



Bibliometric Analysis On Non-Invasive Prenatal Testing

Non-invaziv Prenatal Testler Üzerine Bibliyometrik Analiz

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ABSTRACT

Aim: The study aimed to analyze and assess the global research development, collaborative network, prestigious literature and tendency of themes about Non-invasive prenatal testing(SDisease) (NIPT).

Material and Method: A total of 1639 scientific publications between the years 2011-2025 were examined in this study from the Web Science (WoS) database databases. Country and institutional collaborations, co-citation networks, and keyword collocations were visualized for the data analysis in VOSviewer.

Results: The publications in the area of NIPT were found to have increased over years. The PR China ranked the first both in terms of publications (n=384), with United States of America and United Kingdom being the countries with most significant international collaboration networks included respectively (n=206) and (n=199). The journal Prenatal Diagnosis contributed most, with 171 publications and 4,243 citations. Results Analysis of the Keywords showed that research interest has shifted from simple aneuploidy screening to technical (fetal fraction, mosaicism and next-generation sequencing) as well as social aspects (genetic counseling and ethical issues).

Conclusion: The NIPT field is a very active and mature area of research with an increasing volume of interest. The People's Republic of China is the largest volume contributor, US and UK are in the hub of international collaboration and scientific influence. The emphasis in the field has changed from pure screening to more challenging issues like fetal fraction, mosaicism, genetic counseling and ethics.

Keywords: Bibliometrics, publications, non-invasive prenatal testing

ÖZ

Amaç: Bu çalışmanın amacı, non-invaziv prenatal test (NIPT) alanındaki küresel bilimsel üretimi bibliyometrik yöntemlerle inceleyerek araştırma eğilimlerini, uluslararası iş birliği ağlarını ve alandaki etkili yayınları değerlendirmektir.

Gereç ve Yöntem: Çalışma kapsamında, 2011–2025 yılları arasında Web of Science (WoS) veri tabanında indekslenen toplam 1639 bilimsel yayın bibliyometrik analiz yöntemi ile incelenmiştir. Veri analizinde ülkeler ve kurumlar arası iş birlikleri, eş-atıf ağları ve anahtar kelime birliktelikleri değerlendirilmiş; elde edilen veriler VOSviewer yazılımı kullanılarak görselleştirilmiştir.

Bulgular: NIPT alanındaki bilimsel yayın sayısının yıllar içerisinde belirgin bir artış gösterdiği saptandı. Çin Halk Cumhuriyeti, toplam 384 yayın ile en fazla katkı sağlayan ülke olarak belirlenirken; Amerika Birleşik Devletleri (n=206) ve Birleşik Krallık (n=199) uluslararası bilimsel iş birliği ağlarında en güçlü konuma sahip ülkeler olarak öne çıkmıştır. Prenatal Diagnosis dergisi, 171 yayın ve 4.243 atıf ile alana en fazla katkı sağlayan dergi olarak belirlenmiştir. Anahtar kelime analizleri, araştırma odağının zaman içerisinde yalnızca anöploidi taramasına yönelik çalışmalardan; fetal fraksiyon, mozaizm ve yeni nesil dizileme teknolojileri gibi teknik konular ile genetik danışmanlık ve etik boyutlar gibi sosyal ve klinik konulara doğru genişlediğini göstermiştir.

Sonuç: Bulgular, NIPT alanının giderek artan bilimsel üretim hacmi ile dinamik ve gelişmekte olan bir araştırma alanı olduğunu ortaya koymaktadır. Çin Halk Cumhuriyeti yayın sayısı açısından en büyük katkıyı sağlarken, Amerika Birleşik Devletleri ve Birleşik Krallık uluslararası iş birlikleri ve bilimsel etki açısından merkezi bir konumda bulunmaktadır. Ayrıca, alandaki araştırma eğilimlerinin yalnızca tarama uygulamalarından daha karmaşık teknik ve etik konulara doğru evrildiği görülmektedir.

Anahtar Kelimeler: Bibliyometri, bilimsel yayınlar, non-invaziv prenatal test (NIPT)

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INTRODUCTION

Non-invasive prenatal testing (NIPT) was first discovered in 1988 (1) and is a prenatal test that helps detect common aneuploidies such as Down (T21), Edwards (T18) and Patau (T13) syndromes. This test is a simple, low-risk screening method that involves analysing cell-free foetal DNA (cfDNA) obtained from the mother's blood using next-generation sequencing technologies (1,2). It came into widespread use in 2011 (3). Studies are being conducted on the reliability of NIPT, and significant resources are being allocated for its integration into healthcare systems worldwide (1,2). In a pilot study conducted in China, NIPT was used as a first-line screening test for Trisomy 21, 18, 13, and sex chromosome aneuploidies in 42,924 pregnancies. NIPT demonstrated a high positive predictive value (PPV), particularly for Trisomy 21 and 18, and these values were observed to increase with age. This study suggests that NIPT has the potential to replace current biochemical screening tests (4).

