH), a tertiary healthcare facility, with a chief complaint of poor weight gain. She was born with a birth weight of 1.76kg and in the neonatal period, paediatricians had noticed dysmorphic features. At the first consultation at the RMH genetics unit, he weighed 5.2kg and was 64 cm tall. Physical examinations revealed some dysmorphic features, including hypertelorism, absent philtrum, short and webbed neck and large low set ears. Cytogenetic analysis showed the chromosomal formula of 45, X0.

**CONCLUSION:** The patient was diagnosed with Turner syndrome based on the cytogenetic analysis and managed with physiotherapy of stimulation and re-education that led to improvements.

**Keywords:** Turner Syndrome, Karyotype, Chromosome, Dysmorphism, Physiotherapy, Case Report

### INTRODUCTION

In clinical practice, Turner syndrome is a frequent chromosome disorder characterized by short stature, gonadal dysgenesis and multisystemic diseases associated with high morbidity and reduced life expectancy [1]. This syndrome affects 1 out of 2,000–4,000 females and occurs in different forms depending on frequent chromosomal abnormalities. These include 45,X or monosomy X that occurs in 40-50% of patients, 45,X/46,XX with mosaicism that affects 15% as well as 45,X/46,XY with mosaicism of Y chromosome materials, affecting 10–12% of Turner syndrome patients [2].

Turner syndrome clinical manifestations include short stature for patients with 45,X karyotype, dysmorphic features, delayed appearance of secondary sexual characteristics and primary amenorrhea [3]. It is also associated with dysmorphic features and other abnormalities, which may include down-turning eyelids, low-set and prominent ears, small jaw, narrow mouth roof, neck webbing, lymphedema, broad chest with widely spaced nipples, ovarian insufficiency or infertility, hearing loss, and kidney defects [2]. Turner syndrome leads to congenital heart defects such as coarctation of the aorta, bicuspid aortic valve and aortic root dilatation, in addition

**\*Corresponding author:** Prof. Leon Mutesa. Center for Human Genetics, College of Medicine and Health Sciences, The University of Rwanda, Email:lmutesa@gmail.com; **Potential Conflicts of Interest (Col):** All authors: no potential conflicts of interest disclosed; **Funding:** All authors: no funding was sought; **Academic Integrity.** All authors confirm that they have made substantial academic contributions to this manuscript as defined by the ICMJE; **Ethics of human subject participation:** The study was approved by the local Institutional Review Board. Informed consent was sought and gained where applicable; **Originality:** All authors: this manuscript is original has not been published elsewhere; **Review:** This manuscript was peer-reviewed by three reviewers in a double-blind review process.

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to obesity, diabetes, and atherogenic lipid profile. Turner syndrome patients have normal intelligence, though some may have problems in the skills of different domains, including nonverbal, social, and psychomotor skills [4]. The most common physical abnormalities affecting females ( $\geq 50\%$ ) with Turner syndrome are: short stature, infertility, estrogen deficiency, hypertension, elevated hepatic enzymes, middle ear infection, bone age retardation, decreased bone mineral content, cubitus valgus, and poor growth during the first postnatal year. Females with Turner syndrome also have significantly higher risks for certain diseases, such as hypothyroidism, osteoporosis, congenital renal malformations, neurovascular disease and cirrhosis, as well as colon and rectal cancers.

### CASE PRESENTATION

An 18 month old patient presented to the genetics unit at the Rwanda Military Hospital (RMH) for poor weight gain. The patient's history revealed that she was born by normal delivery with a

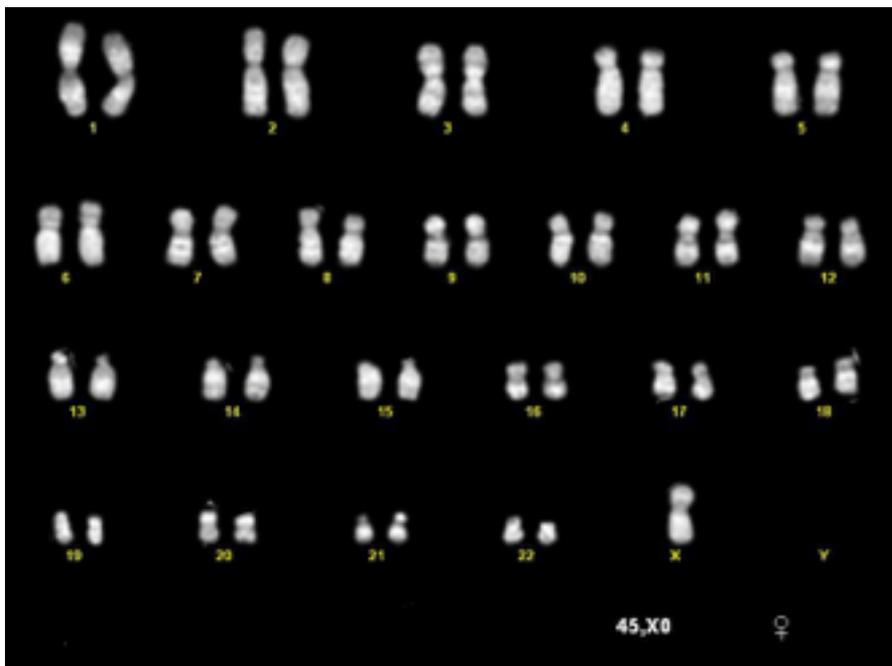
birth weight of 1.76kg and her parents are non-consanguineous whose 3 previous pregnancies were miscarriages. During the neonatal period, the pediatrician noticed dysmorphic features. Our examination revealed that she was small for age, weighing 5.2kg with a height of 64 cm. We noticed some dysmorphic features, such as hypertelorism, absent philtrum, short and webbed neck, large low set ears, and pectus carinatum (Figure 1). On cardiopulmonary examination, she had micrognathia, was cyanotic on both upper and lower limbs and had club feet. Cardiac Doppler ultrasound showed a very small aortic arch, laminar flow, and no significant gradient. She had psychomotor development delays. At her age, she had difficulties in movements and poor sensations, with the absence of basic actions, including sucking, grasping, looking, and listening. A clinical diagnosis of Turner syndrome was made and cytogenetic analysis requested to confirm.

