MRI对胎儿小头畸形与小头畸形简单脑回模式的诊断价值

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【摘要】 **目的** 通过与超声对比,评价MRI对胎儿小头畸形及小头畸形简单脑回模式(microcephaly with simplified gyral pattern,MSGP)的诊断价值。**方法** 回顾性研究复旦大学附属妇产科医院2023年9月至2025年2月24例胎儿小头畸形及MSGP的产前MRI表现,并与同一天的胎儿超声检查结果对比,所有病例均经后续随访及引产后证实。**结果** 24例胎儿的孕周为23.6~36.6周,平均孕周为27.7周,产前MRI诊断胎儿小头畸形14例,MSGP 10例。MSGP中1例伴有Dandy-Walker变异型,2例伴有胼胝体发育不全。产前超声诊断胎儿小头畸形21例,其中1例伴Dandy-Walker变异型,MSGP 3例,漏诊7例MSGP,漏诊2例伴发胼胝体发育不全。胎儿小头畸形的MRI及超声表现为头围小于均值3个标准差(-3SD),不伴有脑发育异常;MSGP的MRI表现为头围小于均值-3SD,颅面比例小,额叶小而倾斜、后缩,大脑沟回浅、少,比正常孕周脑沟回发育延迟2~4周,皮质厚度正常,大脑灰白质正常,MRI对产前MSGP的诊断率为100%。MSGP的超声表现为头围小于均值-3SD,大脑沟回浅、少,超声对产前MSGP的诊断率为30%。**结论** MRI对脑沟回的评估及皮质的显示较超声更有优势,对胎儿小头畸形及MSGP均能明确诊断,并对颅内其他伴发畸形有更高的诊断准确率。对于临床怀疑胎儿小头畸形的胎儿,须行胎脑MRI评估,以确定是否为MSGP及发现可能的颅内伴发畸形。

【关键词】 胎儿小头畸形; 小头畸形简单脑回模式(MSGP); 产前MRI; 产前诊断
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Diagnostic value of MRI in fetal microcephaly and microcephaly with simplified gyral pattern

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【**Abstract**】 **Objective** To evaluate the diagnostic value of MRI in fetal microcephaly and microcephaly with simplified gyral pattern (MSGP) by comparing with ultrasonography. **Methods** Prenatal MRI manifestations of 24 cases of fetal microcephaly and MSGP in Obstetrics and Gynecology Hospital, Fudan University from Sept 2023 to Feb 2025 were retrospectively studied and compared with fetal ultrasonography findings on the same day. All cases were confirmed by follow-up and induced labor. **Results** Twenty-four fetuses were 23.6 to 36.6 gestational weeks with a average of 27.7 gestational weeks. Fourteen cases of fetal microcephaly and 10 cases of MSGP were diagnosed by prenatal MRI. Among those diagnosed with MSGP, 1 case was accompanied by Dandy-Walker variant, and 2 cases were accompanied by Dandy corpus callosum agenesis. Prenatal ultrasound diagnosed microcephaly in 21 cases, among which 1 case was accompanied by Dandy-Walker variant, and MSGP in 3 cases. Seven cases of MSGP and 2 cases of concomitant corpus callosum agenesis were missed diagnosed. MRI and ultrasound

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findings of fetal microcephaly showed a head circumference less than three standard deviations (-3SD) from the mean value. MRI findings of MSGP were head circumference less than -3SD from the mean value, decreased cranio facial ratio, slanted and small frontal lobe, fewer and shallow cerebral sulci and gyri, the development of sulci and gyri is delayed by about 2 to 4 weeks compared to the normal gestational age, but with normal thickness of the cortex, and normal cerebral gray and white matter. The diagnostic rate of MRI for prenatal MSGP was 100%. Ultrasound findings of MSGP were head circumference less than -3SD from the mean value, and cerebral sulci and gyri were shallow and few. The diagnostic rate of ultrasonography for prenatal MSGP was 30%. **Conclusion** MRI is more advantageous than ultrasonography in the evaluation of sulci and gyri, as well as the display of cortex, and can make a definite diagnosis for fetal microcephaly and MSGP, also has a higher diagnostic accuracy for other intracranial concomitant malformations. For fetuses with clinically suspected microcephaly, fetal brain MRI assessment is required to determine whether it is MSGP, or to detect possible intracranial concomitant malformations.

