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Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News

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KEY WORDS

postnatal diagnosis, Down syndrome, trisomy 21, postnatal care, delivery of healthcare, disclosure

ABBREVIATIONS

ACOG—American College of Obstetrics and Gynecology
DS—Down syndrome

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WHAT'S KNOWN ON THIS SUBJECT: Many parents of children with DS have expressed dissatisfaction with how their medical providers informed them of their child's diagnosis. As the most common chromosomal condition, DS is most often diagnosed postnatally.

WHAT THIS STUDY ADDS: This review provides physicians with an up-to-date, evidence-based structure on who should deliver a postnatal diagnosis of DS, when it should be given, where the discussion should occur, what should be said, and how best to say it.

abstract

CONTEXT: Many parents of children with Down syndrome (DS) have expressed dissatisfaction with how they learned about their child's diagnosis. DS remains the most common chromosomal condition, occurring in 1 of every 733 births, with the majority of children still diagnosed postnatally.

OBJECTIVE: Our goal was to review systematically all available evidence regarding how physicians should approach the conversation in which they explain DS for the first time to new parents.

METHODS: We searched online databases from 1960 to 2008, including Medline and PsychInfo, as well as Web sites maintained by academic organizations (eg, American Academy of Pediatrics) and other nonprofit or private organizations (eg, the National Down Syndrome Society), by using the terms "Down syndrome," "trisomy 21," "mongolism," "prenatal diagnosis," "postnatal care," and "delivery of health care." Articles were selected that answered ≥1 research question, established a priori: (1) Who is the best person to communicate the news? (2) When is the best time to share the news? (3) Where is the best place or setting to deliver the news? (4) What information should be delivered? and (5) How should the news be communicated? All studies were evaluated for quality according to the method outlined by the US Preventative Services Task Force. Final recommendations were based on the strength of evidence.

RESULTS: Parents prefer to receive the diagnosis together in a joint meeting with their obstetrician and pediatrician. The conversation should take place in a private setting as soon as a physician suspects a diagnosis of DS. Accurate and up-to-date information should be conveyed, including information about local support groups and resources.

CONCLUSION: By implementing a few cost-neutral measures, physicians can deliver a postnatal diagnosis of DS in a manner that will be deemed by new parents as sensitive and appropriate. *Pediatrics* 2009; 124:e751–e758

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Although the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics now recommends that all pregnant women, regardless of age, be offered prenatal testing for Down syndrome (DS),[1–3] studies suggest that >85% of mothers who have children with DS first received the diagnosis postnatally.[4,5] DS is the most common chromosomal condition, occurring in 1 of every 733 live births with >400 000 persons with DS estimated to be living in the United States.[6] As such, today's neonatologists, geneticists, family practitioners, hospitalists, and general pediatricians can expect to join obstetricians in being among the first physicians who will share the diagnosis with the new parents.

Delivering and receiving that news is not simple. Even the most seasoned physicians admit that they have little, if any, training on how to discuss a new diagnosis of DS in a sensitive manner.[7] In a survey of 2500 medical school deans, students, and residency directors, 81% of medical students report that they "are not getting any clinical training regarding individuals with intellectual disabilities," and 58% of medical school deans say such training is not a high priority.[8] Mothers and fathers report feeling "shocked," "angry," "devastated," "overwhelmed," "depressed," "stunned," and/or "helpless" when they first learn of the diagnosis.[4] In addition, parents from many parts of the world such as England, Scotland, Ireland, Spain, Sweden, Australia, and the United States have reported strong dissatisfaction with the way in which the diagnosis was conveyed and the amount of support provided during the immediate postnatal period.[4]

Our primary goal for this report was to review the current evidence on how physicians should best deliver a postnatal diagnosis of DS to new parents. Specifically, we asked the following questions: Who is the best person to communicate the news? When is the best time to share the news? Where is the best place/setting to deliver the news? What information should be given? How should the news be communicated? We further graded the evidence in an effort to provide today's physicians with evidence-based recommendations on how to approach these sensitive encounters.

