UNIVERSA MEDICINA

pISSN: 1907-3062 / eISSN: 2407-2230

Univ Med 2026;45:XXX

DOI: https://doi.org/10.18051/UnivMed.2026.v45.XXX

CASE REPORT

Genetic counselling problems with parents of child with ROB der(21;21)(q10:q10) Down syndrome: a case report

Nisa. A.T. Hadi[1], Agustini Utari[2,3]*, and Tri I. Winarni[3]

[1]Magister of Biomedical Science - Genetic Counselling Program, Faculty of Medicine, Universitas Diponegoro, Semarang, Indonesia
[2]Department of Pediatrics, Faculty of Medicine, Universitas Diponegoro, Semarang, Indonesia
[3]Center for Biomedical Research (CEBIOR), Faculty of Medicine, Universitas Diponegoro, Semarang, Indonesia

*** Correspondence Author:**
agustiniutari@gmail.com

Date of first submission, January 30, 2025
Date of final revised submission, June 18, 2026
Date of acceptance, July 20, 2026

Cite this article as: Hadi NAT, Utari A, Winarni TI. Genetic counselling problems with parents of child with ROB der(21;21)(q10:q10) Down syndrome: a case report. Univ Med 2026;45:XXX

ABSTRACT

BACKGROUND

Down syndrome (DS) is a genetic disorder caused by addition of a full or partial chromosome 21. The most common type of DS (90–95%) is pure trisomy 21; mosaicism occurs in 6–7%, and about 3–5% comprises translocation DS including Robertsonian translocation, which could be inherited from parents. This case shows a Robertsonian translocation der(21;21)(q10:q10) DS and the common financial problem among Indonesian parents being their inability to pay for examinations to determine the origin of the abnormality (inherited or de novo), which is a big challenge for genetic counselors.

CASE DESCRIPTION

A 2-month-old baby boy was admitted to the hospital with suspect Down syndrome referred by a pediatrician. The patient was the only child of 25-year-old parents. Physical examination showed several characteristics of DS, including upslanting palpebral fissure, a flat nasal bridge, horizontal palmar creases, sandal gap, and continuous murmur in upper left sternal border due to patent ductus arteriosus. Cytogenetic testing revealed the karyotype of derivative chromosome 21, namely Robertsonian translocation 46,XY,+21,der(21;21)(q10:q10). Parents declined to order cytogenetic testing due to financial concerns.

CONCLUSION

Parents' karyotype is critical to confirm the origin of the translocation. It is important to calculate the recurrence risk correctly especially when parents still want another child and to do non-directive counseling in terms of reproductive options. However, it cannot be done in this case because of the lack of parent's karyotypes. Genetic counseling plays an essential role in providing an accurate recurrence risk based on the type of trisomy for future offspring and reproductive planning.

Keywords : Genetic counselling, down syndrome, parental karyotyping

INTRODUCTION

Down syndrome (DS) is a genetic disorder caused by the presence of the whole or part of a third chromosome 21.[1] The incidence of DS is 1 in 1000 to 1 in 1100 live births. Each year, approximately 3,000 to 5,000 children are born with this chromosomal disorder. Down syndrome is the most common genetic cause of intellectual disability occurring in children.[2]

The extra copy of chromosome 21 results from a separation failure during gametogenesis.[1] The type of underlying cytogenetic abnormality determines the phenotypic expression in DS. The most common type of DS (90-95%) is pure trisomy of chromosome 21; mosaicism also occurs in DS in 6-7% of cases, and about 3-5% are caused by a Robertsonian translocation.[2,3]

Robertsonian translocation is the most common structural chromosomal rearrangement in the population, with an incidence of 1 in 1000 newborns.[4] Robertsonian translocation results from the breakage of two acrocentric chromosomes due to a crossing-over error during meiosis I with a subsequent fusion of their long arms to form one chromosome.[4,5] The fusion between two acrocentric chromosomes may occur in chromosomes 13, 14, 15, 21, and 22, which do not have a recognizable short ("p")-arm, and is called a satellite chromosome that appears as a p-arm in a condensed form.[6] Robertsonian translocation is dominated by der(14;21)(q10;q10), in which 70% are de novo cases. Another type of Robertsonian translocation is der(21;21)(q10;q10), although very few cases are inherited from parents (~approximately 5%). However, if the parent is a carrier, the recurrence risk is more than 10%.[7]

The aim of this case report was to provide genetic counseling education on the case of an only child with Robertsonian translocation Down syndrome of parents who refuse cytogenetic testing.