【Key words】 fetal microcephaly; microcephaly with simplified gyral pattern (MSGP); prenatal MRI; prenatal diagnosis

胎儿小头畸型是一种罕见但重要的临床发现,可能作为一系列疾病的一部分,也可能是一种孤立性发现,还可能少数为正常变异[1]。头围越小,发育和智力迟缓的风险越高[1-2]。近年来文献报道提出新的概念:小头畸形简单脑回模式(microcephaly with simplified gyral pattern,MSGP),其特征是小头畸形伴脑沟回数量减少、变浅,无皮质异常[3]。与胎儿小头畸形相比,MSGP有更严重的智力障碍、神经发育迟缓及其他中枢神经系统畸形[4]。因此,在产前诊断及鉴别胎儿小头畸形及MSGP,对产科及孕妇决定妊娠结局至关重要。目前文献对胎儿小头畸形的报道多注重于头围的测量及脑体积的评估,对脑沟回发育的评估较少,以脑沟回异常为诊断依据的产前MSGP的报道也较少。本文着重研究胎儿小头畸形及MSGP的产前MRI表现,并与超声对比,评估MRI对胎儿小头畸形及MSGP的诊断价值,并总结各孕周胎儿脑沟回发育的规律及MSGP的诊断要点。

资料和方法

研究对象 2023年9月复旦大学附属妇产科医院产科与放射科、超声科建立胎儿小头畸形的专项研究,对超声做出小头畸形诊断的胎儿当天行胎脑MRI检查,以保证超声及MRI在相同的胎龄时间内测量胎儿头围及评估脑沟回。收集复旦大学附属妇产科医院2023年9月至2025年2月24例胎儿小头畸形及MSGP的MRI影像资料,其中14例胎儿小头畸形,10例MSGP。24名孕妇,22~42岁,平均31.3岁,均为单胎,孕周23.6~36.6周,平均孕周27.7周。本研究经复旦大学附属妇产科医院伦理委员会同意(批准号2025-010),所有孕妇均阅读并签署知情同意书。

影像学检查及图像采集 孕妇仰卧位或侧卧位。先行孕妇中下腹及盆腔定位扫描,再行胎儿颅脑轴位、矢状位、冠状位T2WI、T1WI及DWI扫描。每一个序列的定位像均以前一个序列为准[5]。MRI有伪影者重复扫描直至影像质量合格(飞利浦3.0T MRI成像仪,16通道相控阵体线圈)。T2WI采用单激发快速自旋回波序列(single shot turbo spin echo,ssh-TSE)及快速平衡式自由稳态进动序列(balanced turbo field echo,BTFE),T1WI采用超快速场回波序列(turbo field echo,TFE),扫描参数如下。ssh-TSE序列:TR 10 000 ms,TE 100 ms,层厚4 mm,层间隔0 mm,反转角90°,视野308 mm×399 mm,矩阵280×275,扫描时间30 s;BTFE序列:TR 2.7 ms,TE 1.36 ms,层厚4 mm,层间隔-2 mm,反转角80°,视野383 mm×387 mm,矩阵256×261,扫描时间63 s;TFE序列:TR 10 ms,TE 2.3 ms,层厚4 mm,层间隔0 mm,反转角15°,视野375 mm×305 mm,矩阵236×142,扫描时间40 s;DWI序列:TR 3 000 ms,TE 53 ms,层厚4 mm,层间隔0 mm,反转角90°,视野360 mm×306 mm,矩阵180×127,扫描时间45 s。控制吸收率(specific

absorption rates，SAR）在 1.5 W/kg 以下[6]。所有MRI图像清晰显示胎脑的轴位、矢状位及冠状位。

数据分析及诊断标准 MRI图像由2名具有10年以上产前诊断经验的放射科副主任医师分析诊断，并经1名10年以上产前诊断经验的放射科主任医师复核；超声图像采集及诊断由2名具有10年以上产前诊断经验的超声科副主任或主任医师完成，以排除因经验不足而导致的误诊或漏诊。在轴位丘脑横切面测量颅骨外缘周长，取3次测量平均值，得到头围数据。头围数据以NICHD亚裔人群为标准对比[胎儿生长受限（fetal growth restriction，FGR）共识推荐]。