Karyotype test results showed that a chromosomal formula of the patient is 45,X0, implying that the



**Figure 1: Patient's clinical features of Turner syndrome**

*The patient has a short neck with a webbed appearance of the back, low hairline at the back of the neck, low-set ears and swollen hands and feet.*



**Figure 2: Patient's karyotype showing the chromosomal abnormality (45,X0)**

second X is missing (Figure 2).

The patient was referred to the department of physiotherapy for 3 months of 3 weekly stimulation physiotherapy and re-education sessions. Parents were advised about the appropriate for progressive weight gain.

For the time of the follow-up after the first round of 3 month physiotherapy, the patient had started acquiring some development millstone, such as learning to stand. However, she was still presenting muscle weakness. We recommended continuing whole body physiotherapy of stimulation and re-education and scheduled to see the patient once every 3 months.

## DISCUSSION

Based on the karyotype formula, the patient was diagnosed with Turner syndrome, a rare condition in women associated with either complete or partial loss of one X chromosome, often in mosaic karyotypes. It is a syndrome characterized by short stature, delayed puberty, ovarian dysgenesis, hypergonadotropic hypogonadism, infertility, congenital malformations of the heart, endocrine disorders such as type 1 and type 2 diabetes mellitus, osteoporosis as well as autoimmune

disorders [4,5]

Different abnormalities in Turner syndrome depend on its different genotypes. A 45,X0 form of turner syndrome is highly linked with short stature and primary amenorrhea, while Isochromosome Xq and X Mosaicism forms of Turner syndrome are highly linked with secondary amenorrhea. Cardiovascular abnormalities, renal defects, and recurrent otitis media are similarly prevalent in both forms of Turner syndrome [6].

While Turner syndrome is associated with growth hormone deficiency, children and adults with it are, on an average, 20 cm less tall than the female population without this condition [7]. Additionally, the intelligence of individuals with Turner syndrome varies from normal to certain difficulties. Some women with Turner syndrome with normal mental development run normal and successful lifestyles [8]. However, others have specific mental and learning disabilities, which include difficulties in visual-spatial processing, poor sense of direction, trouble appreciating subtle social cues such as facial expressions and problems with nonverbal problem-solving [9].

The management is multidisciplinary and depends on a particular case. Patients with Turner syndrome who have cardiac defects should be monitored and receive care for their heart abnormalities

[4]. Hormonal therapy among children with Turner syndrome improves short stature, sexual development and bone density. While sex hormones are given for life, growth hormones are stopped at the age of 14 [4,10]. During follow-up, they are regularly tested for hearing, blood pressure, dental, skeletal and systemic organ function abnormalities and are managed accordingly. For children, the first treatment option is physiotherapy of stimulation at the early stages of child development to prevent the intellectual disability that may occur throughout the lifespan [4].

Since only 4-5% of women with Turner syndrome have menstruation and can reproduce due to 46,XX/45,X0 mosaicism with normal ovarian cells, adult patients should be counseled about sexuality to address reproduction concerns of patients with Turner syndrome and be ready for alternatives, such as adoption [5,11].

## CONCLUSION

Turner syndrome has a wide range of physical and emotional consequences remarkably in the infant's stage of the child development. Thus, treating patients with Turner syndrome should be done by a multidisciplinary team of geneticist, gynecologists, endocrinologists, psychologists or counsellors, and physiotherapists. While Turner syndrome leads to psychomotor development delays, the first treatment option is physiotherapy of stimulation at early stages of child development to prevent the intellectual disability that may occur throughout the lifespan. However, as they grow up, management should be adjusted to address follow-up outcomes

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