**SUPPLEMENTARY MATERIAL**

*Table S1. Search strategy*

Search carried out in PubMed. Date of search November 2023

|  |  |  |
| --- | --- | --- |
| **Number** | **Search** | **Results** |
| 1 | Fetal cortical malformations\*.mp | 14628 |
| 2 | Limit 1 to 2013-2023 | 3701 |
| 3 | Fetal cortical anomalies\*.mp | 14628 |
| 4 | Limit 3 to 2013-2023 | 3701 |
| 5 | Cortical development malformations\*.mp | 143071 |
| 6 | Limit 5 to 2013-2023 | 62955 |
| 7 | 1 or 3 or 5 | 19880 |
| 8 | Limit 7 to 2013-2023 | 8861 |
| 9 | Fetal and cortical malformation | 2190 |
| 10 | Limit 9 to 2013-2023 | 782 |
| 11 | Malformations of Cortical Development/diagnosis[Mesh] | 4585 |
| 12 | Limit 11 to 2013-2023 | 2298 |
| 13 | 1 or 3 or 5 and polymicrogyria | 116 |
| 14 | Limit 13 to 2013-2023 | 67 |
| 15 | 1 or 3 or 5 and megalencephaly | 26 |
| 16 | Limit 15 to 2013-2023 | 20 |
| 17 | 1 or 3 or 5 and lissencephaly | 102 |
| 18 | Limit 17 to 2013-2023 | 42 |
| 19 | 1 or 3 or 5 and schizencephaly | 33 |
| 20 | Limit 19 to 2013-2023 | 17 |

*Table S2. Excluded studies and reason for exclusion*

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| --- | --- |
| **Study reference** | **Reason for exclusion** |
| Eda S, Terai T, Nishikawa Y, Tonari M, Kida T, Oku H, Sugasawa J, Shimakawa S, Hasegawa M, Ogihara T, Ikeda T. A Case of Hydranencephaly in Which Ophthalmic Examinations Were Performed. Case Rep Ophthalmol. 2016 Sep 16;7(3):142-147. doi: 10.1159/000449123. PMID: 27790130; PMCID: PMC5073785. | Did not include cases with fetal MCDs |
| Gandolfi Colleoni G, Contro E, Carletti A, Ghi T, Campobasso G, Rembouskos G, Volpe G, Pilu G, Volpe P. Prenatal diagnosis and outcome of fetal posterior fossa fluid collections. Ultrasound Obstet Gynecol. 2012 Jun;39(6):625-31. doi: 10.1002/uog.11071. Epub 2012 May 14. PMID: 22173885. | Did not include cases with fetal MCDs |
| Gelot AB, Represa A. Progression of Fetal Brain Lesions in Tuberous Sclerosis Complex. Front Neurosci. 2020 Aug 21;14:899. doi: 10.3389/fnins.2020.00899. PMID: 32973442; PMCID: PMC7472962. | Did not include details on CNS anomalies |
| Patek KJ, Kline-Fath BM, Hopkin RJ, Pilipenko VV, Crombleholme TM, Spaeth CG. Posterior fossa anomalies diagnosed with fetal MRI: associated anomalies and neurodevelopmental outcomes. Prenat Diagn. 2012 Jan;32(1):75-82. doi: 10.1002/pd.2911. PMID: 22367673. | Did not provide enough information about fetal MRI to assess if the MCDs were found prenatally |
| Sarno M, Sacramento GA, Khouri R, do Rosário MS, Costa F, Archanjo G, Santos LA, Nery N Jr, Vasilakis N, Ko AI, de Almeida AR. Zika Virus Infection and Stillbirths: A Case of Hydrops Fetalis, Hydranencephaly and Fetal Demise. PLoS Negl Trop Dis. 2016 Feb 25;10(2):e0004517. doi: 10.1371/journal.pntd.0004517. PMID: 26914330; PMCID: PMC4767410. | Did not include cases with fetal MCDs |
| Thorup E, Jensen LN, Bak GS, Ekelund CK, Greisen G, Jørgensen DS, Hellmuth SG, Wulff C, Petersen OB, Pedersen LH, Tabor A. Neurodevelopmental disorder in children believed to have isolated mild ventriculomegaly prenatally. Ultrasound Obstet Gynecol. 2019 Aug;54(2):182-189. doi: 10.1002/uog.20111. Epub 2019 Jun 26. PMID: 30168217. | Did not include cases with fetal MCDs |
| Wüest A, Surbek D, Wiest R, Weisstanner C, Bonel H, Steinlin M, Raio L, Tutschek B. Enlarged posterior fossa on prenatal imaging: differential diagnosis, associated anomalies and postnatal outcome. Acta Obstet Gynecol Scand. 2017 Jul;96(7):837-843. doi: 10.1111/aogs.13131. Epub 2017 Apr 10. PMID: 28295149. | Did not include cases with fetal MCDs |