METHODS

Sources

This study was part of a larger literature review where we searched online databases including Medline and PsychInfo for all studies published in English from 1960 through February 2008, by using the following National Library of Medicine Medical Subject Headings terms: "Down syndrome," "trisomy 21," "mongolism," "prenatal diagnosis," "postnatal care," and "delivery of health care." To ensure completeness of the literature search, we reviewed reference lists and articles from the authors' libraries. We further supplemented the primary literature search by searching the Web sites of the following entities: Google Scholars; the American Academy of Pediatrics; the ACOG; the American College of Medical Genetics; the National Newborn Screening and Genetics Resource Center; the National Down Syndrome Society; the National Down Syndrome Congress; March of Dimes; the National Federation of Voluntary Bodies; the International Mosaic Down Syndrome Association; the Down Syndrome Educational Trust; the Canadian Down Syndrome Society; and the United Kingdom Down Syndrome Association. Additional studies were identified by reviewing references of previously screened articles.

Study Selection and Data Extraction

Before the start of this review, we solicited input and advice from an expert in study design and public health policy. To keep our initial search as unbiased and comprehensive as possible, we chose to review a wide range of study designs, selecting to analyze, on a consistent basis, the titles and abstracts of the articles that seemed to answer ≥1 of our research questions, established a priori: (1) Who is the best person to communicate the news? (2) When is the best time to share the news? (3) Where is the best place or setting to deliver the news? (4) What information should be delivered? (5) How should the news be communicated? Our research questions for the prenatal period were reported elsewhere.[9]

After independently reading, in full, all of the articles meeting the initial criteria, the primary authors discussed and then eliminated those articles that (1) on full read did not answer any of the research questions established a priori, (2) did not have results that were specific to DS, (3) contained only duplicative and not original data, (4) contained only opinion based on clinical or personal experience, or (5) had a participant pool of <10 persons. No discordant opinion among the authors occurred with this criteria. In total, we identified 19 articles, collectively sampling 3359 parents who received a postnatal diagnosis of DS for their child.[4,10–27] The studies came from a variety of countries, were performed from 1964 to 2005, and sampled primarily mothers (Table 1). All studies were evaluated for quality by 1996 US Preventative Services Task Force guidelines (levels of evidence are indicated in Table 1).[28] Final recommendations for practitioners were based on the strength of evidence, as assessed by both the 3-tiered levels used by the

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TABLE 1 Details of Articles Included in the Literature Review, Listed in Chronological Order

Reference No.	Year of Publication	Location of Study	Participants, *n*	Study Design	Level of Evidence[a]
14	1964	United Kingdom	71 mothers	Retrospective case study with in-person interviews	II-3
10	1969	United Kingdom	95 mothers	Retrospective case study with in-person interviews	II-3
11	1970	United Kingdom	46 mothers	Retrospective case study with in-person interviews	II-3
24	1973	Scotland	31 mothers and 31 fathers	Retrospective case study with in-person interviews	II-3
16	1974	United States	85 mothers and 85 fathers	Retrospective case study with telephone interviews	II-3
21	1976	United States	414 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
13	1977	United Kingdom	30 mothers and 21 fathers	Retrospective case study with in-person interviews	II-3
22	1978	United Kingdom	54 mothers and fathers combined	Retrospective case study with in-person interviews	II-3
23	1980	United States	35 mothers and 3 fathers	Retrospective case study with telephone interviews	II-3
18	1980	Ireland	79 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
19	1983	Ireland	123 mothers	Retrospective case study with mailed questionnaires	II-3
12	1984	United Kingdom	59 mothers and 58 fathers	Prospective nonrandomized control trial between mothers of children with DS who received the diagnosis by a "model service" and those who received customary disclosure; evaluated with in-person interviews	II-1
20	1985	United States	285 mothers and fathers combined	Retrospective case study with mailed questionnaires	II-3
25	1986	United Kingdom	63 mothers and fathers combined	Retrospective case study with in-person interviews	II-3
26	1994	United Kingdom	56 mothers and fathers	Retrospective case study with in-person interviews	II-3
15	1995	United States	18 mothers and fathers	Retrospective case study with in-person interviews	II-3
17	2002	Sweden	165 mothers and fathers	Retrospective case study with mailed questionnaires	II-3
27	2005	Spain	467 mothers	Retrospective case study with mailed questionnaires	II-3
4	2005	United States	985 mothers	Retrospective case study with mailed questionnaires	II-3

