

Errata Sheet

Errata sheet with the correction to errors in the doctoral thesis entitled “Novel genetic causes of childhood cancer predisposition” by Carolina Maya González (2025). Karolinska Institutet. Thesis. <https://doi.org/10.69622/28194191> ISBN: 978-91-8017-470-1.

Section name or page number in the thesis	Error in the thesis	Correction
List of constituent scientific papers in this thesis, Related scientific papers and Additional scientific papers	The volume and article number are missing from the published constituent, related and additional scientific papers in the thesis.	Corrected references below.
Page 14 (Figure 5)	Both pathogenic variants are shown on the sister chromatids of a chromosome.	Each chromosome should have a genetic variant. The corrected figure is presented below.
Page 26	Currently approved targeted cancer treatments include immunotherapies and immune checkpoint inhibitors.	Currently approved targeted cancer treatments include immunotherapies as cancer vaccines, CAR T-cells, immune checkpoint inhibitors and some monoclonal antibodies.
Page 30 (Table 3)	Swedish National patient Register - Congenital diagnoses. Details on inpatient and outpatient stays since 1987 and 2001.	Swedish National Patient Register - Congenital diagnoses. The register starts in 1964. Details on nationwide inpatient and outpatient stays since 1987 and 2001, respectively, although outpatient care coverage does not reach 100%.
Page 32 (Figure 10)	Short read whole genome sequencing workflow used.	Short read whole genome sequencing workflow used (Illumina Inc).
Page 34 (Figure 12)	Overview of droplet digital PCR.	Overview of droplet digital PCR used (Bio-Rad).
Page 34 (Figure 13)	The principle of MS-MLPA.	The principle of MS-MLPA (MRC-Holland, Amsterdam, The Netherlands).
Page 42 (Figure 17)	<i>De novo</i> nonsense <i>FBN1</i> p.C805*.	<i>De novo</i> frameshift variant resulting in premature stop codon (<i>FBN1</i> p.C805*).
Page 42	The second patient was a woman with MFS caused by a <i>de novo FBN1</i> nonsense variant (p.C805*) –a mutation resulting in an early stop in RNA translation.	The second patient was a woman with MFS caused by a <i>de novo FBN1</i> frameshift variant (p.C805*) –a two base-pair deletion changing the reading frame and resulting in an early stop in RNA translation.

Corrected references for published papers:

Page 13. Constituent scientific papers

II. Maya-González C, Wessman S, Lagerstedt-Robinson K, Taylan F, Tesi B, Kuchinskaya E, McCluggage WG, Poluha A, Holm S, Nergårdh R, Díaz De Ståhl T, Höybye C, Tettamanti G, Delgado-Vega AM, Skarin Nordenvall A[#], Nordgren A[#]. Register-based and genetic studies of Prader-Willi syndrome show a high frequency of gonadal tumors and a possible mechanism for tumorigenesis through imprinting relaxation. *Front Med (Lausanne)*. 2023 Jul 28;10:1172565. doi: 10.3389/fmed.2023.1172565.

III. Maya-González C, Delgado-Vega AM, Taylan F, Lagerstedt Robinson K, Hansson L, Pal N, Fagman H, Puls F, Wessman S, Stenman J, Georgantzi K, Fransson S, Díaz De Ståhl T, Ek T, Palmer R, Tesi B, Kogner P[#], Martinsson T[#], Nordgren A[#]. Occurrence of cancer in Marfan syndrome: Report of two patients with neuroblastoma and review of the literature. *Am J Med Genet A*. 2024 Dec;194(12):e63812. doi: 10.1002/ajmg.a.63812.

IV. Maya-González C, Díaz De Ståhl T, Wessman S, Taylan F, Tesi B, Lagerstedt-Robinson K, Tettamanti G, Dukic M, Poluha A, Ljungman G, Nordgren A. Pediatric Soft Tissue Sarcoma in Limb-Girdle Muscular Dystrophy: Molecular Findings and Clinical Implications. *Am J Case Rep*. 2024 Dec 29;25:e945715. doi: 10.12659/AJCR.945715.

V. Maya-González C*, Tettamanti G*, Taylan F, Skarin Nordenvall A, Sejersen T, Nordgren A. Cancer Risk in Patients With Muscular Dystrophy and Myotonic Dystrophy: A Register-Based Cohort Study. *Neurology*. 2024 Oct 22;103(8):e209883. doi: 10.1212/WNL.0000000000209883.

Page 14. Related scientific papers

VII. Tesi B*, Robinson KL*, Abel F*, Díaz de Ståhl T, Orrsjö S, Poluha A, Hellberg M, Wessman S, Samuelsson S, Frisk T, Vogt H, Henning K, Sabel M, Ek T, Pal N, Nyman P, Giraud G, Wille J, Pronk CJ, Norén-Nyström U, Borssén M, Fili M, Stålhammar G, Herold N, Tettamanti G, Maya-Gonzalez C, Arvidsson L, Rosén A, Ekholm K, Kuchinskaya E, Hallbeck AL, Nordling M, Palmeback P, Kogner P, Smoler GK, Lähteenmäki P, Fransson S, Martinsson T, Shamik A, Mertens F, Rosenquist R, Wirta V, Tham E, Grillner P, Sandgren J, Ljungman G[#], Gisselsson D[#], Taylan F[#], Nordgren A[#]. Diagnostic yield and clinical impact of germline sequencing in children with CNS and extracranial solid tumors-a nationwide, prospective Swedish study. *Lancet Reg Health Eur*. 2024 Mar 19;39:100881. doi: 10.1016/j.lanepe.2024.100881.

