

CASE REPORT

Woman with Down Syndrome delivered a Normal Child

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ABSTRACT

A 25 years old woman with Down syndrome (DS) presented at 9 weeks gestational age. She belonged to a very supportive family. She was married to a person with normal karyotype and conceived within 3 months of marriage. Her antenatal period was uneventful, except for mild IUGR and oligohydroamnios 32 weeks onwards. At 38 weeks, she underwent induction of labor. She tolerated labor pains well but because of failure to progress, cesarean section was performed. She delivered a 2 kg male baby, who was phenotypically and genotypically normal. Screening for trisomy 21 should be offered to all women as part of routine antenatal care. This offer should include detailed counseling about the implications and limitations of the test used in the screening program. The available second trimester screening tests are the double, triple and quadruple tests. Children with Down syndrome may benefit from speech therapy, physical therapy and occupational therapy. They may receive special education and assistance in school. Full potential of Down patients can be utilized only with complete support of family and society.

Keywords: Down syndrome, Fertility, Trisomy 21, Pregnancy.

INTRODUCTION

Down syndrome is the most commonly encountered chromosomal anomaly in medical practice. Trisomy 21, or trisomy G, is a chromosomal disorder caused by the presence of all or part of an extra 21st chromosome. It is named after John Langdon Down, the British physician, who described the syndrome in 1866. The disorder was identified as a chromosome 21 trisomy by Jérôme Lejeune in 1959. The condition is characterized by a combination of major and minor differences in structure. Often, Down syndrome is associated with facial characteristics. Down syndrome in a fetus can be identified with amniocentesis during pregnancy, or in a baby at birth. These patients are of special concern because of their associated problems with regard to mental disability, low IQ, respiratory, cardiovascular and other systemic problems. People with Down syndrome rarely reproduce. Fifteen to thirty percent of women with trisomy 21 are fertile and they have about 50% risk of having a child with Down syndrome. There are few case reports of a man with Down syndrome fathering a child.¹ Here, we are reporting a case of woman with Down syndrome, who had delivered a normal child.

CASE REPORT

A 25 year old woman, known case of Down syndrome (Trisomy 21), was born at mother's age of 21 years. She has

phenotypical features of Down syndrome, such as short stature, moon facies, flat nasal bridge, upward slanting eyes, edema of dorsum of hands and feet. Her karyotype report shows Trisomy 21 (Fig. 1). She belonged to a very supportive family. She was brought up in a much planned way by her parents and grandparents. They taught her to respond well socially. Her IQ was 60 and EQ was 70. She dresses well, can read and write a little. She can do household work, such as cleaning, arranging, cooking, etc. No past history of any admission in hospital or any surgical intervention. Thyroid profile was within normal limits. Patient's 2D echocardiography revealed normal findings. She got married to a man with normal karyotype. Within 3 months of marriage, she conceived. She presented with 9 weeks of pregnancy. Amniocentesis was advised to the patient

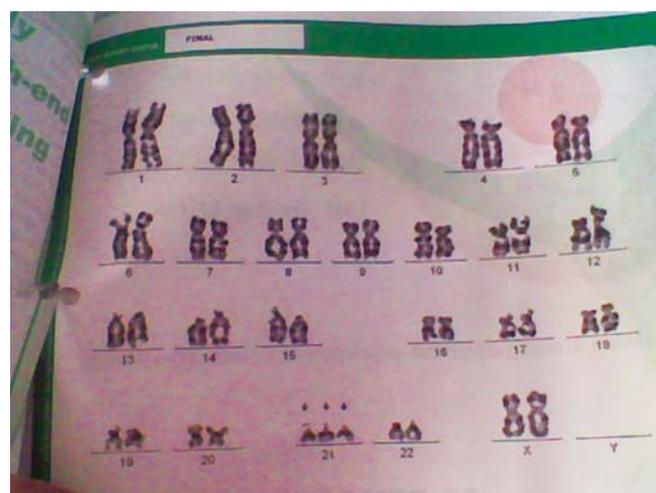


Fig.1: Chromosomal karyotype report showing trisomy 21

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in 15 to 16 weeks. But parents refused to do amniocentesis and wished to continue the pregnancy irrespective of the outcome. Her antenatal period was uneventful except for mild IUGR and oligohydroamnios, 32 weeks onwards. At 38 weeks, she underwent induction of labor. She tolerated labor pains well, but because of failure to progress, lower segment cesarean section under spinal anesthesia was performed uneventfully. She delivered a 2 kg male baby, who was phenotypically and genotypically normal (Fig. 2).