CASE REPORT

A 2-month-old male patient was admitted to the hospital with Down syndrome diagnosed by a pediatrician. The patient was the only child of 25-year-old parents (the mother being P1G1A0) and was delivered by C-section due to macrosomia (4950 grams) at term and a history of jaundice on his first day of life. The perinatal history was unremarkable; the mother had routine prenatal checkups every month and no history of illness during pregnancy. There was no consanguineous marriage and no history of a similar case in the family [Figure 1].

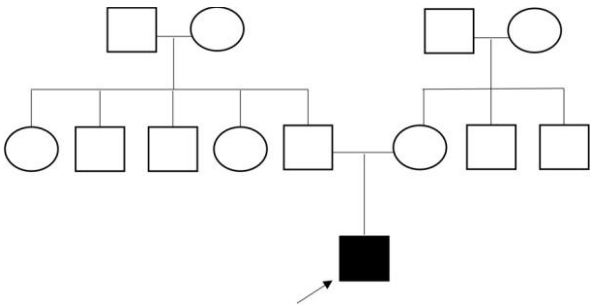

Figure 1. Pedigree of the patient. Patient is the only child, there is no family history with similar condition

Physical examination showed an upslanting palpebral fissure, flat nasal bridge, horizontal palmar creases, sandal gap, and continuous murmur at upper left sternal border. Patent ductus arteriosus (PDA) was diagnosed by echocardiography. In addition, there was distichiasis in both eyes, normal hearing function in the right ear, and sensory hearing problem in the left ear. A thyroid profile was done to exclude the possibility of congenital hypothyroidism; the result was within the normal range.

The patient was referred to the Center for Biomedical Research (CEBIOR) Laboratory, Faculty of Medicine, Universitas Diponegoro, Semarang, Central Java, Indonesia, for cytogenetic testing due to a presumptive diagnosis of DS. Informed consent was given throughout the examination. There was no numerical chromosomal abnormality but a structural chromosomal abnormality was found on chromosome 21 (translocation on chromosome 21 to 21) i.e., 46,XY,+21,der(21;21)(q10:q10). Parents declined to perform cytogenetic testing after genetic counseling post-testing was performed [Figure 2]. The karyotype of parents is needed to confirm the origin of the translocation, whether it is de novo or inherited, to calculate the recurrence risk because the parents want to have another child.

Informed consent from the patient was obtained and ethics approval was granted by the Ethics Committee of Faculty of Medicine, Diponegoro, University (Number 100/EC/KEPK/FK- /VI/2020).

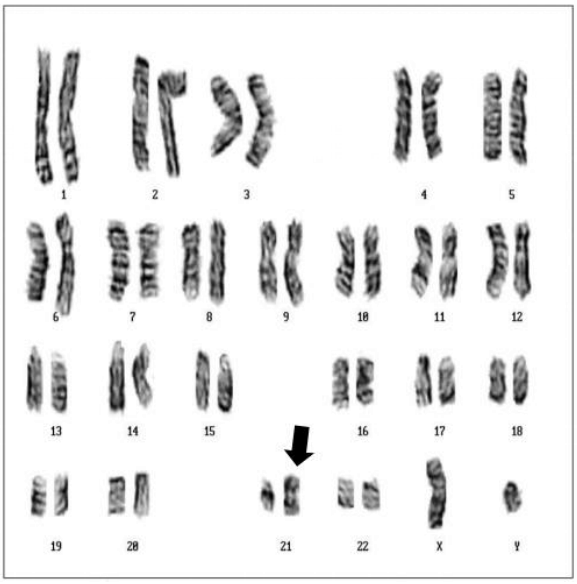

Figure 2. Karyogram of the patient showing a male karyotype with 46,XY,+21,DER(21;21)(Q10:Q10)