Although it offers an important opportunity for screening for foetal aneuploidies, there are various challenges and limitations regarding the widespread adoption and standardisation of the test (1,2). The success of the test depends on the accurate measurement of the amount of foetal DNA (foetal fraction) in the blood sample. The widespread use of NIPT has also brought with it ethical debates, such as potential over-screening and the psychosocial effects of the results. Therefore, it is crucial for obstetricians to play the right role in genetic counselling and to manage patient expectations. Studies on the use of NIPT as a first-line screening test are ongoing (3).

This study quantitatively examines global research trends in the scientific literature on NIPT, the key contributing institutions/countries, and the distribution of publications over the years. It also visualises country, institution, collaboration networks, the journals with the highest number of publications, and key word co-occurrence maps to reveal current research themes and potential future directions in the field.

MATERIAL AND METHOD

This is a bibliometric retrospective study.

Material (Data Source) Database Selection: To ensure the comprehensiveness and reliability of the study, the Web of Science (WoS) Core Collection, one of the most reputable bibliographic databases, was selected.

Search Strategy: A comprehensive search query specific to the defined topic was created. The keywords for the search string were: ("Non-Invasive Prenatal Testing" OR "Non Invasive Prenatal Testing" OR "cell-free fetal dna" OR "cffdna" OR "fetal fraction" OR nipt OR "non-invasive prenatal diagnosis" OR "non-invasive prenatal screening"

OR "non-invasive prenatal test" OR "non-invasive prenatal testing nipt" OR "prenatal screening"). This string has been combined using logical operators (OR) to cover all relevant publications in the selected Web of Science bibliographic database. This search strategy aims to collect all variations of screening and diagnostic methods based on cell-free foetal DNA (including abbreviations and full terms) by covering both new and old publications in the field.

This database review was conducted on 16 September 2025.

Inclusion criteria and filtering

Time period: The time period from the beginning of publications in the field of NIPT to the present day was determined (1 January 2011 – 15 September 2025).

Document Type: Publication type was limited to 'Article' and 'Review' types.

Language: The language of the articles was limited to 'English'.

The data collection process for the bibliometric analysis of the literature was carried out using a multi-stage filtering process. A comprehensive keyword search strategy initially identified a total of 2,235 publications. Subsequently, a time range restriction (from 2011 to September 2025) was applied to reflect the period during which NIPT entered clinical practice, reducing the number of publications to 2,077. In order to focus the study on fundamental scientific outputs, the document type was limited to articles and reviews only, resulting in 1,739 publications. After removing duplicate publications to eliminate repetitions in the dataset, the number of publications was 1,702. Finally, only English publications were selected with a language restriction, resulting in 1,639 publications that formed the final sample of the study. These 1,639 publications formed the basic dataset for the VOSviewer analysis.

Data Download: Search results were downloaded from the selected database in Full Record and Cited References format. The downloaded file was in a format supported by VOSviewer (Plain Text format from WoS).

Analysis Method

Data Cleaning and Harmonisation: The downloaded dataset was examined for spelling errors, variant names and duplicate records before being loaded into the bibliometric analysis software, and was cleaned and harmonised.

Analysis Software: VOSviewer V 1.6.20 software was used for bibliometric analysis and visualisation.

Within the scope of these analyses, Country/Institution Collaboration (Co-authorship Analysis) was conducted to map international collaboration networks; Keyword Co-occurrence Analysis was

performed to identify core themes and current and emerging topics in the field of NIPT, and the results were presented using Network and Overlay Visualisations. Furthermore, Citation Analysis was applied to identify the most cited authors, articles, and journals in order to determine the impact power within the literature. Finally, Bibliographic Coupling or Co-citation Analysis was used to map the conceptual structure and theoretical foundations between studies, and all results obtained were interpreted in detail through network maps and layered visualisations created by VOSviewer.