| Braga VL, da Costa MDS, Riera R, Dos Santos Rocha LP, de Oliveira Santos BF, Matsumura Hondo TT, de Oliveira Chagas M, Cavalheiro S. Schizencephaly: A Review of 734 Patients. Pediatr Neurol. 2018 Oct;87:23-29. doi: 10.1016/j.pediatrneurol.2018.08.001. Epub 2018 Aug 8. PMID: 30501885. | Did not include cases with fetal MCDs |
| Çitli Ş, Serdaroglu E. Maternal Germline Mosaicism of a *de Novo* TUBB2B Mutation Leads to Complex Cortical Dysplasia in Two Siblings. Fetal Pediatr Pathol. 2022 Feb;41(1):155-165. doi: 10.1080/15513815.2020.1753270. Epub 2020 Apr 13. PMID: 32281916. | Did not include cases with fetal MCDs |
| Bhatnagar S, Kuber R, Shah D, Kulkarni V. Unilateral closed lip schizencephaly with septo-optic dysplasia. Ann Med Health Sci Res. 2014 Mar;4(2):283-5. doi: 10.4103/2141-9248.129065. PMID: 24761255; PMCID: PMC3991957. | Did not include cases with fetal MCDs |
| Candelo E, Sanz AM, Ramirez-Montaño D, Diaz-Ordoñez L, Granados AM, Rosso F, Nevado J, Lapunzina P, Pachajoa H. A Possible Association Between Zika Virus Infection and CDK5RAP2 Mutation. Front Genet. 2021 Mar 19;12:530028. doi: 10.3389/fgene.2021.530028. PMID: 33815457; PMCID: PMC8018576. | Did not include cases with fetal MCDs |
| Gutiérrez Sánchez LA, Sandoval Martínez DK, Díaz-Martínez LA, Becerra Mojica CH. Zika virus infection: A correlation between prenatal ultrasonographic and postmortem neuropathologic changes. Neuropathology. 2019 Dec;39(6):434-440. doi: 10.1111/neup.12603. Epub 2019 Nov 11. PMID: 31710135. | Did not include cases with fetal MCDs |
| Nagaraj UD, Hopkin R, Schapiro M, Kline-Fath B. Prenatal and postnatal evaluation of polymicrogyria with band heterotopia. Radiol Case Rep. 2017 May 27;12(3):602-605. doi: 10.1016/j.radcr.2017.04.007. PMID: 28828134; PMCID: PMC5551996. | Did not include at least 3 cases |
| Rosado Santos R, Rodrigues M, Loureiro T. Prenatal Diagnosis of Lissencephaly Associated with Biallelic Pathologic Variants in the COQ2 Gene. Acta Med Port. 2023 Jun 1;36(6):428-431. doi: 10.20344/amp.18606. Epub 2022 Sep 28. PMID: 36168972. | Did not include at least 3 cases |
| De Angelis C, Byrne AB, Morrow R, Feng J, Ha T, Wang P, Schreiber AW, Babic M, Taranath A, Manton N, King-Smith SL, Schwarz Q, Arts P, Scott HS, Barnett C. Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. BMC Med Genomics. 2021 Feb 27;14(1):64. doi: 10.1186/s12920-021-00911-4. PMID: 33639934; PMCID: PMC7916305. | Did not include at least 3 cases |
| Wojtowicz A, Duczkowska A, Huras H. A rare case of hemimegalencephaly diagnosed prenatally. Ginekol Pol. 2022;93(8):677-678. doi: 10.5603/GP.a2022.0030. Epub 2022 Jul 27. PMID: 35894497. | Did not include at least 3 cases |
| Inan C, Sayin NC, Gurkan H, Atli E, Gursoy Erzincan S, Uzun I, Sutcu H, Dogan S, Ikbal Atli E, Varol F. Schizencephaly accompanied by occipital encephalocele and deletion of chromosome 22q13.32: a case report. Fetal Pediatr Pathol. 2019 Dec;38(6):496-502. doi: 10.1080/15513815.2019.1604921. Epub 2019 May 26. PMID: 31130048. | Did not include at least 3 cases |
| Lanna MM, Fabbri E, Zavattoni M, Doneda C, Toto V, Izzo G, Casati D, Faiola S, Cetin I. Severe Fetal Symptomatic Infection from Human Cytomegalovirus following Nonprimary Maternal Infection: Report of Two Cases. Fetal Diagn Ther. 2022;49(1-2):36-40. doi: 10.1159/000521711. Epub 2021 Dec 30. PMID: 34969040. | Did not include at least 3 cases |