DISCUSSION

A baby boy with translocation Down syndrome involving Robertsonian der(21;21)(q10:q10) was reported. The most common type of DS is non-inherited Down syndrome called classical type in approximately 90-95%, while in our case, a translocation type was found. Thus, the origin of the abnormality should be explored, whether it is de novo or inherited.

Once an unbalanced translocation in the child has been identified, parental cytogenetics is essential to rule out the possibility of a balanced translocation of the parents. Cytogenetic evaluation is needed to perform recurrence risk calculation during counseling to discuss reproductive possibilities and options. Genetic counseling has to be performed to help parents understand the significance of genetic conditions and facilitate them with the best available reproductive options. In order to provide genetic counseling and reproductive planning, it is important to ascertain whether the identified variant is inherited or represents a de novo mutation. Testing in multiple family members can yield a more accurate estimation of the penetrance and expressivity of a particular variant within the family regarding inherited findings.[8]

A few similar cases of Robertsonian translocation Down syndrome involving der(21;21)(q10:q10) have been recorded only in a limited number of case reports worldwide. Earlier reports from India [9], Kosovo [10], Sri Lanka [11], and Iran [12] described patients presenting with typical Down syndrome phenotypes associated with Robertsonian translocation [Table 1]. The present case has many similarities with those previously reported in the literature, including characteristic dysmorphic features such as upslanting palpebral fissures, flat nasal bridge, and congenital heart disease. Most reported cases emphasized the importance of parental cytogenetic analysis.

In this case, the parents refused to carry out cytogenetic testing due to financial problems, which is a prevalent issue among Indonesian parents. Genetic counselors are most concerned with how to manage incomplete data. For incomplete parental data, partially observed parental phenotypes matching the offspring's phenotype configurations are sufficient statistics to conclude whether it is inherited or de novo.[13,14]

The risk of classical DS increases gradually with maternal age. A previous study found that young maternal age has a risk of 1/1466 for classic de novo DS.[15] On the other hand, maternal age does not influence the risk of having translocation DS. However, genetic counselors should be suspicious of the possibility of a balanced translocation carrier parent. Genetic counselors cannot conclude the origin of abnormality (de novo or inherited) without parent cytogenetic testing results and thus the calculation of recurrence risk. In such a case a literature-based study can be used to deal with incomplete data.

Theoretically, more than 50% of the translocations in a fetus are de novo. If the parents have a normal karyotype, the recurrence risk is approximately 1%, similar to classical trisomy.[16] On the other hand, for familial translocations, the recurrence risk varies between 1-15% depending on the type of translocations and whether the mother or the father is the carrier. However, the recurrence risk of der(21;21)(q10:q10) is 100% when both father or mother are carriers.[5,17] In such a case, adoption is the best possible solution that a genetic counselor should communicate if the affected individual wishes to have another child.

Table 1. Comparison of published cases of Robertsonian translocation der(21;21)(q10:q10): clinical findings and genetic evaluation with the present case

Authors	Karyotype	Parent testing results	Physical findings	Outcomes
Kusre et al., India[9]	der(21;21)(q10:q10)	Normal parental karyotype	Typical DS features, congenital heart disease	Considered de novo translocation
Kolgeci et al., Kosovo[10]	der(21;21)(q10:q10)	Mother was balanced carrier	DS phenotype with family history of miscarriage	High recurrence risk identified
Kollurage et al., Sri Lanka[11]	rob(21;21)(q10:q10)	Normal parental karyotype	Typical DS clinical manifestations	Considered de novo translocation
Nikfar et al., Iran[12]	der(21;21)(q10:q10)	Normal parental karyotype	Typical DS clinical manifestations	Considered de novo translocation
Present case, Indonesia	der(21;21)(q10:q10)	Decline due to financial issues	Typical DS features and PDA	Recurrence risk could not be accurately determined