[a] Levels of evidence as established by 1996 US Preventative Services Task Force guidelines.[28]

ACOG[1,2] (with which level A recommendations are based on "good and consistent scientific evidence," level B recommendations are based on "limited or inconsistent scientific evidence," and level C recommendations are primarily based on "consensus and expert opinion") and the 2-tiered levels used by the Grading of Recommendations, Assessment, Development, and Evaluations (GRADE) system[29] (with which a "strong recommendation" means that "most informed patients would choose the recommended management and that clinicians can structure their interactions with patients accordingly" and a "weak recommendation" means that "patients' choices will vary according to their values and preferences, and clinicians must ensure that patients' care is in keeping with their values and preferences").

RESULTS

Who Is the Best Person to Communicate the News?

Mothers first learned that their infants had DS from a variety of people: "ward sisters"[11,12,19]; nurses[4,11,19,21,24]; spouses[16,19–21,23,24]; lactation specialists[4]; hospital volunteers[4]; genetic counselors[4]; midwives[12]; medical students[21]; medical residents[19]; primary care practitioners[12,16,19–21,23]; pediatricians (including geneticists and neonatologists)[4,12,16,17,20,21,23,24]; and obstetricians.[4,12,16,20,21,23] Since 1964 when researchers began tracking maternal reactions, however, mothers have offered consistent advice on from whom among this list they would prefer—and not prefer—to hear.

The person to first deliver the news should be a physician knowledgeable about DS, according to most mothers.[4,11,13,16,18,19,21,27] In some cases, mothers preferred this clinician to be their obstetrician[16,21] because a "trust relationship with the obstetrician had been established during the antenatal period."[21] Other mothers valued a pediatrician or pediatric subspecialist given their expertise and training in childhood conditions.[16] Recommendations from the largest studies, to date, suggest that a "combined counseling effort of obstetrician and pediatrician might relieve some of the parental anxieties,"[21] and "all hospitals should have a plan in place so that all relevant physicians know how best to coordinate their messages."[4]

Mothers have been clear from even the earliest studies that they do not want to hear the news first from their husbands or partners.[4,16,23,24] Doing so, they argue, creates an unfair burden on a person who is experiencing the same initial feelings of shock, grief, and sadness. Nor should the person who delivers the news be a health care professional who is not fully trained to explain DS, such as lactation specialists, hospital volunteers, medical students, medical residents, and most midwives and nurses.[4,21] Although these persons might not be the first communicators, the largest study on this topic, to date, emphasizes that "it takes a committed team to make the experience of having a child with DS a positive one."[4]

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When Is the Best Time to Share the News?

In general, mothers prefer learning about DS as soon as possible even when the diagnosis is not confirmed[16,24,27]; however, physicians must also use their best judgment and delay informing the mother if she seems ill[10] or is still recovering from the delivery.[4,10,17] Across time, mothers most often complained about a delay in receiving information, feeling that health care professionals were reluctant to disclose information regarding their child's condition.[24] The percentage of mothers who were notified of the diagnosis within 24 hours of their child's birth has been improving: 35% ($n = 31$) in 1973,[24] 28% ($n = 406$) in 1976,[12] 56% ($n = 54$) in 1978,[22] 37% ($n = 78$) in 1980,[18] 37% ($n = 123$) in 1983,[19] 29% ($n = 59$) in 1984,[12] 68% ($n = 139$) in 1985,[20] 63% ($n = 63$) in 1986,[25] 63% ($n = 56$) in 1994,[26] and 75% ($n = 81$) in 2002.[17]

In most instances, a diagnosis of DS is based on distinctive physical features, quite apparent at birth, so parents are notified on the day of delivery or on the following day.[19,20] Even when a physician has not reached a definitive diagnosis, he should report his or her suspicions to the parents as soon as possible.[13,20,27] Most mothers recognize that something is different about their infant before ever having a conversation with their physician.[19,22] By delaying the conversation or waiting for a confirmatory karyotype result, physicians cause unnecessary anxiety in parents.[19,22,27] Parents further report that by having an early discussion, they can prepare themselves for more intense subsequent discussions, during which more of their questions can be answered.[13,21]

Where Is the Best Place or Setting to Deliver the News?