| Wang H, Li S, Li S, Jiang N, Guo J, Zhang W, Zhong M, Xie J. De Novo Mutated TUBB2B Associated Pachygyria Diagnosed by Medical Exome Sequencing and Long-Range PCR. Fetal Pediatr Pathol. 2019 Feb;38(1):63-71. doi: 10.1080/15513815.2018.1538273. Epub 2018 Dec 26. PMID: 30585108. | Did not include at least 3 cases |
| Nakamura Y, Okanishi T, Yamada H, Okazaki T, Hosoda C, Itai T, Miyatake S, Saitsu H, Matsumoto N, Maegaki Y. Progressive cerebral atrophies in three children with COL4A1 mutations. Brain Dev. 2021 Nov;43(10):1033-1038. doi: 10.1016/j.braindev.2021.06.008. Epub 2021 Jul 17. PMID: 34281745. | Did not include at least 3 cases |
| Karner E, Kasprian GJ, Farr A, Krampl-Bettelheim E. Polymicrogyria in a patient after twin-twin transfusion syndrome. BMJ Case Rep. 2023 Sep 22;16(9):e255510. doi: 10.1136/bcr-2023-255510. PMID: 37739446; PMCID: PMC10533711. | Did not include at least 3 cases |
| Su XR, Ma B, Zhang C, Li TG, Han BL, Wu WR, Nie F. Prenatal Ultrasound Diagnosis of Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus Syndrome with Persistent Hyperplastic Primary Vitreous: A Case Report. Fetal Diagn Ther. 2024;51(2):154-158. doi: 10.1159/000535509. Epub 2023 Nov 26. PMID: 38008077. | Did not include at least 3 cases |
| Angelidou A, Michael Z, Hotz A, Friedman K, Emani S, LaRovere K, Christou H. Is There More to Zika? Complex Cardiac Disease in a Case of Congenital Zika Syndrome. Neonatology. 2018;113(2):177-182. doi: 10.1159/000484656. Epub 2017 Dec 16. PMID: 29248924. | Did not include at least 3 cases |
| Cagneaux M, Paoli V, Blanchard G, Ville D, Guibaud L. Pre- and postnatal imaging of early cerebral damage in Sturge-Weber syndrome. Pediatr Radiol. 2013 Nov;43(11):1536-9. doi: 10.1007/s00247-013-2743-9. Epub 2013 Jul 23. PMID: 23877502. | Did not include at least 3 cases |
| Valence S, Poirier K, Lebrun N, Saillour Y, Sonigo P, Bessières B, Attié-Bitach T, Benachi A, Masson C, Encha-Razavi F, Chelly J, Bahi-Buisson N. Homozygous truncating mutation of the KBP gene, encoding a KIF1B-binding protein, in a familial case of fetal polymicrogyria. Neurogenetics. 2013 Nov;14(3-4):215-24. doi: 10.1007/s10048-013-0373-x. Epub 2013 Sep 27. PMID: 24072599. | Did not include at least 3 cases |
| Sepulveda W, Sepulveda F, Schonstedt V, Stern J, Diaz-Serani R. Neuroimaging Findings in Fetal Hemimegalencephaly: Case Study and Review. Fetal Diagn Ther. 2024;51(2):133-144. doi: 10.1159/000535406. Epub 2023 Nov 25. PMID: 38008087. | Did not include at least 3 cases |
| Harel T, Hacohen N, Shaag A, Gomori M, Singer A, Elpeleg O, Meiner V. Homozygous null variant in CRADD, encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. Am J Med Genet A. 2017 Sep;173(9):2539-2544. doi: 10.1002/ajmg.a.38347. Epub 2017 Jul 7. PMID: 28686357. | Did not include at least 3 cases |
| Štrafela P, Vizjak A, Mraz J, Mlakar J, Pižem J, Tul N, Županc TA, Popović M. Zika Virus-Associated Micrencephaly: A Thorough Description of Neuropathologic Findings in the Fetal Central Nervous System. Arch Pathol Lab Med. 2017 Jan;141(1):73-81. doi: 10.5858/arpa.2016-0341-SA. Epub 2016 Oct 11. PMID: 27726416. | Did not include at least 3 cases |
| Chartier S, Alby C, Boutaud L, Thomas S, Elkhartoufi N, Martinovic J, Kaplan J, Benachi A, Lacombe D, Sonigo P, Drunat S, Vekemans M, Agenor J, Encha Razavi F, Attie-Bitach T. A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. Birth Defects Res. 2018 Apr 17;110(7):598-602. doi: 10.1002/bdr2.1204. Epub 2018 Jan 22. PMID: 29356416. | Did not include at least 3 cases |
