

Mosaic Trisomy 22 References

Abdelgadir, D., Nowaczyk, M. J., & Li, C. (2013). Trisomy 22 mosaicism and normal developmental outcome: report of two patients and review of the literature. *American Journal of Medical Genetics. Part A*, 161A(5), 1126–1131.

<https://doi.org/10.1002/ajmg.a.35812>

Abdelmoula, NB, Ayadi, N., Amouri, A, Meddeb, M., Elguezal, H. Saad, A., Karray A., Chaabouni, M., and Rebai, T. (2005) Mosaic trisomy 22 in a malformed newborn female: a new case. *J.J. M. Sfax*. 1(9):51-55.

<https://www.medecinesfax.org/useruploads/files/article11-9-10.pdf>

Baur Cavegn B, Popelka J, Arioglu S, Todesco Bernasconi M (2023) Live-Born with a Trisomy 22 Mosaic: A Case Report. *Ann Case Report* 8: 1405.

<https://doi.org/10.29011/2574-7754.101405>

Basaran, N., Berkil, H., Ay, N., Durak, B., Ataman, C., Ozdemir, M., Ozon, Y. H., & Kaya, I. (2001). A rare case: mosaic trisomy 22. *Annales de Genetique*, 44(4), 183–186. [https://doi.org/10.1016/s0003-3995\(01\)01095-4](https://doi.org/10.1016/s0003-3995(01)01095-4)

Carazo, M.J., Lopez-Exposito, L., Glover, G & del Carpio, DE. (2005) Trisomia 22 mosaico como causa disgenesia ovarica asociada a un fenotipo Turner-Ullrich. *Prog. Obstet Ginecol*. 48(8): 398-403.

<https://www.elsevier.es/es-revista-progresos-obstetricia-ginecologia-151-articulo-trisomia-22-mosaico-como-causa-13078324>

Chen, C. P., Huang, M. C., Chern, S. R., Wu, P. S., Chen, S. W., Chuang, T. Y., Town, D. D., & Wang, W. (2019). Mosaic trisomy 22 at amniocentesis: Prenatal diagnosis and literature review. *Taiwanese Journal of Obstetrics & Gynecology*, 58(5), 692–697. <https://doi.org/10.1016/j.tjog.2019.07.020>

Crowe, C. A., Schwartz, S., Black, C. J., & Jaswaney, V. (1997). Mosaic trisomy 22: a case presentation and literature review of trisomy 22 phenotypes. *American Journal of Medical Genetics*, 71(4), 406–413.

[https://doi.org/10.1002/\(SICI\)1096-8628\(19970905\)71:4<406::AID-AJMG7>3.0.CO;2-R](https://doi.org/10.1002/(SICI)1096-8628(19970905)71:4<406::AID-AJMG7>3.0.CO;2-R)

Cunha, S., Meireles, D., Figueiredo, C., Soares, A., Freitas, J., Oliveira, M., & Borges, T. (2021). Mosaic trisomy 22 in a 14-year old adolescent: A case report. *Portuguese Journal of Pediatrics*. (52)127-30.

<https://ojs.pjp.spp.pt/article/view/20175/17812>

Dayasiril, KC, De Silva, D., and Weerasekara, K. (2018) Confirmation of mosaic trisomy 22 in an infant with failure to thrive. *Sri Lanka Journal of Child Health*. 47(2): 174-176. <https://sljch.sljol.info/articles/10.4038/sljch.v47i2.8488>

de Ravel, T. J., Legius, E., Brems, H., Van Hoestenbergh, R., Gillis, P. H., & Fryns, J. P. (2001). Hemifacial microsomia in two patients further supporting chromosomal mosaicism as a causative factor. *Clinical Dysmorphology*, 10(4), 263–267. <https://doi.org/10.1097/00019605-200110000-00005>

Dulitzky, F., Shabtal, F., Zlotogora, J., Halbrecht, I., & Elian, E. (1981). Unilateral radial aplasia and trisomy 22 mosaicism. *Journal of Medical Genetics*, 18(6), 473–476. <https://doi.org/10.1136/jmg.18.6.473>

Florez, L., & Lacassie, Y. (2005). Mosaic trisomy 22: report of a patient with normal intelligence. *American Journal of Medical Genetics. Part A*, 132A(2), 223–225. <https://doi.org/10.1002/ajmg.a.30401>

Gholkar, N, Singh, C., & Kaul, A. (2018) Antenatal detection of mosaic trisomy 22 with a finding of Blake's pouch cyst. *J. Fetal Med*. 5:53-57. <https://link.springer.com/article/10.1007/s40556-017-0144-0>

Guzé, C., Qin, N., Kelly, J., Yang, X., Bruni, R., Baker, D., & Hassan, R. (2004). Isochromosome 22 in trisomy 22 mosaic with five cell lines. *American Journal of Medical Genetics. Part A*, 124A(1), 79–84. <https://doi.org/10.1002/ajmg.a.20365>

Hall, T., Samuel, M., & Brain, J. (2009). Mosaic trisomy 22 associated with total colonic aganglionosis and malrotation. *Journal of Pediatric Surgery*, 44(1), e9–e11. <https://doi.org/10.1016/j.jpedsurg.2008.09.032>

