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THE ROLE OF MAGNETIC RESONANCE IMAGING IN THE PRENATAL DIAGNOSIS OF CLEFT LIP AND PALATE

ROLA REZONANSU MAGNETYCZNEGO W PRENATALNYM ROZPOZNANIU WADY ROZSZCZEPOWEJ WARGI I PODNIEBIENIA

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Abstract

Introduction: *Cleft lip and/or palate is the most common congenital craniofacial anomaly. Ultrasonography plays a key role in the early diagnosis of this anomaly and is completed by MRI. The purpose of this paper is to present and summarize the experience in diagnosis of cleft lip/palate by means of MRI.*

Material and methods: *The material consists of 62 fetuses that required more detailed evaluation which was conducted with the use of a 1.5 T scanner in SSFSE/T2 sequence in the sagittal, transverse and coronal plane.*

Results: *The cleft was diagnosed in 15 fetuses: an isolated cleft lip in one case (6.7%), a cleft lip and alveolar process in 2 (13.3%), a cleft lip and palate in 12 (80%). In eight fetuses (53.3%) the defect was unilateral, in 6 (40%) on both sides, in one case (6.7%) – a bilateral cleft lip and unilateral cleft palate was diagnosed. In three cases (20%), the cleft lip and/or palate defect was isolated, in 12 (80%) – it coexisted with other fetal abnormalities. MRI was less useful than ultrasound in 1 case (6.7%), in 4 cases (26.7%) it did not add any significant new information, in the remaining 10 cases (66.6%) important additional information was obtained on MRI. MRI revealed more details of the cleft in 5 cases (33.3%). In 10 fetuses (66.7%), cleft diagnosis was based on ultrasound and MRI only confirmed it. In 47 cases MRI allowed to show normal fetal faces, while there were difficulties of visualisation on ultrasound.*

Conclusions: *Prenatal MRI is a method supporting fetal ultrasound and is used to confirm/expand sonographic diagnosis, but can also change it. In the case of cleft lip and palate fetal MRI produces a better picture of the connections between the cavities, the degree of involvement of the secondary palate and cleft extent, and also helps to detect/assess other associated fetal abnormalities.*

Key words: magnetic resonance imaging (MRI), fetus, cleft lip, cleft palate

Streszczenie

Wprowadzenie: *Wada rozszczepowa wargi i/lub podniebienia jest najczęstszą wrodzoną anomalią rozwojową twarzoczaszki. Kluczową rolę we wczesnym rozpoznaniu rozszczepu pełni badanie ultrasonograficzne. Uzupełnia je badanie MR. Celem niniejszej pracy jest przedstawienie i podsumowanie doświadczenia w diagnostyce MR wady rozszczepowej.*

Materiał i metody: *Materiał stanowią badania MR 62 płodów zbadanych skanerem o natężeniu pola magnetycznego 1,5 T w sekwencji SSFSE w obrazach T2-zależnych w projekcji strzałkowej, poprzecznej i czołowej.*

Wyniki: *Wadę rozszczepową rozpoznano u 15 płodów: izolowany rozszczep wargi w 1 przypadku (6,7%), rozszczep wargi i wyrostka zębodołowego szczęki w 2 (13,3%), rozszczep wargi, wyrostka i podniebienia w 12 (80%). U 8 płodów (53,3%) wada była jednostronna, u 6 (40%) obustronna; w 1 przypadku (6,7%) wystąpił obustronny rozszczep wargi i jednostronny – podniebienia. W 3 przypadkach (20%) rozszczep wargi i/lub podniebienia był wadą izolowaną, w 12 (80%) – współistniały inne nieprawidłowości płodu. Badanie MR było mniej przydatne niż USG w 1 przypadku (6,7%), w 4 przypadkach (26,7%) nie wniosło*

istotnych nowych informacji do rozpoznania, w pozostałych 10 przypadkach (66,6%) dzięki badaniu MR uzyskano dodatkowe informacje. Badanie MR uwidoczniło więcej szczegółów rozszczepu w 5 przypadkach (33,3%). U 10 płodów (66,7%) rozpoznanie wady rozszczepowej ustalono na podstawie USG, a MR jedynie je potwierdził. W 47 przypadkach MR pozwolił ocenić twarzoczaszkę jako prawidłową przy trudnościach wizualizacyjnych w USG.