结　　果

诊断结果 24例胎儿中，MRI诊断为胎儿小头畸形14例，MSGP10例，MSGP占41.67%，10例MSCP中1例伴有Dandy-Walker变异型，2例伴有胼胝体发育不全，3例伴有心脏异常，2例羊水穿刺提示有小头畸形相关遗传学异常，3例存在FGR。14例胎儿小头畸形中12例存在FGR，2例孕妇多发巨大子宫肌瘤。超声诊断胎儿小头畸形21例，MSGP 3例，漏诊7例MSGP，漏诊2例伴发的胼胝体发育不全。通过头围测量、MRI及超声均能对胎儿小头畸形做出准确诊断，但超声对MSGP及颅内伴发畸形的漏诊率较高，MSGP的MRI诊断率为100%，超声诊断率为30%。

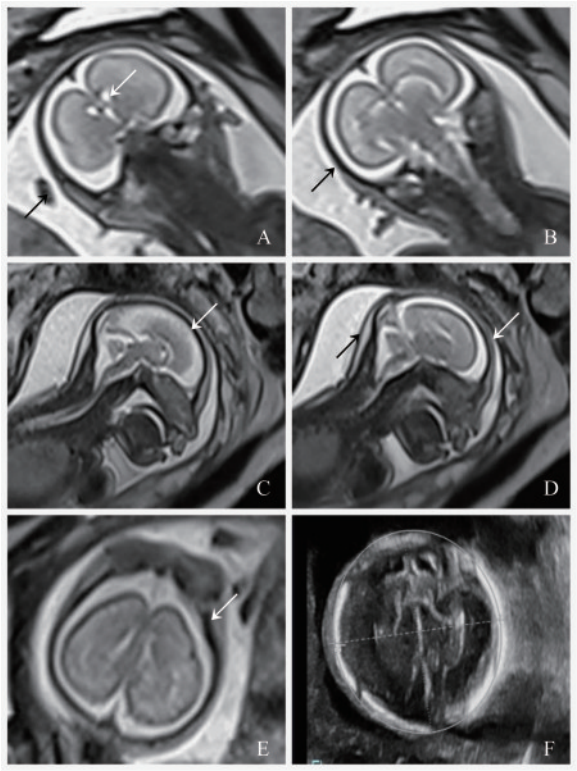
随访结果 本组病例均经后续随访及引产后证实。10例MSGP均回当地或由我院引产（经伦理委员会批准），引产后头围均小于均值3个标准差（-3SD），颅面比例小。14例胎儿小头畸形中2例因其他异常引产，引产后头围小于均值-3SD，12例出生后随访至2025年6月，出生后6例头围在正常范围（0，-2SD）内，6例出生后头围小于均值-3SD，头颅影像学检查均无脑沟回发育异常及其他颅内结构异常。

胎儿小头畸形的影像学表现 MRI及超声均表现为头围小于均值-3SD，MRI显示脑发育无异常；14例胎儿小头畸形中，12例超声提示腹围及体重小于均值10%（85.71%），存在FGR，2例MRI提示子宫巨大肌瘤压迫胎儿头颅。

MSGP的影像学表现 MRI表现为头围小于均值-3SD，脑组织小、脑沟回发育异常，皮质厚度及灰白质无异常。脑组织小表现为颅面比例小，额叶小而后缩、倾斜，脑外间隙增宽，小脑多不受影响，其小脑横径多数正常。脑沟回发育异常表现为：孕25周以下外侧裂、顶枕沟、距状沟不显示或浅、平（图1）；孕25～29周中央沟、中央前沟、中央后沟不显示或浅（图2）；孕29周以上表现为额颞叶的脑沟回浅、少（图3、图4）。MSGP存在一定程度的脑沟发育，但脑沟回的发育不符合孕周，一般较正常孕周脑沟回发育推迟2～4周，且脑沟深度变浅、数量减少，皮质厚度正常，与脑沟回完全不发育的平滑脑及脑回增厚、增多的巨脑回、多小脑回不同。超声诊断的3例MSGP表现为头围小于均值-3SD，外侧裂、顶枕沟、距状沟浅，或大脑皮层沟回少，孕周为29.5～36.6周；6例孕29周以下胎儿MSGP超声未发现脑沟回异常；1例孕31.4周胎儿MSGP伴Dandy-Walker变异型，超声提示胎儿小头畸形伴Dandy-Waler变异型，未发现脑沟回异常；2例孕28周以上胎儿MSGP伴胼胝体发育不全，超声未发现胼胝体异常；3例MSGP超声提示心脏异常；3例超声提示腹围及体重小于均值10%（30%），存在FGR。