Note : DS : Down syndrome; PDA : Patent ductus arteriosus

Genetic counseling plays an essential role in providing accurate and relevant medical information and recurrence risk for the subsequent offspring and reproductive planning. In addition, it is also important for genetic counselors to have the expertise to provide empathy, anticipatory guidance, support materials, and concern about parent's psychology to accept and nurture their child, by assisting families to concentrate on their child's potential and quality of life,[18] as well as to facilitate understanding and acceptance to increase parents' perceived personal control by explaining scientific causes and exploring parents' personal beliefs.[19] Psychosocial aspects are an important part that a genetic counselor needs to handle carefully through each phase.[20]

Furthermore, there are issues when the process of post-genetic counseling sessions is not performed appropriately. This is shown by the parents' limited understanding after counseling sessions leading them to decline cytogenetic testing, which is the crucial part in determining the origin of the abnormality, such that the recurrence risk calculation and precise reproductive planning can be performed accurately. Otherwise, parents should be offered sufficient opportunity to process information, deliberate on options, express their values and beliefs, and make informed decisions. The re-genetic counseling sessions should be performed after parents have taken enough time to deal with their psychosocial aspects.

Future direction

Genetic counselors need to be concerned about the psychosocial aspects of the parents or affected individuals, as the psychosocial impact is of utmost importance in our society. The better psychosocial aspect can significantly support knowledge-based outcomes. The rejection or denial phase may be observed at the first or second genetic counseling session, underscoring the need for giving the parents sufficient time and opportunity to process all medical information and deliberate on options and appreciate them. Re-genetic counseling sessions should be implemented during the precise psychosocial phase.

CONCLUSIONS

The present case was Down syndrome with a rare unbalanced Robertsonian translocation der(21;21)(q10:q10) and parents who declined to do cytogenetic testing. For the important task of calculating the recurrence risk correctly and genetic counseling, parental karyotyping should have been performed to determine the possible cause of this patient's case. It is crucial to highlight the significant role of genetic counseling in genetic disorders, guiding the family by providing vital information and options for better long-term outcomes.

Conflict of interest

All the authors declare that there are no conflicts of interest

Acknowledgement

We thank the patient and his parents for the willingness participation in this study

Authors' Contributions

NH, AU, TW contributed to the study's design and data analysis. NH performed the

manuscript preparation. AU performed the physical examination and genetic counselling. TW analyzed the cytogenetic examination. AU and TW provided critical revisions to the manuscript and supervised the research project. All authors have read and approved the final manuscript.

Funding

This study received no external funding.

Data Availability Statement

Derived data supporting the findings of this study are available from the corresponding author on request

Declaration of AI Usage in Scientific Writing

We hereby confirm that no artificial intelligence (AI) tools were used in the drafting and editing of the manuscript, including assistance with language enhancement, structure, and citation formatting.