Wnioski: *Prenatalne badanie MR jest metodą uzupełniającą badanie USG płodu i służy do potwierdzenia/rozszerzenia rozpoznania ultrasonograficznego, choć również może je zmienić. W obrazowaniu rozszczepu wargi i podniebienia płodu badanie MR służy do lepszego obrazowania połączeń pomiędzy jamami, stopnia zajęcia podniebienia wtórnego i zasięgu rozszczepu, a także do wykrycia/oceny innych, towarzyszących wad.*

Słowa kluczowe: rezonans magnetyczny (MR), płód, rozszczep wargi, rozszczep podniebienia

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INTRODUCTION

Cleft lip and/or palate is the most common congenital craniofacial anomaly and second most common birth defect, accounting for 13% of fetal abnormalities. It is estimated that it accounts for about 65% of craniofacial defects and occurs with a frequency of 2:1, 000 live births. Cleft may involve any part of the face, but is usually located on the line running between the nostril and the central part of the posterior palate. Cleft lip occurs in 25% of cases, cleft lip and palate in 50%, cleft palate in 25%. A cleft can be an isolated malformation or part of one of more than 400 genetic syndromes (1).

The Institute of Mother and Child is a reference centre for multi-specialist diagnosis and treatment of this defect; diagnostics begins in the fetal life and is established on the basis of ultrasound (US) and magnetic resonance imaging (MRI). US plays a key role in the early diagnosis of this malformation. MRI is always performed after US if it is inconclusive, or requires confirmation, or other anomalies are suspected. In the postnatal period radiologists use X-rays, cone beam tomography and computed tomography in order to accurately assess the nature and extent of the changes for the purposes of surgical and orthodontic treatment (2).

Ten years of the Institute's Magnetic Resonance Unit's work have passed; the MR Unit is the first and one of the two centres in Poland dealing with prenatal diagnostics by means of MRI. Among many hundreds of fetal studies performed, there are also examinations of fetuses with congenital orofacial clefts. The purpose of this paper is to present and summarize our experience in the diagnosis of this particular defect.

MATERIAL AND METHODS

The material consists of MR examinations of 62 fetuses which were chosen as a result of retrospective analysis of prenatal studies performed in the Department of Diagnostic Imaging of the Institute of Mother and Child in the years 2004-2013 with the use of a 1.5 T scanner. MR studies were always performed after US if ultrasound was equivocal or required confirmation. Ultrasound examinations were performed at our Institute in 7 cases, eight fetuses were examined in other centres.

SSFSE/T2-weighted sequence (single-shot fast spin echo) was the main and primary MR sequence used to evaluate the fetal face. It was performed in three projections: the axial, sagittal and coronal one with the following parameters: repetition time, TR=5000 ms, echo time, TE=160 ms, number of acquisitions, NEX=0,56, matrix, MX=384 × 256, field of view, FOV=38 × 38 cm, slice thickness/interslice gap, ST=3,0/0,3 or 2,5/0,0 mm.

RESULTS

Among the 15 fetuses who were diagnosed with a cleft, there was an isolated cleft lip in one case (6.7%), a cleft lip and alveolar process in 2 (13.3%), a cleft lip and palate in 12 (80%). In eight fetuses (53.3%) the defect was unilateral, in 6 (40%) on both sides, in one case (6.7%) we dealt with bilateral cleft lip and unilateral cleft palate. In three cases (20%), the cleft lip and/or palate defect was isolated, in 12 (80%) – it coexisted with other fetal abnormalities. These results are presented in Table I.

The last column of the table shows the assessment of the role of US and MRI in the detection of fetal pathology. MRI turned to be less useful than ultrasound in 1 case (6.7%) – no. 9, in which the diagnosis of tetralogy of Fallot was established on the basis of US only. In 4 cases (26.7%) – nos. 1, 3, 14, 15 – MRI did not add any significant new information. In the remaining 10 cases (66.6%) important additional information was obtained on MRI and allowed the diagnosis of the whole spectrum of malformations. As far as cleft lip/palate is concerned, MRI revealed more details of the cleft in 5 cases (33.3%)%): nos. 2, 6, 7, 8 i 13 – showing cleft lip and palate that were not visible on US (no. 13), cleft alveolar process in fetus no. 2 and cleft palate in fetuses no. 6, 7 and 8. In the remaining 10 fetuses (66.7%), cleft diagnosis was based on ultrasound and MRI only confirmed it. Besides, in 47 cases MRI allowed to show a normal fetal face, while there were difficulties of visualisation on ultrasound.