Hirschhorn K. (1998). Mosaic trisomy 22: a case presentation and literature review of trisomy 22 phenotypes. *American Journal of Medical Genetics*, 76(5), 447. [https://doi.org/10.1002/\(sici\)1096-8628\(19980413\)76:5<447::aid-ajmg15>3.0.co;2-m](https://doi.org/10.1002/(sici)1096-8628(19980413)76:5<447::aid-ajmg15>3.0.co;2-m)

Kalayinia, S., Shahani, T., Biglari, A., Maleki, M., Rokni-Zadeh, H., Razavi, Z., & Mahdieh, N. (2019). Mosaic trisomy 22 in a 4-year-old boy with congenital heart disease and general hypotrophy: A case report. *Journal of Clinical Laboratory analysis*, 33(2), e22663. <https://doi.org/10.1002/jcla.22663>

Leclercq, S., Baron, X., Jacquemont, M. L., Cuillier, F., & Cartault, F. (2010). Mosaic trisomy 22: five new cases with variable outcomes. Implications for genetic counselling and clinical management. *Prenatal Diagnosis*, 30(2), 168–172. <https://doi.org/10.1002/pd.2427>

Lewis, B., Fulton, S., Short, E., Nelson, S., Lombardi, G., Rosenbaum, D., Kerckmar, C., Baley, J., & Singer, L. T. (2007). A longitudinal case study of a child with mosaic trisomy 22: language, cognitive, behavioral, physical, and dental outcomes. *American Journal of Medical Genetics. Part A*, 143A(17), 2070–2074. <https://doi.org/10.1002/ajmg.a.31866>

Lessick, M. L., Szego, K., & Wong, P. W. (1988). Trisomy 22 mosaicism with normal blood chromosomes. Case report with literature review. *Clinical Pediatrics*, 27(9), 451–454. <https://doi.org/10.1177/000992288802700908>

Mazza, V., Latella, S., Fenu, V., Ferrari, P., Bonilauri, C., Santucci, S., & Percesepe, A. (2010). Prenatal diagnosis and postnatal follow-up of a child with mosaic trisomy 22 with several levels of mosaicism in different tissues. *The Journal of Obstetrics and Gynaecology Research*, 36(5), 1116–1120. <https://doi.org/10.1111/j.1447-0756.2010.01278.x>

Mollica, F., Sorge, G., & Pavone, L. (1977). Trisomy 22 mosaicism. *Journal of Medical Genetics*, 14(3), 224–225. <https://doi.org/10.1136/jmg.14.3.224>

Nardelli, A., Laskoski, L. V., Luiz, A. F., Silveira, M. A. D., & d'Arce, L. P. G. (2023). Occurrence of mosaic trisomy 22 and pericentric inversion of chromosome 9 in a patient with a good prognosis. *BMC Medical Genomics*, 16(1), 286. <https://doi.org/10.1186/s12920-023-01709-2>

Pridjian, G., Gill, W. L., & Shapira, E. (1995). Goldenhar sequence and mosaic trisomy 22. *American Journal of Medical Genetics*, 59(4), 411–413. <https://doi.org/10.1002/ajmg.1320590402>

Ruiter, E. M., Toorman, J., Hochstenbach, R., & de Vries, B. B. A. (2004). Mosaic trisomy 22 in a boy with a terminal transverse limb reduction defect. *Clinical Dysmorphology*, 13(2), 99–102. https://journals.lww.com/clindysmorphol/abstract/2004/04000/mosaic_trisomy_22_in_a_boy_with_a_terminal.9.aspx

Schinzel A. (1981). Incomplete trisomy 22. III. Mosaic-trisomy 22 and the problem of full trisomy 22. *Human Genetics*, 56(3), 269–273. <https://doi.org/10.1007/BF00274677>

Thomas, S., Parker, M., Tan, J., Duckett, D., & Woodruff, G. (2004). Ocular manifestations of mosaic trisomy 22: a case report and review of the literature. *Ophthalmic Genetics*, 25(1), 53–56. <https://doi.org/10.1076/opge.25.1.53.29004>

Trevisan, V., Meroni, A., Leoni, C., Sirchia, F., Politano, D., Fiandrino, G., Giorgio, V., Rigante, D., Limongelli, D., Perri, L., Sforza, E., Leonardi, F., Viscogliosi, G., Contaldo, I.,

Orteschi, D., Proietti, L., Zampino, G., & Onesimo, R. (2024). Trisomy 22 Mosaicism from Prenatal to Postnatal Findings: A Case Series and Systematic Review of the Literature. *Genes*, 15(3), 346. <https://doi.org/10.3390/genes15030346>

Wang, J. C., Dang, L., Mondal, T. K., & Khan, A. (2007). Prenatally diagnosed mosaic trisomy 22 in a fetus with left ventricular non-compaction cardiomyopathy. *American Journal of Medical Genetics. Part A*, 143A(22), 2744–2746. <https://doi.org/10.1002/ajmg.a.32004>

Wertelecki, W., Breg, W. R., Graham, J. M., Jr, Iinuma, K., Puck, S. M., & Sergovich, F. R. (1986). Trisomy 22 mosaicism syndrome and Ullrich-Turner stigmata. *American Journal of Medical Genetics*, 23(3), 739–749. <https://doi.org/10.1002/ajmg.1320230302>

Zhang, Z., Cao, D., Xu, Z., & Jiang, W. (2021). Prenatal diagnosis of low-level trisomy 22 mosaicism with a favorable outcome. *Taiwanese Journal of Obstetrics & Gynecology*, 60(2), 380–381. <https://doi.org/10.1016/j.tjog.2021.01.025>