Page 14. Additional scientific papers

VIII. Bakhuizen JJ, Bourdeaut F, Wadt KAW, Kratz CP, Jongmans MCJ, Waespe N, SIOPE Host Genome Working Group. Genetic testing for childhood cancer predisposition syndromes: Controversies and recommendations from the SIOPE Host Genome Working Group meeting 2022. *EJC Paediatric Oncology*. 2024 Dec;4:100176. doi: 10.1016/j.ejcped.2024.100176.

IX. Delgado-Vega AM*, Cederroth H*, Taylan F*, Ekholm K, Ek M, Thonberg H, Jemt A, Nilsson D, Eisefeldt J, Bilgrav Saether K, Höijer I, Akgun-Dogan O, Asano Y, Barakat TS, Batkovskye D, Baynam G, Bodamer O, Chetruengchai W, Corcoran P, Couse M, Danis D, Demidov G, Dohi E, Erhardsson M, Fernandez-Luna L, Fujiwara T, Garg N, Giugliani R, Gonzaga-Jauregui C, Grigelioniene G, Groza T, Gunnarsson C, Hammarsjö A, Hammond CK, Hatirnaz Ng Ö, Hesketh S, Hettiarachchi D, Johansson Soller M, Kirmani UA, Kjellberg M, Kvarnung M, Kvilidze O, Lagerstedt-Robinson K, Lasko P, Lassmann T, Lau LYS, Laurie S, Lim WK, Liu Z, Lysenkova Wiklander M, Makay P, Maiga AB, Maya-González C, Meyn MS, Neethiraj R, Nigro V, Nordgren F, Nordlund J, Orrsjö S, Ottosson J, Ozbek U, Özdemir Ö, Partin C, Pearce DA, Peck R, Pedersen A, Pettersson M, Pongpanich M, Posada de la Paz M, Ramani A, Romero JA, Romero VI, Rosenquist R, Saw AM, Spencer M, Stattin EL, Srichomthong C, Tapia-Paez I, Taruscio D, Taylor JP, Tkemaladze T, Tully I, Tümer Z, van Zelst-Stams WAG, Verloes A, Västerviga E, Wang S, Yang R, Yamamoto S, Yépez VA, Zhang Q, Shotelersuk V, Wiafe SA, Alanay Y, Botto LD, Kirmani S, Lumaka A, Palmer EE, Puri RD, Wirta V, Lindstrand A, Buske OJ[#], Cederroth M[#], Nordgren A[#]. Pushing the boundaries of rare disease diagnostics with the help of the first Undiagnosed Hackathon. *Nat Genet*. 2024 Nov;56(11):2287-2294. doi: 10.1038/s41588-024-01941-1.

* Shared first authors.

[#] Shared senior authors.

Corrected Figure 5:

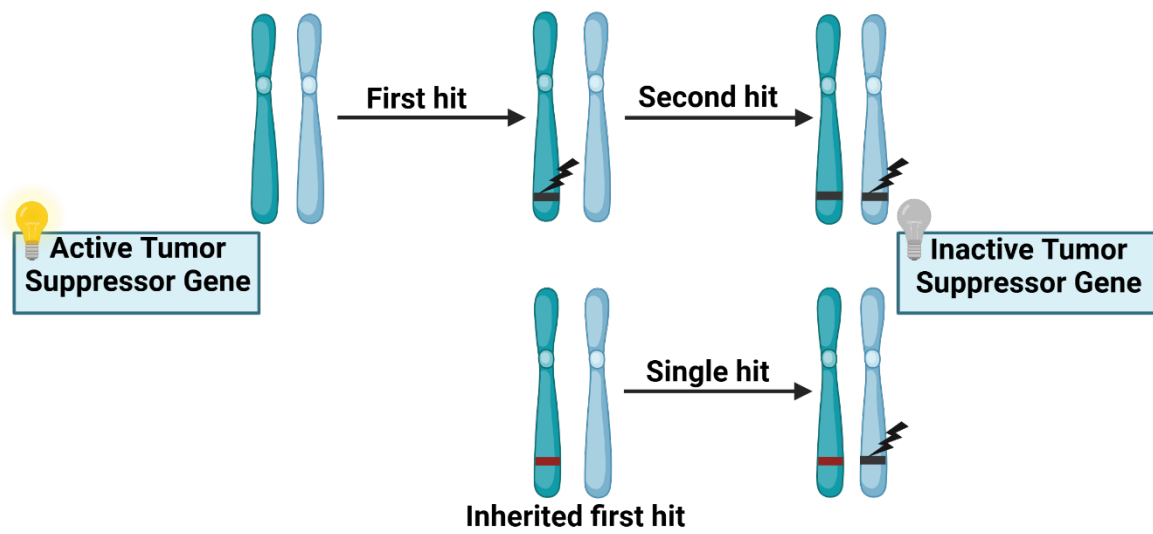


Figure created with *BioRender.com*.