Table I. The diagnosis of cleft lip/palate and of other anomalies in the analysed material.
Tabela I. Rozpoznania wady rozszczepowej oraz innych wad towarzyszących w analizowanym materiale.

No	Gestational age	Lip	Alveolus	Palate	Unilateral	Bilateral	Other malformations/problems; remarks	MRI vs. US
1		+	+	+	+	–	Underdevelopment of eyeballs	0
2	31	+	+	–	+	–	-	+
3		+	–	–	+	–	Semilobar holoprosencephaly, microphthalmia	0
4	22	+	+	+	–	+	Callosal agenesis, omphalocoele, double renal collecting systems	+
5	35	+	+	–	+	–	Persistent cloaca, right kidney agenesis	+
6	23	+	+	+	+	–	Alobar holoprosencephaly, hypertelorism	+
7	24	+	+	+	+	–	-	+
8	32	+	+	+	+ P	+ L	Oligohydramnios; cleft palate diagnosed only on MRI	+
9	27	+	+	+	+	–	Tetralogy of Fallot (diagnosed only on US)	-
10	29	+	+	+	–	+	Enlargement of right heart, thickening of walls and septum diagnosed only on US, dilatation of renal collecting systems diagnosed only on MRI, omphalocoele	+/-
11	26	+	+	+	–	+	Double outlet of the right ventricle diagnosed only on US, posterior urethral valve diagnosed only on MRI	+/-
12		+	+	+	–	+	Dilatation of renal collecting systems; megaureter on the right diagnosed only on MRI	+
13	25	+	+	+	–	+	Callosal agenesis, cleft lip and palate diagnosed only on MRI, shortening of long bones diagnosed only on US; oligohydramnios	+/-
14	31	+	+	+	–	+	Holoprosencephaly, hypotelorism, pleural and peritoneal effusion, generalised fetal hydrops	0
15		+	+	+	+	-	Alobar holoprosencephaly	0

DISCUSSION

The phrase: „The face could not be assessed due to inadequate visualisation of the fetus or fetal position" is quite often found in descriptions of prenatal ultrasound and then MRI is used (Fig. 1a, b). Although ultrasound remains the first diagnostic method in the prenatal period, it has its limitations. The field of view, the penetration of the ultrasound beam through fetal bones, including the facial bones and through the subcutaneous tissue of obese mothers are limited. Besides, the small volume of amniotic fluid, the location of the placenta on the anterior wall of the uterus, the fetal head being advanced in the birth canal and the fetal limbs overriding the face, make the study very difficult. The accuracy of sonographic assessment depends on the skill and experience of the operator (3-5).

Fetal MRI is not influenced by these factors, although, of course, the experience of the radiologist is crucial for the proper performance and analysis of the study. Fetal movements are a problem – sedation of the mother (and fetus) is not used in Poland and in most countries during MRI. But technical progress allowed for a marked reduction of the examination time, even to several seconds in the case of SSFSE/T2 sequence – during such a short time the influence of fetal movement on image quality is minimized. One should instruct the mothers to avoid caffeine-containing drinks (coffee, Coca-Cola) before the

examination in order not to provoke the movements of the fetus. It is also recommended to perform fetal MRI in the morning, after at least 4 hours of fasting, since fetal movements are reduced by hypoglycemia (6).