Across time, parents have received the diagnosis in a variety of locations, including private hospital rooms,[18,19] shared inpatient rooms,[19,24] surgical suites,[19] and at home.[11,19] Overwhelmingly, parents prefer receiving the diagnosis in a private place, where no other medical personnel are present.[12,15,19] Parents have expressed intense dissatisfaction when they have received the diagnosis in the presence of hospital roommates and visitors.[4,15,27] Mothers most often desired the company of their husband or partner when hearing the diagnosis, while being secluded from all others.[12,13,15,16,23,24,27] The percentage of mothers who have received the diagnosis with their partner present has been improving over time: 33% ($n = 36$) in 1970,[11] 24% ($n = 170$) in 1974,[16] 20% ($n = 417$) in 1976,[21] 30% ($n = 30$) in 1977,[13] 27% ($n = 37$) in 1980,[23] 17% ($n = 78$) in 1980,[18] 47% ($n = 123$) in 1983,[19] 75% ($n = 59$) in 1984,[12] 33% ($n = 139$) in 1985,[20] and 79% ($n = 77$) in 2002.[17] Whenever possible, health care professionals should provide parents with a private place to talk with each other without interruption immediately after disclosure.[4,12,13]

What Information Should Be Given?

Across time, parents have been as consistent about information that they would not like to receive as they have been about the details that should be discussed. They frequently resented information perceived as vague, inaccurate, or outdated.[10–12,18,22,25] An emphasis on overly pessimistic or offensive terminology (eg, "mongolism") was commonly regarded as both unbalanced and hurtful.[15–17,20,21,23,24,27] Too much information about possible medical conditions occurring later in life, such as obesity, leukemia, and Alzheimer disease, was also felt by parents to be overwhelming for the first conversation.[17,23,27]

By contrast, parents generally expressed 3 desires about the information that they would like to receive. First, parents want to have access to complete and accurate information for the following questions: What is DS? What is its cause? And, what does it mean for a family to have a member with DS, in practical terms?[13,16,18,26] This should include providing parents with the contact information for local parent support groups and community resources.[4,16,18,21] Second, parents want the information to be balanced, realistic, and contemporary given the current possibilities for people with DS in today's society. This should include providing the parents with an up-to-date bibliography of resource books on DS.[4,13,20,22,23,27] Third, the information provided during this initial discussion should be limited to the most immediate or common medical conditions.[12,15,16,18,21] The percentage of mothers who have felt that they received complete, accurate information has varied over time, with a trend for less satisfaction in more recent years: 60% ($n = 43$) in 1970,[11] 61% ($n = 85$) in 1974,[16] 84% ($n = 414$) in 1976,[21] 55% ($n = 33$) in 1977,[13] 53% ($n = 79$) in 1980,[23] 65% ($n = 139$) in 1985,[20] 11% ($n = 27$) in 1986,[25] and 30% ($n = 165$) in 2002.[17]

How Should the News Be Communicated?

Again, parents were equally clear about the ways in which the diagnosis should not be communicated as they were about how the news should be conveyed. Parents resented when the information was delivered in a manner perceived to be insensitive, unkind, or unconcerned with the welfare of the mother.[12,20,22,25,26] They further thought the delivery was unprofessional when news was provided to 1 parent before the other.[15,25] The use of language conveying pity (eg, "I am sorry to have to tell you this, but . . ."), personal tragedy (eg, "Unfortunately, I have some bad news to share . . ."), or extreme sorrow (eg, "I know this might seem like a devastating loss . . .") was considered un-