| Mishima T, Watari M, Iwaki Y, Nagai T, Kawamata-Nakamura M, Kobayashi Y, Fujieda S, Oikawa M, Takahashi N, Keira M, Yoshida H, Tonoki H. Miller-Dieker Syndrome with unbalanced translocation 45, X, psu dic(17;Y)(p13;p11.32) detected by fluorescence in situ hybridization and G-banding analysis using high resolution banding technique. Congenit Anom (Kyoto). 2017 Mar;57(2):61-63. doi: 10.1111/cga.12193. PMID: 27644460. | Did not include at least 3 cases |
| Pagnamenta AT, Howard MF, Wisniewski E, Popitsch N, Knight SJ, Keays DA, Quaghebeur G, Cox H, Cox P, Balla T, Taylor JC, Kini U. Germline recessive mutations in PI4KA are associated with perisylvian polymicrogyria, cerebellar hypoplasia and arthrogryposis. Hum Mol Genet. 2015 Jul 1;24(13):3732-41. doi: 10.1093/hmg/ddv117. Epub 2015 Apr 8. PMID: 25855803; PMCID: PMC4459391. | Did not include at least 3 cases |
| Izzo G, Toto V, Faiola S, Cattaneo E, Cavallari U, Passarini A, Gladin CR, Scelsa B, Parazzini C, Righini A. Cobblestone-like brain malformation with a new bi-allelic ADGRG1 (GPR-56) mutation: Fetal imaging-pathology correlation. J Neuroimaging. 2023 Jul-Aug;33(4):527-533. doi: 10.1111/jon.13130. Epub 2023 May 31. PMID: 37259271. | Did not include at least 3 cases |
| Itoh K, Pooh R, Shimokawa O, Fushiki S. Somatic mosaicism of the PI3K-AKT-MTOR pathway is associated with hemimegalencephaly in fetal brains. Neuropathology. 2023 Apr;43(2):190-196. doi: 10.1111/neup.12875. Epub 2022 Nov 3. PMID: 36325654. | Did not include at least 3 cases |
| Chapman T, Perez FA, Ishak GE, Doherty D. Prenatal diagnosis of Chudley-McCullough syndrome. Am J Med Genet A. 2016 Sep;170(9):2426-30. doi: 10.1002/ajmg.a.37806. Epub 2016 Jun 17. PMID: 27312216; PMCID: PMC8210655. | Did not include at least 3 cases |
| Laquerriere A, Maillard C, Cavallin M, Chapon F, Marguet F, Molin A, Sigaudy S, Blouet M, Benoist G, Fernandez C, Poirier K, Chelly J, Thomas S, Bahi-Buisson N. Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations. J Neuropathol Exp Neurol. 2017 Mar 1;76(3):195-205. doi: 10.1093/jnen/nlw124. PMID: 28395088. | Did not include at least 3 cases |
| De Keersmaecker B, Van Esch H, Van Schoubroeck D, Claus F, Moerman P, De Catte L. Prenatal diagnosis of MPPH syndrome. Prenat Diagn. 2013 Mar;33(3):292-5. doi: 10.1002/pd.4039. Epub 2013 Jan 24. PMID: 23348821. | Did not include at least 3 cases |
| Tonni G, Pattacini P, Bonasoni MP, Araujo Júnior E. Prenatal Diagnosis of Lissencephaly Type 2 using Three-dimensional Ultrasound and Fetal MRI: Case Report and Review of the Literature. Rev Bras Ginecol Obstet. 2016 Apr;38(4):201-6. doi: 10.1055/s-0036-1582126. Epub 2016 Apr 18. PMID: 27088705; PMCID: PMC10309364. | Did not include at least 3 cases |
| Kakish D, Tominna M, Krishnan A. Hemimegalencephaly: Evolution From an Atypical Focal Early Appearance on Fetal MRI to More Conventional MR Findings. Cureus. 2022 Aug 13;14(8):e27976. doi: 10.7759/cureus.27976. PMID: 36120272; PMCID: PMC9468183. | Did not include at least 3 cases |
| Poirier K, Martinovic J, Laquerrière A, Cavallin M, Fallet-Bianco C, Desguerre I, Valence S, Grande-Goburghun J, Francannet C, Deleuze JF, Boland A, Chelly J, Bahi-Buisson N. Rare ACTG1 variants in fetal microlissencephaly. Eur J Med Genet. 2015 Aug;58(8):416-8. doi: 10.1016/j.ejmg.2015.06.006. Epub 2015 Jul 16. PMID: 26188271. | Did not include at least 3 cases |