Cleft lip and/or palate is reflected on MRI by interruption of low-signal (dark) facial tissues by a kind of a channel filled with hyperintense (bright) amniotic fluid (Fig. 2). Additional abnormalities, such as a nasal septum distorted contralaterally to the cleft in a unilateral cleft (Fig. 2) and the elevated median nasal prominence (premaxillary protrusion, maxillary pseudomass – Fig. 3) and/or a flattened nose with a short columella in bilateral complete clefts are also clearly visible (1, 7, 8). The diagnosis of the rarely present, isolated cleft palate, which is particularly difficult to assess on ultrasound, is more complicated but possible on MRI (8.9). The lack of secondary palate is found in these cases in midsagittal view. Cine imaging during the act of swallowing is particularly useful (7, 10). It should be recalled in this place that the anomaly in question is divided into cleft of the primary palate, which includes lip and alveolar process anterior to the incisive fossa and cleft of the secondary palate, including the hard palate from the incisive fossa and the soft palate (1). Another additional MR technique is dedicated to imaging of the hard palate – echoplanar gradient echo sequence (GRE EPI) (11). MRI shows the connections between the cavities, the degree of involvement of the secondary palate and the lateral and anterior-posterior extent of the cleft better than US; the planning of surgical treatment is based on these latter elements (1, 12). However, although in many congenital defects and other congenital pathologies it has been shown that repeated MRI after birth usually does not bring significant new information as compared to the prenatal one and that postnatal treatment planning can be based on prenatal MRI (13, 14), cleft lip/palate is not one of them. Moreover, MRI is not repeated after birth in these cases - as mentioned in the introduction postnatal diagnostics is based on X-rays, cone beam CT, or computed tomography. However, the correct diagnosis of the defect and its extent in the prenatal period is especially important in terms of the parents' mental preparation for the forthcoming problems, such as those with feeding, and the prospect of long-term, multidisciplinary treatment.

Prenatal diagnosis concerns not only the cleft itself, but also other associated defects. These include defects of the musculoskeletal, cardiovascular and central nervous (CNS) systems (15). In our material CNS anomalies dominated; they were present in six fetuses (40%), while heart abnormalities were diagnosed in three (20%) and shortening of the long bones in one (6.7%). This explains the high proportion of cases in which MRI was superior to ultrasound – the evaluation of the CNS is the domain of magnetic resonance imaging. Ventricular enlargement and suspected defects of the midline structures of the brain

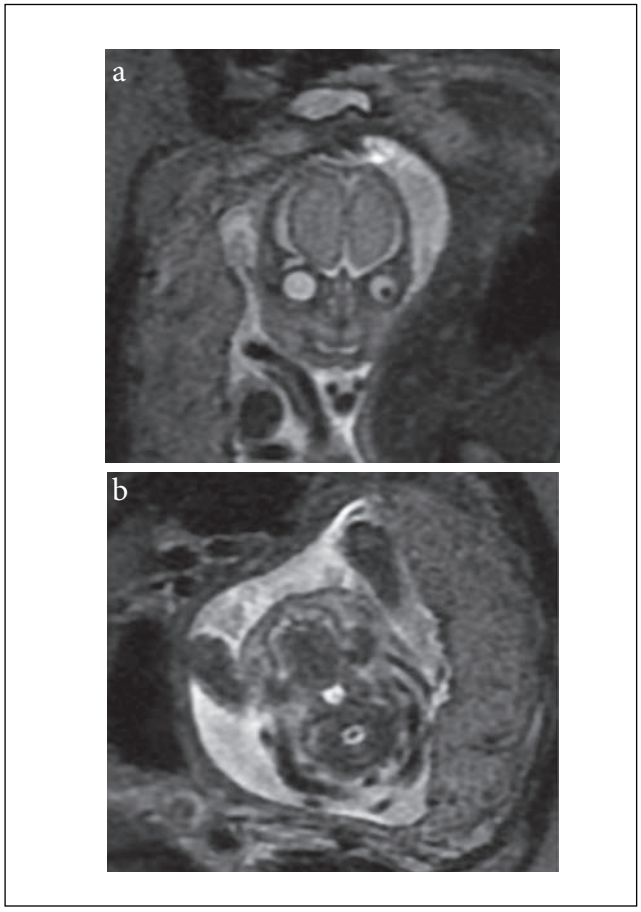

Fig. 1. Fetus, 23 weeks of gestational age (GA). MRI, SSFSE sequence, T2-weighted image (T2WI). Normal MR appearance of the lip, alveolus and palate in coronal (a) and axial (b) plane.

Ryc. 1. Płód 23 Hbd. Badanie MR, sekwencja SSFSE, obrazy T2-zależne. Prawidłowy obraz wargi, wyrostka zębodołowego i podniebienia w projekcji czołowej (a) i poprzecznej (b).