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necessary and not always reflective of mothers' emotional states.[4,24] The percentage of mothers who felt that their physicians used respectful, nonjudgmental language varied over time, with no particular trends: 30% ($n = 95$) in 1969,[10] 24% ($n = 79$) in 1980,[18] 74% ($n = 59$) in 1984,[12] 65% ($n = 139$) in 1985,[20] 44% ($n = 62$) in 1986,[25] and 44% ($n = 65$) in 2002.[17]

Research has shown that mothers forever remember the first words that their physicians use.[30] They expressed the most satisfaction when their physicians offered congratulations over the fact that they had just had an infant.[4] Parents further indicated a desire to be told the news with both partners and the infant present, whenever feasible.[12,31] They also request sufficient time to receive a thorough explanation, with the opportunity to have all of their questions answered. At the end of the initial discussion, a follow-up appointment should be arranged within several weeks.[11,12,14,18,19,24,26]

CONCLUSIONS

Although delivering a diagnosis of DS for the first time to new parents is not a comfortable situation for most physicians and families, researchers have been offering evidence-based suggestions since 1964 on how to make these encounters more sensitive. In fact, despite their commonly shared feelings of shock, anger, and fear after receiving such a diagnosis, parents can and do rate their physicians positively when some simple measures are followed.[4,12]

Recommendations

The following recommendations are based on consistent evidence from the articles that were reviewed and are rated according to the criteria established both by the ACOG[1,2] and the GRADE system.[29] These suggestions are meant to serve as helpful guideposts for today's physicians but should not be considered inclusive of all possible recommendations. Likewise, adherence to these suggestions does not necessarily ensure a satisfactory experience for both the physician and patient. Recommendations are offered for the ideal situations, with the understanding that some measures might need to be adapted to fit the resources available within a particular health care community. Nevertheless, the evidence suggests that most parents receiving a postnatal diagnosis of DS would want the following measures implemented:

- Obstetricians and pediatricians (or pediatric subspecialists) should coordinate their messaging and be the persons who first deliver the news to the parents. In ideal circumstances, the parents' obstetrician and pediatrician would meet jointly with the couple to explain DS. If this is not possible because of limited resources or if this would cause a prolonged delay to coordinate efforts, the obstetrician and pediatrician should connect with each other to ensure that a consistent message is conveyed (level A; strong recommendation).
- Physicians should inform parents of their suspicion for DS immediately, even if the diagnosis has not been confirmed by a karyotype result. Physicians can usually recognize DS immediately after the birth of a child, and often, parents can also discern that something is different about their infant before ever speaking with the doctor. Physicians must use their best judgment, however, in determining the precise timing of disclosure. For example, if the mother is ill after the childbirth, the physician might wait until the mother has recovered (level A; strong recommendation).
- Physicians should deliver the diagnosis in a private hospital room, away from other medical personnel, patients, and visitors (including other family members). Whenever possible, physicians should also offer the parents a private place to talk without interruption immediately after they have received the diagnosis (level A; strong recommendation).
- Parents should be informed together. Exceptions to this include when availability of the father or partner would significantly delay the conversation or in circumstances where the mother does not wish the child's father to be present. In the case where a mother is alone and preemptively asks her physician if "anything is wrong with her infant," the physician should ask if she would like to wait for her husband or partner to be present to talk about some observations. If not, the physician should plan to reexplain the diagnosis to the husband or partner when they arrive, if desired by the mother (level A; strong recommendation).
- The infant with DS should be present during the conversation and referred to by name by the physician (level C; weak recommendation).
- Physicians should begin their conversations with positive words, such as congratulating the parents on the birth of their child. They should avoid language conveying pity, personal tragedy, or extreme sorrow; moreover, they should avoid offering unsolicited personal opinions. The first few words that doctors use have been shown to set the tone for the remainder of the conversation. Moreover, mothers remember the first words >20 years after the initial discussion (level A; strong recommendation).
- Physicians should provide parents with accurate information that emphasizes in very practical terms, what DS is, what causes the condi-

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tion, and what it means to live with DS in today's society. As part of this explanation, physicians should hand parents an up-to-date bibliography list of DS resources, such as the reference lists available from the National Down Syndrome Society (www.ndss.org) or the National Down Syndrome Congress (www.ndsccenter.org) (level A; strong recommendation).