讨　　论

胎儿小头畸形及MSGP的概念及诊断 胎儿小头畸形是一个临床测量值，不是以头颅形态结构异常得出的，而是由生物学统计数据得出，其诊断依赖于超声测量的胎儿头围[7]。1984年Chervenak等[8]首先定义了胎儿小头畸形的诊断标准，即头围小于胎龄均值-3SD，加拿大妇产科医师协会（SOGC）及美国母胎医学会（SMFM）均采用此标准，并将头围小于胎龄均值-5SD定义为病理性小头畸形[9-10]。胎儿小头的病因有外源性或内源性，外源性包括宫内生长限制、宫内感染、药物暴露、母体因素等，内源性多为遗传性[1]，又称为真性小头症，主要与细胞增殖减少或过度凋亡相关[4,7]。一些胎儿小头畸形表现为简化脑回模式，即MSGP[4,11]，其特征是脑容量减少（额面比例小，额叶小而后缩、倾斜）和简单化脑回（脑沟回数量减少、深度变浅），皮质厚度正常，与生发基质中神经元细胞数量减少有关。与不伴有脑发育异常的胎儿小头畸形相比，



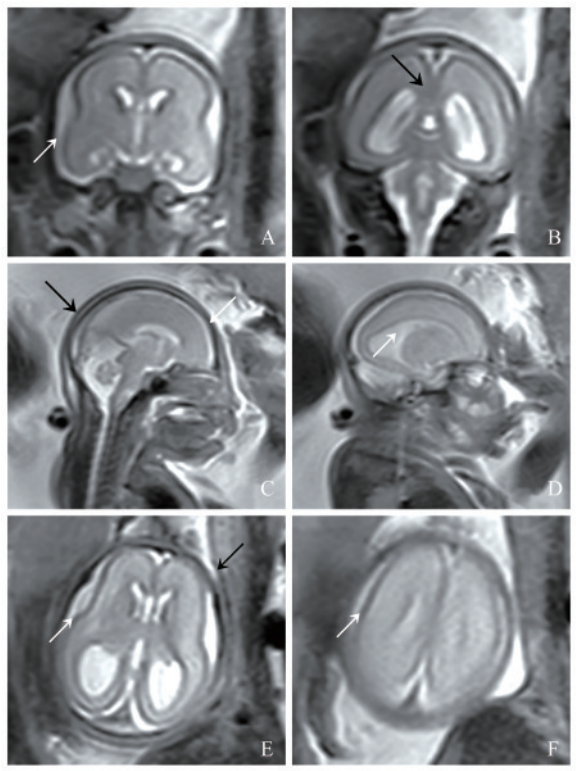
A-B: coronal T2WI, demonstrating that sylvian fissure is not shown (black arrow), and calcarine sulcus is not shown (white arrow). C-D: Sagittal T2WI, showing decreased cranio facial ratio, slanted and small frontal lobe (white arrow) and shallow parieto-occipital fissure (black arrow), with otherwise normal brain and posterior fossa. E: Axial T2WI, showing small frontal lobe (white arrow). F: An axial image of ultrasonography, showing that the cerebrum is small.