RESULTS

Main features

According to the inclusion criteria, 1,639 publications were included in the study. The majority of this sample, 1,406 publications (85.784%), were of the article type, while the remainder were of the review type. The distribution of these publications in Web of Science (WoS) subdatabases showed that the research was predominantly scientific and clinically oriented. The majority of the publications came from the Science Citation Index Expanded (SCI-EXPANDED), which covers core scientific journals, with 1,461 publications (89.140%). The Emerging Sources Citation Index (ESCI), which reflects the contribution from journals new to the field, was included with 178 publications (10.860%). The Social Sciences Citation Index (SSCI), which focuses on the ethical and social dimensions of NIPT, included 143 publications (8.725%), while the Book Citation Index – Science (BKCI-S), which includes scientific books and book chapters, contributed 30.183% of the total publications. This distribution revealed that the NIPT literature is largely dominated by SCI-EXPANDED. Since a publication can be included in more than one index, the sum of the percentages exceeds 100%.

Publications and Citations per Year

As illustrated in **Figure 1**, there are 1,639 manuscripts on NIPT from 2011 to 2025 and it seems the number of publications increased over the years. There were only 22 publications in the analysis' first year (2011), the volume of material is constantly growing. The number of publications exceeded 100 for the first time in 2017, and a peak was seen in the year with the highest number of publication, that is 173 (10.555%) in 2022.

The included publications were cited 30,675 times. When all of the citations were subtracted from single first author cells, these numbers amounted to 22,240. Average citations per item was 18.72. Citation numbers have gradually increased. 2023 was the year with the most citations, 3,732. It dipped after 2023 instead. However, 2025 has not been finished, the data of 2025 can be neglected (**Figure 1**).

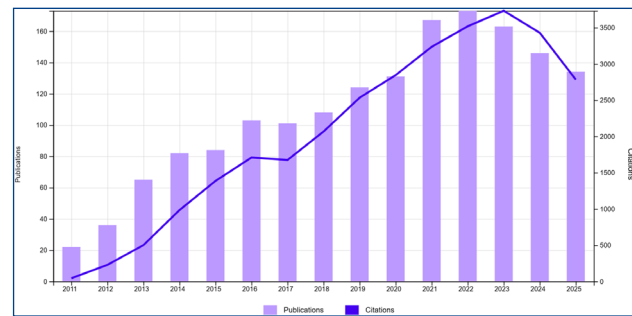


Figure 1. Times Cited and Publications Over Time

Top countries of publication and country collaboration

Global NIPT literature has apparent scope, 79 countries covering the publication. 49 of such countries had five or more published works ($n \geq 5$) on this subject. **Table 1** is a structured table on leading publication countries on NIPT. According to **Table 1** the most prolific 5 countries are: People's Republic of China ($n=384$), USA ($n=262$), England ($n=196$), Netherlands ($n=134$) and Italy ($n=88$). Abbreviations The 5 most frequent countries are: England ($n=8784$), USA ($n=7716$), People's Republic of China ($n=5569$), Netherlands ($n=4264$), Denmark ($n=1808$). Top 5 countries by total link strength (TLS) were: USA ($n=206$), England ($n=199$), Netherlands (1330), Germany (100), People's Republic of China (?82). This gives the USA an aggregate of 206 TLS units with all others of the network. This renders it the most central and connected country of the network.

Table 1. Top published countries

Country	Number of documents	Number of citations	Total link strength
People's Republic of China	384	5569	82
USA	262	7716	206
England	196	8784	199
Netherlands	134	4264	133
Italy	88	1538	85
Australia	86	1394	66
Germany	72	1289	100
Canada	69	1947	68
Belgium	52	1548	81
Denmark	51	1808	62
Spain	50	1369	85
France	46	851	58
Japan	44	395	11
Taiwan	38	199	4
Sweden	33	1338	78
Iran	32	166	12
India	29	95	2
Turkey	29	226	12
Israel	28	427	19
Switzerland	26	603	60
South Korea	25	268	3
Singapore	21	322	25
Slovakia	20	370	12
Cyprus	19	331	20
Greece	19	285	17

Also England (n=199) has a highly dense and widespread collaboration network, which is very similar to the USA. The international scientific collaboration network in the NIPT area was mapped using **Figure 2**. Inspection of the structure of the map reveals that countries with high connectivity, including the USA, England, the Netherlands, Germany and China are situated in its center; they connect to a large number of other countries and therefore act as major nodes in the network. Strong partnership clusters revolves around these principal countries have emerged. In contrast, nodes from weak-interconnected countries (e.g., Taiwan, Iran and India) appear at the map-peripheral region connected by fewer or weaker edges. This network architecture indicates that scientific collaboration within this research area is organized around a few strong hubs.

Top published institutions

8055 authors from 2430 institutions contributed to these publications.