| Bruet S, Francannet C, Marguet F, Biard M, Sarret C, Laurichesse Delmas H. Prenatal diagnosis of hemimegalencephaly revealing tuberous sclerosis complex. Ultrasound Obstet Gynecol. 2020 May;55(5):688-689. doi: 10.1002/uog.21874. PMID: 31568608. | Did not include at least 3 cases |

*Table S3.* *Characteristics of liveborn cases with fetal malformation of cortical development (MCD)*

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Case**  | **GA at diagnosis (weeks)** | **Prenatal US findings** | **Prenatal MRI findings** | **Postnatal findings** | **Type of MCD** | **Neurodevelopmental outcome** | **Follow-up** |
| 138  | 31+3 | Cortical tubers | - | - | Complex | Normal | 9 months |
| 228  | 30 | Hypertelorism, suspected frontonasal dysplasia | Hypertelorism, suspected frontonasal dysplasia, PV focal gray matter heterotopia | Frontonasal dysplasia, hypertelorism, thinning of CC, multiple focal PV heterotopia, mild thinning of frontal cortex with abnormal imaging of white matter | Complex | Normal | 1 year |
| 328  | 34 | Severe bilateral VM, dilatated third ventricle | Severe bilateral VM, subacute frontal parenchymatous and intraventricular hemorrhage, dilatated third ventricle | Severe bilateral VM, intraventricular hemorrhage signs associated with frontoparietal parenchymatous lesion | Complex | Normal | 2 years |
| 438  | 32+6 | Cortical tubers, subependymal nodules | - | - | Complex | Normal | 2 years |
| 538 | 26+5 | Subependymal nodules | - | - | Complex | Normal | 7 years |
| 623  | 23 | Lateral ventricle asymmetry without VM, right deviation of midline and CSP, asymmetric frontal lobes, wide and dysmorphic left frontal horn, abnormal right Sylvian fissure | Dysmorphic right Sylvian fissure, enlarged left anterior horn, midline deviation, multiple shallow right posterior frontal sulci, asymmetric lateral ventricles, thin CC, small vermis | Likely *TUBB3* variant | Complex | Normal | 6 months |
| 742  | 26+6 | Lateral ventricular asymmetry, right deviation of midline and CSP, dysmorphic right Sylvian fissure, asymmetric left frontal lobes, wide dysmorphic left frontal horn | - | Microcephaly, plagiocephaly, dysgyria, *TUBB3* tubulinopathy | Complex | Normal | 6 months |
| 829 | 31+4 | Suspected cerebral abnormality | Bilateral GE enlargement, asymmetric overgrowth of one hemisphere, bilateral abnormal opercularization, perisylvianpolymicrogyria | Overlapping features of megalencephaly, capillary malformation and megalencephaly-polydactyly-polymicrogyria-hydrocephalus syndrome | Complex | Mild abnormality | - |
| 929 | 31 | Suspected cerebral abnormality | Bilateral GE enlargement | PV nodular heterotopia, persistent mega cisterna magna, mesial temporal sclerosis, *OPHN1* pathogenic mutation | Complex | Mild abnormality | - |
| 1035 | 30 | Schizencephaly | Right parietal and left temporal schizencephaly | - | Isolated | Mild abnormality | 8 months |
| 1138  | 34+6 | Cortical tubers | - | *TSC2* variant | Complex | Moderate abnormality | 20 months |
| 124 | 23+6 | Abnormal sulcation, ACC, asymmetric hemispheres | - | - | Complex | Moderate abnormality | 6 years |
| 1329  | 30+3 | Suspected cerebral abnormality | Unilateral GE enlargement, multiple small hemispheric mass lesions and subependymal nodules | MRI diagnostic for tuberous sclerosis, *TSC2* pathogenic copy-number variant | Complex | Moderate abnormality | - |