图1 孕23.6周MSGP的MRI及超声图像

Fig 1 MRI and ultrasound images of MSGP at 23.6 gestational weeks

MSGP有更严重的智力障碍、神经发育迟缓及其他中枢神经系统畸形[4]。本研究纳入的24例头围减小的胎儿病例,10例经MRI诊断为MSGP,占比41.67%,这意味着MSGP在头围减小的胎儿中占一定比例。

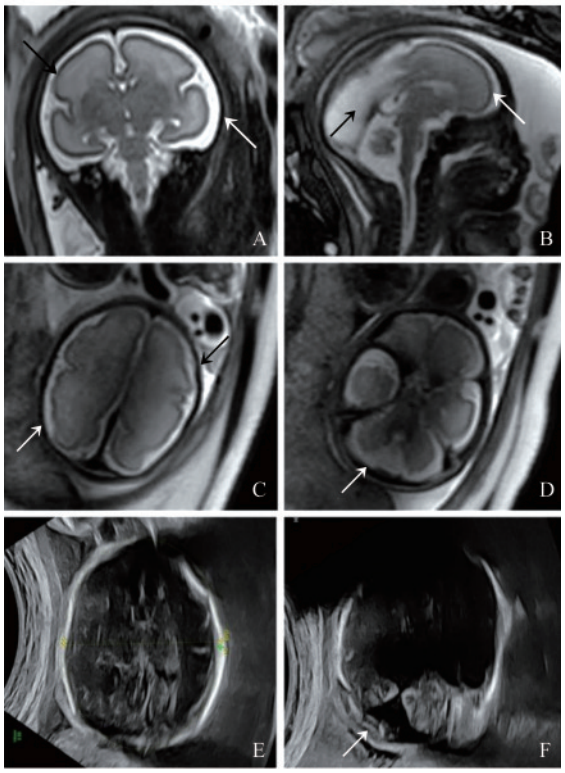
本研究中MSGP均表现为头围减小、额面比例小,额叶小而后缩、倾斜,脑沟回浅、少、延迟于孕周,但小脑横径与孕周基本相符,表明其影响主要为大脑尤其是额叶,这与文献[12-14]报道一致。Clouchoux等[12]发现胎儿小脑在体积上比大脑其他区域具有更高的成熟率,脑容积减小主要影响大脑,Chen等[13]和Polat等[14]观察到胎儿大脑体积减小最常影响的区域是额叶。本研究10例MSGP中有2例(20%)胼胝体发育不全,1例(10%)Dandy-Walker畸形,这意味着MSGP与胼胝体发育不全、小脑或桥小脑发育不全相关,与文献[15-17]报道一致,Vermeulen等[15]报道58%的MSGP伴有胼胝体部分或完全发育不全,Cabet等[11]报道50%的MSGP伴有胼胝体发育不全,12.5%伴有桥小脑发育不全,而这类伴发颅内其他畸形的MSGP预后更差。本研究10例MSGP中,2例胎儿羊水穿刺检出与小头畸形相关的遗传学异常,3例伴有颅内其他畸形,3例伴有心脏异常,这意味着MSGP伴有更多的颅内畸形、其他系统畸形及遗传学异常,提示与遗传相关性更高,更倾向于内源性疾病,预后更差,这与Okafor等[4]的研究结果相同。Cabet等[11]报道了



A: Coronal T2WI, showing shallow sylvian fissure without formation of an acute angle with the adjacent temporal lobe (white arrow). B: Coronal T2WI, and calcarinesulcus is not shown (black arrow). C: Sagittal T2WI, showing decreased cranio facial ratio, slanted and small frontal lobe (white arrow), and the parieto-occipital sulcus is not shown (black arrow), with otherwise normal brain and posterior fossa. D: Sagittal T2WI, showing no evidence of central sulcus (white arrow). E: Axial T2WI, showing small frontal lobe (black arrow), shallow sylvian fissure without formation of an acute angle with the adjacent temporal lobe (white arrow). F: Axial T2WI, and the precentral sulcus is not shown.