REFERENCES

1. Pal A, Daley SF. Down syndrome. Treasure Island (FL): StatPearls Publishing; 2026.
2. Anil V, Anirban M. De novo Robertsonian translocation t(21;21) in a child with Down syndrome. J Nepal Paediatr Soc 2017;37:92-4. https://doi.org/10.3126/jnps.v37i1.16184.
3. Karan V, Rosemary I, Kayla M, Preshon P, Alexander F, Mary M. Disparities and outcomes of patients living with Down syndrome undergoing healthcare transitions from pediatric to adult care: a scoping review. Am J Med Genet A 2022;1882293-2302. https://doi.org/10.1002/ajmg.a.62854.
4. Lu W, Zhou J, Rao H, et al. A retrospective analysis of Robertsonian translocations from a single center in China. Reprod Sci 2024;31:851–6. doi: 10.1007/s43032-023-01398-3.
5. Zhang S, Lei C, Wu J, et al. Meiotic heterogeneity of trivalent structure and interchromosomal effect in blastocysts with Robertsonian translocations. Front Genet 2021;12:609563. doi:10.3389/fgene.2021.609563.
6. Song J, Li X, Sun L, et al. A family with Robertsonian translocation: a potential mechanism of speciation in humans. Mol Cytogenet 2016;9:48. doi: 10.1186/s13039-016-0255-7.
7. Kalpana VL, Gowri Sai Ram PVV, Soujanya P, Anuradha A, Papa Kusuma B, Uma Bharati H. Robertsonian translocations t(21q;21q) and t(14q;21q) in Down syndrome. Int J Med Res Health Sci 2017;6:53-8.
8. Maya I, Sukenik-Halevy R, Basel-Salmon L, Sagi-Dain L. Ten points to consider when providing genetic counseling for variants of incomplete penetrance and variable expressivity detected in a prenatal setting. Acta Obstet Gynecol Scand 2020;99:1427-9. doi: 10.1111/aogs.13963.
9. Kusre G, Sarma M, Nirmolia T, Shankarishan P. Robertsonian translocation t(21; 21) in a female born to normal parents: a case report. J Clin Diagn Res 2015;9:GD01-2. doi: 10.7860/JCDR/2015/10638.5372.
10. Kolgeci S, Azemi M, Ahmeti H, Dervishi Z, Sopjani M, Kolgeci J. Recurrent abortions and Down syndrome resulting from Robertsonian translocation 21q; 21q. Med Arch 2012;66:350-2. doi: 10.5455/medarh.
11. Kollurage DUA, Kudagammana ST. Down syndrome caused by 21;21 Robertsonian translocation. Sri Lanka J Med 2018;27:43-5. doi:10.4038/sljm.v27i2.85.
12. Nikfar A, Mansouri M, Fatemi Abhari G. A rare de novo Robertsonian translocation t(21q;21q) in an Iranian child with Down syndrome: a case report. Acta Med Iran 2019;57:522-4. doi:10.18502/acta.v57i8.2430.
13. Van Steen K, Laird NM, Markel P, Molenberghs G. Approaches to handling incomplete data in family-based association testing. Ann Hum Genet 2007;71(Pt 2):141-51. doi: 10.1111/j.1469-1809.2006.00325.x.
14. Harris A, Bather JR, Bradshaw RL. Incomplete family history and meeting algorithmic criteria for genetic evaluation of hereditary cancer. JAMA Netw Open 2025;8:e2539870. doi: 10.1001/jamanetworkopen.2025.39870.
15. Mai CT, Isenburg JL, Canfield MA, et al. National population-based estimates for major birth defects, 2010-2014. Birth Defects Res 2019;111:1420-35. doi: 10.1002/bdr2.1589.
16. Priya PK, Mishra VV, Roy P, Patel H. A study on balanced chromosomal translocations in couples with recurrent pregnancy loss. J Hum Reprod Sci 2018;11:337-42. doi: 10.4103/jhrs.JHRS_132_17.
17. Wiland E, Olszewska M, Woźniak T, Kurpisz M. How much, if anything, do we know about sperm chromosomes of Robertsonian translocation carriers? Cell Mol Life Sci 2020;77:4765–85. doi:10.1007/s00018-020-03560-5.
18. Crellin E, Martyn M, McClaren B, et al. What matters to parents? A scoping review of parents' service experiences and needs regarding genetic testing for rare diseases. Eur J Hum Genet 2023;31:869–78. doi: 10.1038/s41431-023-01376-y.
19. Miller EG, Young JL, Rao A, Ward-Lev E, Halley MC. Demographic characteristics associated with perceptions of personal utility in genetic and

genomic testing: a systematic review. JAMA Netw Open 2023;6:e2310367. doi: 10.1001/jamanetworkopen.2023.10367.

20. Higgs E, Wain KE, Wynn J, et al. Measuring quality and value in genetic counseling: the current landscape and future directions. J Genet Couns 2023;32:315-24. doi: 10.1002/jgc4.1657.