- For the first conversation, physicians should limit their discussions on possible medical conditions to those that the infant is suspected of having (or has) and those conditions seen in young children with DS under the age of 1 year. Physicians should convey the frequency of developing any particular condition as well as available treatments or therapies, such as speech, occupational, or physical therapy. Discussion about transient myeloproliferative disorder, acute myelogenous leukemia, or acute lymphoblastic leukemia should be saved for subsequent conversations. The health care guidelines for children with DS in the United States[30–32] are available at www.ndss.org and www.ndsccenter.org (level C; weak recommendation).
- The physician should offer contact information for local support groups and community resources to the new parents. If the family is interested and consents, the physician might even proactively contact the local support group and forward the contact information for the new family. Connecting the new parent with another parent has been shown to be among the most helpful measures a physician can do during the first conversation. Local DS support groups in the United States can be quickly located through the National Down Syndrome Society (www.ndss.org) and the National Down Syndrome Congress (www.ndsccenter.org) (level A; strong recommendation).
- Follow-up appointments should be arranged, as desired by the parents, with the medical professionals who have an expertise in DS (eg, geneticists, genetic counselors, developmental-behavioral pediatricians). A directory of DS specialty clinics within the United States is available at www.ndss.org (level C; weak recommendation).

Future Research

Despite the enormous body of literature that exists on this subject, several crucial questions remain without evidence-based answers. First, how much accurate and up-to-date information should be divulged to parents during the first conversation? And, in how much detail should physicians describe the medical conditions associated with infants with DS? Research clearly shows that mothers retain with great accuracy the first words that physicians use[4]; other studies demonstrate that they can recall with nearly 82% accuracy most of the conversation ~20 years later.[33] Yet, how much is too much? And, what information might be better conveyed through books and handouts distributed after this first conversation?

Although this review focuses exclusively on the first conversation with new parents, equally important are the dynamics of the subsequent conversations. Who should meet with the parents next? When and where should this meeting take place? What information should be introduced and discussed then? Research is noticeably absent in addressing these questions, and many others, regarding continued outreach to parents.

In addition, our research incorporates only those studies printed in English, and most of the participants were white. Future research should seek to incorporate parents with more socioeconomic and cultural diversities from the United States and other countries so that support and outreach could target unique needs.

Implications

The recommendations offered in this review article do not require many financial resources, if any, to be implemented by physicians. In many ways, the suggestions might even seem obvious to some. Yet, if these suggestions are cost-efficient and commonsensical, then why have thousands of mothers over decades of research indicated that their physicians have not incorporated these measures? Part of the explanation can likely be attributed to physicians' lack of training.[8] For the recommendations in this review article to be implemented, medical schools, nursing schools, genetic counseling schools, pediatric residency programs, obstetrician/gynecologist residency programs, family medicine residency programs, and associated fellowship programs need to work collaboratively with leaders in the DS community on proper training. Educational opportunities include lecture series, grand rounds presentations, clinical experiences, and online simulation.[34] After nearly 50 years of research on how physicians communicate a diagnosis of DS, the time has long come for progress.

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Dr Skotko holds a salaried position at Children's Hospital Boston and is sometimes remunerated as a speaker on Down syndrome for hospitals and support groups; serves without pay on the Board of Directors of the National Down Syndrome Society and the Massachusetts Down Syndrome Congress; and serves without pay on the Professional Advisory Council to the National Down Syndrome Congress. Dr Kishnani holds a salaried position in the Division of Medical Genetics, Department of Pediatrics at Duke University; has received occasional honoraria from academia and pharmaceutical companies such as Eisai, Pfizer, and Novartis for expert opinion on Down syndrome; and serves without pay on the Clinical Advisory Board of the National Down Syndrome Society. Dr Capone holds a salaried position as the director of a Down syndrome clinic, is sometimes remunerated as a speaker on health issues of persons with this condition, and serves without pay on the Board of Directors of the National Down Syndrome Congress and on the Scientific Advisory Board of the National Down Syndrome Society and the Down Syndrome Research Coalition.

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