图2 孕25.3周MSGP的MRI图像

Fig 2 MRI images of MSGP at 25.3 gestational weeks



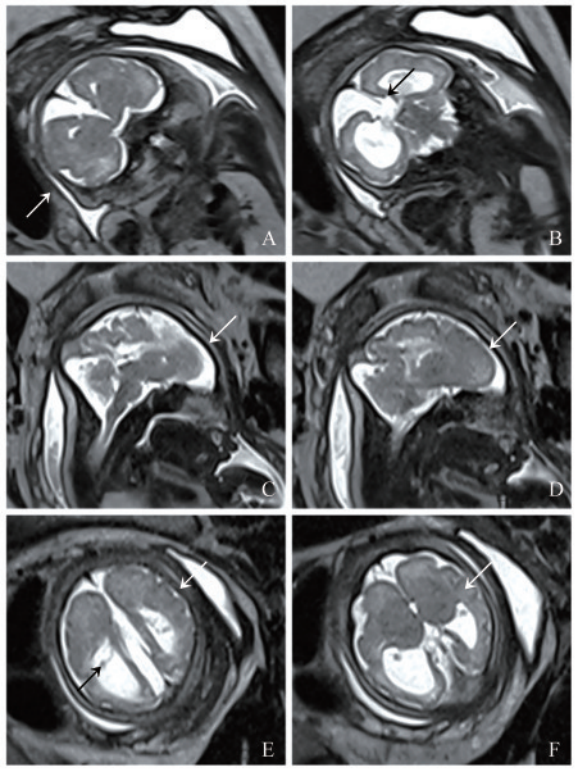
A：Coronal T2WI, showing that the superior frontal sulcus and inferior frontal sulcus are not displayed (black arrow), and the superior temporal sulcus and inferior temporal sulcus are not displayed (white arrow). B：Sagittal T2WI, showing decreased cranio facial ratio, slanted and small frontal lobe (white arrow), and no evidence of central sulcus (black arrow). C：Axial T2WI, showing shallow precentral sulcus (black arrow) and postcentral sulcus (white arrow), and fewer and more shallow frontal parietal sulci gyri. D：Axial T2WI, showing fissure-like communicating fourth ventricle with posterior cranial fossa (white arrow). E：Axial ultrasound imaging, the measured head circumference is small, and sulci and gyri display unclear. F：Axial ultrasound imaging, showing fissure-like communicating fourth ventricle with posterior cranial fossa.

图3 孕31.4周MSGP的MRI及超声图像

Fig 3 MRI and ultrasound images of MSGP at 31.4 gestational weeks

37.5%的MSGP存在FGR，本研究中30%的MSGP存在FGR(3/10)，这意味着MSGP与FGR的相关性较低。

本研究中胎儿小头畸形的脑沟回发育与孕周相符合，不伴有额叶缩小，12例腹围减小，存在FGR(85.71%)，2例子宫巨大肌瘤压迫胎儿头颅，这意味着不伴有脑发育异常的胎儿小头畸形多为外源性因素(如FGR或子宫肌瘤压迫)引起，与FGR的相关性更高，这与文献[13-14]报道一致。Chervenak等[8]



A-B：Coronal T2WI, showing fewer and shallow frontal and temporal sulci (white arrow), and absence of corpus callosum (black arrow). C-D：Sagittal T2WI, showing decreased cranio facial ratio, slanted and small frontal lobe, fewer and shallow frontal and parietal sulci (white arrow). E-F：Axial T2WI, showing small frontal lobe, fewer and shallow sulci gyri of frontal, temporal and occipital lobe (white arrow), and absence of corpus callosum (black arrow).

图4 孕36.6周MSGP的MRI图像

Fig 4 MRI images of MSGP at 36.6 gestational weeks

报道了16例胎儿小头畸形，其中9例(56.23%)出生后仍为小头畸形(头围小于-3SD)，7例(43.75%)为正常范围头围(头围大于-2SD)，本研究中14例胎儿小头畸形，57.14%引产及出生后仍为小头畸形(2例引产，6例出生)，42.86%(6例)在后续的随访及出生后，头围逐渐追赶至正常范围，且出生后颅面比例正常，脑内结构无异常，这与Chervenak等[8]的研究结果相似。Leibovitz等[7]认为胎儿小头畸形常常是小于胎龄儿的一个表现，这意味着不伴有脑发育异常的胎儿小头畸形有一定概率出生后预后良好。

胎儿各孕周脑沟回出现的意义 孕中期胎儿脑沟开始发展，最具代表性的是外侧裂，是大脑正常发育的重要标志[18]。孕18～19.6周显示顶枕沟、外侧裂，孕20～21.6周显示距状沟[19]。孕24～25.6