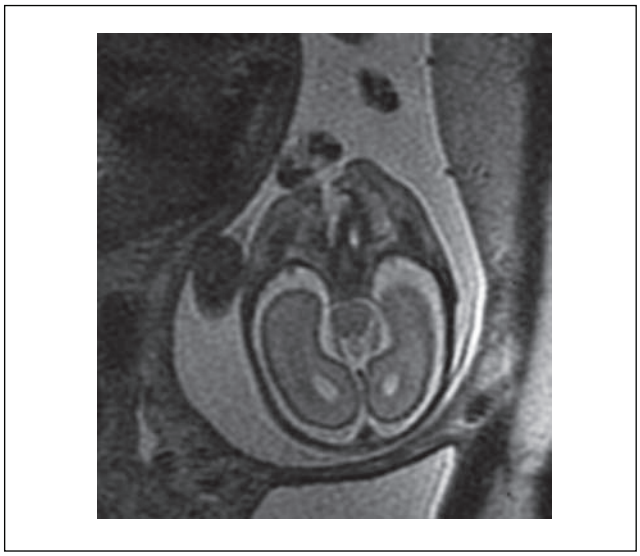

Fig. 2. Fetus, 24 weeks GA, case no 7. MRI, SSFSE sequence, T2WI, axial plane. Cleft lip, alveolus and palate on the right side, nasal ceptum deviation.

Ryc. 2. Płód 24 Hbd, przypadek nr 7. Badanie MR, sekwencja SSFSE, obraz T2-zależny, projekcja poprzeczna. Prawostronny rozszczep wargi, wyrostka zębodołowego i podniebienia, skrzywienie przegrody nosa.

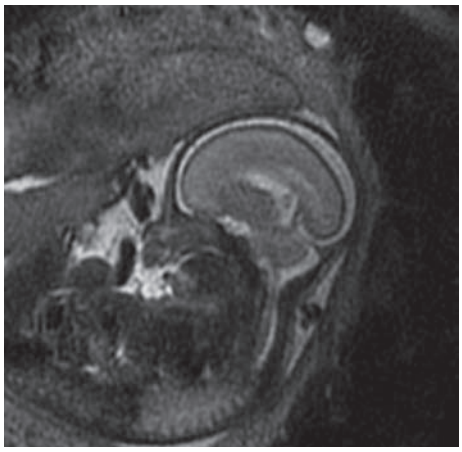

Fig. 3. Fetus, 25 weeks GA, case no 13. MRI, SSFSE sequence, T2WI, sagittal plane. Premaxillary protrusion resulting from the elevation of the median nasal prominence in the bilateral cleft.

Ryc. 3. Płód 25 Hbd, przypadek nr 13. Badanie MR, sekwencja SSFSE, obraz T2-zależny, projekcja strzalkowa. Uwypuklenie tkanek górnej wargi wskutek uniesienia wyniosłości nosowej pośrodkowej w obustronnej wadzie rozszczepowej.

are the first and main indications for prenatal MRI (16). Our experience indicates that both agenesis of the corpus callosum and holoprosencephaly, observed in fetuses nos. 3, 4, 6, 13, 14 and 15, may remain unrecognized in difficult conditions, or by an ultrasonographist with limited experience, and described as ventriculomegaly or hydrocephalus, which was the case in the above mentioned fetuses. MRI made it possible to establish a correct diagnosis of these associated anomalies (Fig. 4).

However, evaluation of the fetal heart and skeleton remains the domain of ultrasonography, although there have been significant advances in magnetic resonance imaging technology that allowed to assess these structures (1). However, in fetuses nos. 9, 10, 11 and 13, examined before the upgrade of the MR scanner at our Institute and before installing the new software, the diagnosis of heart defects and limb shortening was established on ultrasound. Only cardiomegaly was shown on MRI.

Our material collected in the tertiary referral centre is unique, and consists of cases with severe malformations, requiring highly specialized examinations, often associated with genetic disorders, and therefore is dominated by cases of multiple congenital abnormalities. However, craniofacial clefts usually occur as an isolated defect of the fetus. Nowadays multidisciplinary treatment (carried out by a maxillo-facial surgeon, orthodontist, otolaryngologist, phoniatrist, speech therapist, psychologist) provides a complete correction of the defect before the patient's coming of age, therefore the cleft alone does not fulfill the conditions of the Act of 7 January 1993 on family planning, protection of the human fetus and the conditions for termination of pregnancy. Correct prenatal parental counseling, which explains any doubts, stops the further steps to the termination of pregnancy.