| 1429 | 25+2 | Suspected cerebral abnormality | Unilateral GE enlargement, bilateral VM | MRI diagnostic for tuberous sclerosis | Complex | Moderate abnormality | - |
| 1535 | 27 | No brain abnormality suspected | Right parietotemporal schizencephaly | - | Complex | Moderate abnormality | 6 months |
| 1645 | 29 | Enlarged cisterna magna, borderline VM, mild decrease of cerebral hemispheres | Bilateral GE enlargement, mild decrease in cerebral hemisphere biometry, unilateral borderline VM, ‘squared’ shape of frontal horns, mild reduction in cerebellar vermis biometry, enlarged cisterna magna | Enlarged cisterna magna, reduced cerebellar biometry, widened frontal horns | Complex | Moderate abnormality | 48 months |
| 1742 | 30+4 | Bilateral irregularity of frontal horns, underdeveloped Sylvian fissures | - | Polymicrogyria compatible with ischemic changes | Complex | Moderate abnormality | - |
| 1828  | 27 | Severe bilateral VM | Severe bilateral VM, bilateral opercular syndrome, frontoparietal polymicrogyria | Severe bilateral VM, bilateral opercular syndrome with associated cortical dysplasia, frontoparietal polymicrogyria | Complex | Severe abnormality | 6 months |
| 1929 | 25+1 | Suspected cerebral abnormality | Unilateral GE enlargement hemorrhage, asymmetric overgrowth of onehemisphere | Right hemimegalencephaly | Complex | Severe abnormality | 8 months |
| 2029  | 22+2 | Suspected cerebral abnormality | Bilateral GE enlargement, ACC, bilateral abnormality of opercularization, hemispheric dysgyria, small and asymmetric pons, dilatated Blake's pouch with vermis rotation and hypoplastic vermis | ACC, enlarged basal ganglia with absent anterior limb internal capsule, bilateral abnormal opercularization and extensive dysgyria, brainstem dysplasia with small and asymmetric pons, vermis upwardly rotated by persistent Blake's pouch, hypoplasia of vermis and cerebellar hemispheres, *TUBA1A* pathogenic variant | Complex | Severe abnormality | 22 months |
| 2129  | 23+3 | Suspected cerebral abnormality | Bilateral GE cavitation, ACC, bilateral VM, cerebellar hypoplasia | ACC, severe loss of PV white matter, *PDHA1* pathogenic variant | Complex | Severe abnormality | 6 years |
| 2229  | 22+3 | Suspected cerebral abnormality | Bilateral GE enlargement and cavitation, bilateral abnormal opercularization, perisylvian polymicrogyria | Right frontal polymicrogyria, abnormal orientation of Sylvian fissures, bilateral subependymal pseudocysts | Complex | Severe abnormality | - |
| 2329  | 33 | Suspected cerebral abnormality | Bilateral GE enlargement, underopercularization, underdeveloped primary and secondary sulcation, abnormal persistence of laminated appearance, bilateral VM, cerebellar hypoplasia | Lissencephaly, enlarged basal ganglia, absent anterior limb of internal capsule, cerebellar hypoplasia, *TUBA1A* pathogenic variant | Complex | Severe abnormality | - |
| 2447  | - | Cystic tumor | - | - | Complex | Severe abnormality | - |
| 2535 | 28 | No brain abnormality suspected | Bilateral temporoparietal schizencephaly | - | Complex | Severe abnormality | 13 months |
| 2635 | 38 | VM, mega cisterna magna | Left occipital and temporo-occipital schizencephaly | - | Complex | Severe abnormality | 8 months |