周显示扣带沟,孕24～25.6周显示中央沟及额上沟,孕26～27.6周显示颞上沟、中央前沟及中央后沟,孕29周显示额下沟及颞枕沟,孕30周显示颞下沟[20-21]。本研究MRI最早诊断MSGP的孕周为23.6周,3例孕25周以下MSGP的MRI表现为外侧裂、距状沟、顶枕沟未发育或浅、平,额叶小而后缩,3例孕25～28.6周MSGP的MRI表现为中央沟、中央前沟、中央后沟未发育或浅,额叶小而后缩,这6例MSGP超声未能发现脑沟回异常,1例未发现伴发的胼胝体发育不全,证明MRI在孕29周之前诊断MSGP具有优势,可较超声更早期、更准确地诊断MSGP,并发现其他颅内伴发畸形。孕25周之前胎脑MRI应着重观察外侧裂、顶枕沟及距状沟的形态,孕25～28.6周之前胎脑MRI应着重观察中央沟、中央前沟及中央后沟。4例孕29周以上MSGP MRI表现为额上沟、额下沟、颞上沟及颞下沟的未发育或浅,超声1例未发现伴发的胼胝体发育不全,1例未发现脑沟回异常,证明MRI对大脑沟回的定位较超声更准确、细致,孕29周之后对脑沟回的观察不应再局限于外侧裂及顶枕沟,而应当着重观察额颞叶二三级脑沟回的发育。胎儿小头畸形容易诊断,但MSGP容易漏诊,需要影像技术上对各个孕期的脑沟回的精准显示,更需要诊断医师熟悉胎儿各孕周脑沟回出现的时间。

胎儿小头畸形及MSGP产前MRI诊断的优势 胎儿小头畸形表现为头围小于均值-3SD,但超声测量头围是对胎儿颅骨生物测量的评估,无法评估脑容量,对脑沟回及皮质的显示亦有局限性。Tanabe等[22]认为使用产前超声诊断胎儿小头畸形仍然具有挑战性,产前超声排除小头畸形比检测小头畸形更准确。胎脑MRI对脑容量的评估、脑沟回及皮质的显示优于超声[6],被广泛认为是检测胎儿脑沟回异常的最佳方法[23]。Cabet等[11]回顾性研究8名超声观察到胎儿小头畸形后被转诊的患者,均无任何脑外异常,胎脑MRI证实在所有病例中,胎儿脑回均为简单化表现,皮质厚度正常,白质信号正常,排除了迁移/皮质生成疾病(无脑畸形/多小脑回),提示MSGP,这意味着在头围减小的胎儿中MSGP发生率高。本研究中24例头围减小的胎儿,10例为MSGP,占比41.67%,超声漏诊7例,而MRI对10例MSGP均明确诊断。此外,MRI对颅内多发畸形及伴发畸形的诊断较超声更准确,本研究中,1例胎儿MSGP伴Dandy-Walker变异型,超声未发现脑沟回异常,2例胎儿MSGP伴胼胝体发育不全,超声未发现胼胝体发育不全。Pei等[23]研究中,对超声诊断小头畸形的胎儿行胎脑MRI检查,均发现额外信息,包括脑白质发育不良、脑沟回发育异常等;von der Hagen等[24]认为除头围小外,76%的小头畸形患儿通过胎脑MRI检测到其他异常,包括白质异常、胼胝体异常、幕下病变和皮质回转异常。MRI可以克服超声的局限性并检测到额外的脑结构异常,是识别小头畸形相关异常更敏感的影像学检查方式,已成为表征正常和异常大脑发育的首选成像方式[21]。因此,当怀疑胎儿小头畸形时,需转诊进行胎脑MRI,以获取更多大脑发育的信息,从而确定是否存在MSGP,以及发现可能的颅内伴发畸形[25],这对产科决策和妊娠结局起到关键作用。

本研究尚有不足之处:由于病例均为产前超声筛查提示胎儿小头畸形后进一步行MRI检查,因此未能对更早期的病例进行研究,也未能在正常胎儿筛查中发现MSGP;本研究病例数较少,对MSGP与颅内其他伴发畸形的关系尚需要进一步研究。

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利益冲突声明 所有作者均声明不存在利益冲突。

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