If the cleft is associated with other defects or fetal chromosomal aberrations, the terms of the above mentioned Act may be fulfilled and the pregnancy can be terminated in accordance with the Act, before the fetus reaches the ability to live independently outside the uterus.

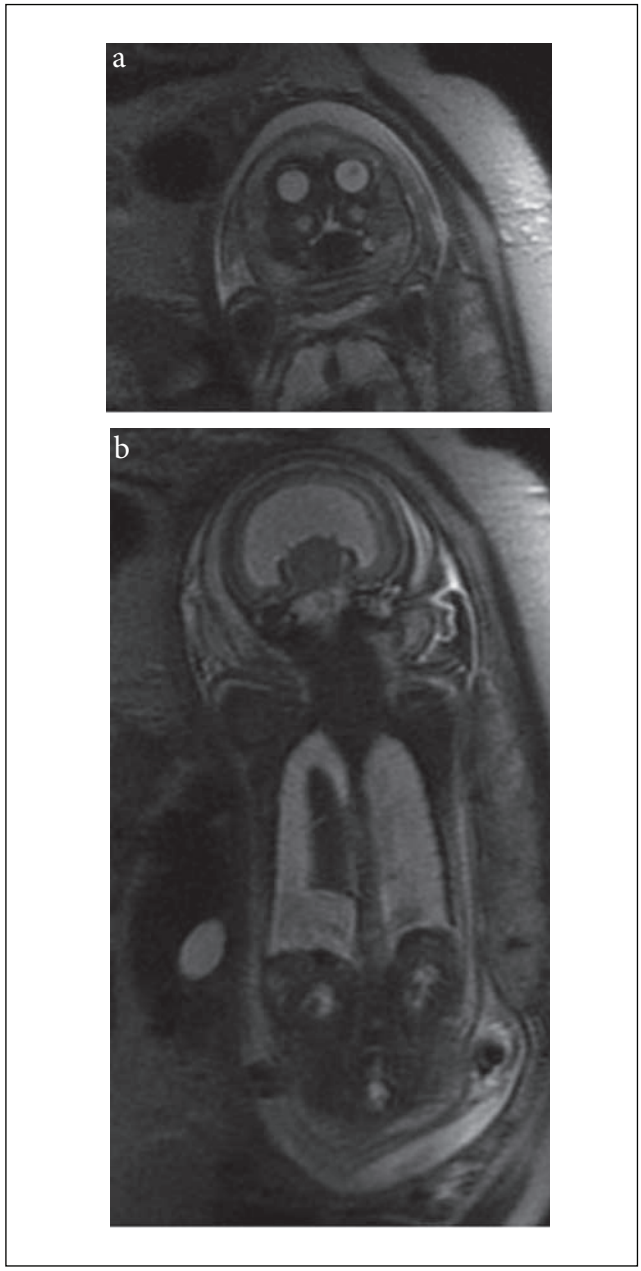

Fig. 4. Fetus, 31 weeks GA, case no 14. MRI, SSFSE sequence, T2WI, coronal plane.
a. Cleft lip on the left side, hypotelorism.
b. Holoprosencephaly, bilateral pleural effusions.

Ryc. 4. Płód 31 Hbd, przypadek nr 14. Badanie MR, sekwencja SSFSE, obrazy T2-zależne, projekcja czołowa.
a. Lewostronny rozszczep podniebienia, hipoteloryzm.
b. Holoprosencefalia, płyn w jamach opłucnowych.

CONCLUSIONS

Prenatal MRI is an adjunct to fetal ultrasound and in tertiary referral centres is used to confirm or expand sonographic diagnosis, but can also change it. In the case of equivocal US or when the diagnostics must be extended, pregnant women should be referred to the centres equiped with a 1.5 T scanner and expertise in the field of fetal MRI. MR imaging improves visualisation of the connections between the cavities, of the degree of the secondary palate involvement and of the extent of the cleft, as well as of other associated abnormalities.

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