| 2736  | 24 | Unilateral microphthalmia, delayed cortical sulcation, mild VM with irregular ventricular wall, dilatated third ventricle, dysmorphic CC, Z-shaped brainstem, small dysplastic cerebellar hemispheres, tectocerebellar dysraphia | - | Narrow and anteriorly blurred interhemispheric fissure, thick and flat cortex with irregular pebbled cortical white matter border, PV heterotopia, subependymal hemorrages and calcifications, deformed basal ganglia and thalami, immature and hypomyelinated white matter, enlarged third ventricle, thin CC, severe brainstem kinks, enlarged tectum, small occipital skull defect, small vermian remnant, dysmorphic and small cerebellar hemispheres, small right eye; diagnosis of Walker–Warburg syndrome associated with tectocerebellar dysraphia | Complex | Severe abnormality | 3 years |
| 2842 | 26 | Delayed cortical sulcation, shallow Sylvian fissures, mild VM, irregular ventricular wall, dysgenesis of CC, Z-shaped brainstem, small dysplastic cerebellar hemispheres, occipital encephalocele, vermian agenesis, elongated and thick tectum, unilateral microphthalmia | - | Cobblestone malformation, homozygous *DAG1* frameshift mutation; diagnosis of Walker–Warburg syndrome | Complex | Severe abnormality | 3 years |
| 2942 | 26+6 | Mild VM, parenchymal calcifications, small Sylvian fissures, enlarged perisylvian subarachnoid space, irregular cortex, delayed sulcation, small CC | - | Microcephaly, brain atrophy, homozygous *FLVCR2* frameshift mutation; diagnosis of Fowler syndrome. | Complex | Severe abnormality | - |
| 3042 | 24+6 | Malformed shallow Sylvian fissures, small frontal lobes, irregular cortical contour, enlarged subarachnoid space | - | Diffuse polymicrogyria | Isolated | Severe abnormality | - |
| 3129 | 21+1 | Suspected cerebral abnormality | Bilateral GE enlargement, no significant opercularization, thin hemispheric parenchyma, severe bilateral lateral VM, hypoplastic cerebellum, ACC, small pons | *TUBA1A* pathogenic variant | Complex | Neonatal death | 1 day |
| 3229 | 24+1 | Suspected cerebral abnormality | Bilateral GE enlargement and cavitation, marked underopercularization, parieto-occipital sulcus and cingulate sulcus not seen, severe lateral VM | Walker–Warburg phenotype with cobblestone lissencephaly | Complex | Infant death | 2 months |
| 3329  | 22+4 | Suspected cerebral abnormality | Bilateral GE enlargement and cavitation, severe VM and underopercularization, hypoplastic cerebellum, ACC | Walker–Warburg phenotype with cobblestone lissencephaly | Complex | Infant death | 4 months |
| 3443  | - | MCD, microcephaly, cerebellar hypoplasia, hypoplastic left heart | - | MCD, hypoplastic left heart syndrome, terminal deletion of 6q26-qter, interstitial duplication of 6q24.2-q26 | Complex | Infant death | - |
| 3545 | 22 | Vermian hypoplasia, enlarged cisterna magna | Unilateral GE enlargement, small vermis | - | Complex | Infant death due to cardiac abnormality | 2 months |
| 3627 | - | Obliterated CSP | Obliterated CSP, polymicrogyria of left parietal lobe | - | Complex | Neonatal death | - |
| 3724 | - | Microcephaly, polymicrogyria | Abnormal gyration | *ATP2A2* homozygous truncating variant | Complex | Neonatal death | 1 h |
| 3824 | - | Microcephaly, polymicrogyria | Abnormal gyration, undulated and mildly thickened cortical ribbon | Microcephaly, polymicrogyria, meningeal arterial calcifications, necrotic areas in basal ganglia, dentato-olivary dysplasia, hypoplasia of pyramidal tracts, fetal akinesia sequence, *ATP2A2* homozygous truncating variant | Complex | Neonatal death | 30 min |

ACC: agenesis of corpus callosum; CC: corpus callosum; CSP: cavum septum pellucidum; GE: ganglionic eminence; MRI: magnetic resonance imaging; PV: periventricular; VM, ventriculomegaly.