DISCUSSION

The live incidence of Down syndrome is about 1 in 700 births. Approximately, 30% of Down syndrome fetuses miscarry between 12 weeks gestation and term, while it is estimated that 24% will miscarry between 16 weeks and term.² Down syndrome was first described in 1866 and is named after John Langdon Down, the doctor who first identified the syndrome.³ Affected babies are likely to suffer from severe mental disability and have a high incidence of associated physical disabilities, affecting particularly the heart, gastrointestinal tract, eyes, and ears. Individuals with Down syndrome also have a higher incidence of Alzheimer's disease and a 15 to 20 times higher

risk of leukemia.⁴ Twenty percent die by the age of 5 years, usually from cardiac causes, but over half are expected to survive into their fifties.

Women with Down syndrome are less fertile and often have difficulties with miscarriage, premature birth, and difficult labor. Without preimplantation genetic diagnosis, approximately half of the offspring of someone with Down syndrome also have the syndrome themselves. Men with DS are almost uniformly infertile, exhibiting defects in spermatogenesis. Screening for trisomy 21 should be offered to all women as part of routine antenatal care. This offer should include detailed counseling about the implications and limitations of the test used in the screening program. The available second trimester screening tests are double, triple and quadruple tests. When resources are limited, the best first trimester test is combining maternal age with the measurement of nuchal translucency while in the second trimester the quadruple test outperforms the other available tests.⁵ When fetus is diagnosed to have Down syndrome, most parents abort it under social pressure. This case report sets an example that Down babies need not be terminated, if mother's willingness and family support exist. Society must understand that those with Down syndrome have the right to live and can be brought up like normal individuals with adequate behavioral training.

People with Down syndrome experience the same range of sexual feelings as the general population. Teenagers with Down syndrome undergo the same changes at puberty as all adolescents, though these changes may be slightly delayed for boys with Down syndrome. While the overall fertility of men with Down syndrome may be reduced, it is still advisable that couples use contraception whenever pregnancy prevention is desired. At least 50 percent of women with Down syndrome are fertile. Healthy females can use contraception without added medical risk.⁶ Education is an appropriate and highly desirable component in developing positive sexual awareness for the individuals with Down syndrome. There are few case reports (Table 1) across the world on reproduction in mother with Down syndrome.⁷⁻⁹ For the second time in India (of the reported cases



Fig. 2: Woman with Down syndrome carrying a normal baby

Table 1: Reported results of pregnancy in Down syndrome

Paper	Parent	Offspring
Sheridan et al, 1989 ⁷	1 man with Down syndrome	1 normal male
Pradhan M, 2006 ¹	1 man with Down syndrome	1 normal baby
Bovicelli et al, 1982 ⁸	26 women with Down syndrome	10 normal 10 Down syndrome 2 mentally retarded 1 set of premature, nonviable normal twins. 3 malformed 1 slightly microcephaly 1 still born 2 abortion, phenotype unknown
Rani et al, 1990 ¹⁰	1 woman with Down syndrome	1 normal
Kristesashvili DI, 1988 ¹¹	1 woman with Down syndrome	1 normal
Grall JY. Le Goux AM, 1978 ¹²	1 woman with Down syndrome	Multiple malformations with normal karyotype.
ScharrerS. Stengel-Rutkowski S, 1975 ¹³	1 woman with Down syndrome	Aplasia of the left fifth finger, chromosome analysis revealed few hyperdiploid but no G-trisomic cells.

in literature) a mother suffering from Down syndrome has delivered a normal child.¹⁰

There is no medical cure for Down syndrome. However, children with Down syndrome would benefit from early medical assistance and developmental interventions beginning during infancy. Children with Down syndrome may benefit from speech therapy, physical therapy and occupational therapy. They may receive special education and assistance in school. The general health and quality of life for people with Down syndrome has improved drastically in recent years. Many adult patients are healthier, live longer and participate more actively in society due to early intervention and therapy.

CONCLUSION

A woman with Down syndrome has mothered a phenotypically and genotypically normal child. Full potential of Down patients can be utilized only with complete support of family and society. Children with Down syndrome may benefit from speech therapy, physical therapy and occupational therapy. They may receive special education and assistance in school.

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