Table 2 shows that the institutions with the highest number of documents originate from China (China Medical University and Nanjing Medical University, 36 documents each) and Australia (University of Melbourne, 35 documents). However, when citation count and total link strength metrics are considered, the picture changes; institutions such as Great Ormond Street Hospital for Children NHS Foundation Trust (1,558 citations, 37,906 link strength) and the Chinese University of Hong Kong (1,538 citations,

33,767 link strength) from the UK stand out in terms of the impact of their research outputs and their central position in the international collaboration network. Another striking finding is that institutions from the Netherlands (Erasmus MC, Vrije Universiteit Amsterdam, Maastricht University) and the UK (UCL, University of Oxford) demonstrate their active role in the network with their high citation and link strength values, despite their relatively lower document counts. **Figure 3** presents the VOSviewer institutional collaboration network map. This map visualizes various trends in global collaboration dynamics within the NIPT research field. The map is structured around several main clusters with strong connections, which are understood to reflect geographical and strategic partnerships. UK-based institutions (Great Ormond Street Hospital, University College London, Kings College Hospital) and Dutch institutions (Erasmus MC, Vrije Universiteit Amsterdam, Maastricht University) are located at the center of the network with strong connections, becoming the main hubs of international collaboration. Chinese institutions (Nanjing Medical University, Chinese University of Hong Kong, Guangzhou Medical University) have both established a dense collaboration network among themselves and are highly influential within the global network. Conversely, some institutions (such as Mackay Memorial Hospital and Asia University) are located on the periphery of the network with more limited connections, consistent with their low connection strength values.

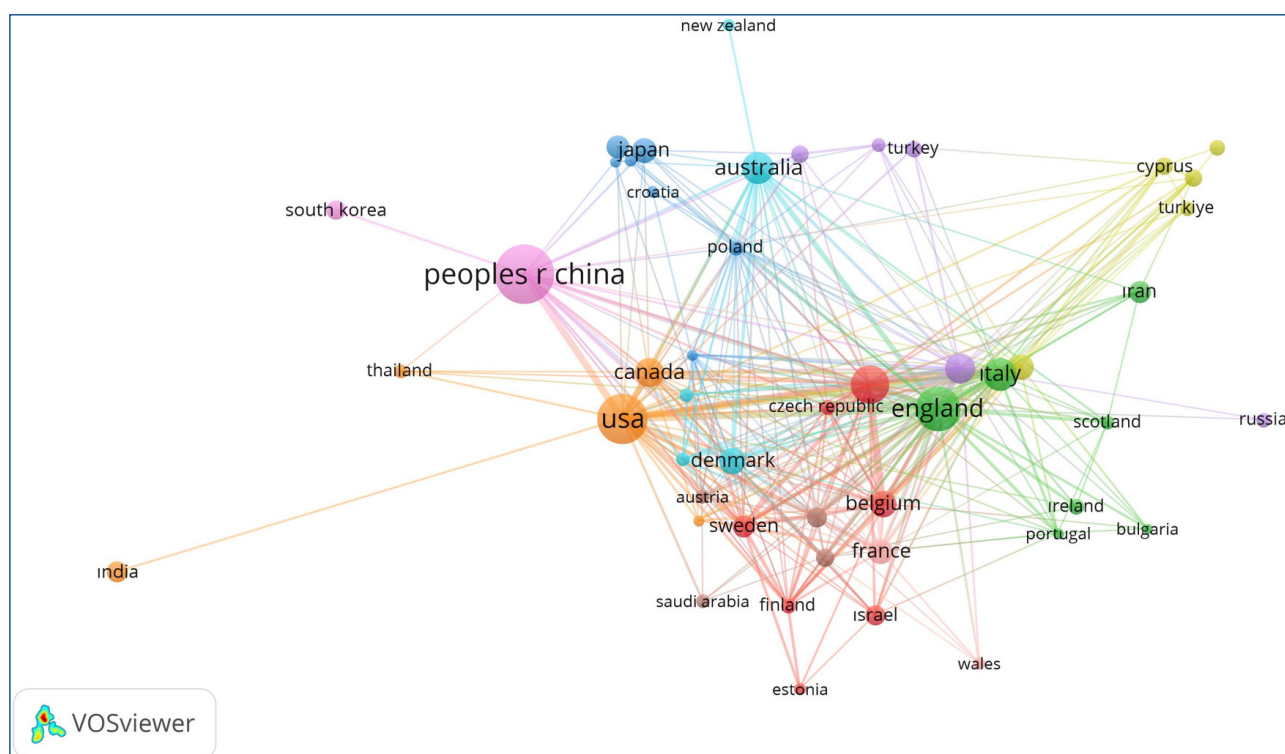


Figure 2. Top published countries

Table 2. Top published institutions

Institution	Number of documents	Number of citations	Total link strength
China Medical University (China)	36	202	7095
Nanjing Medical University (China)	36	417	23364
University of Melbourne (Australia)	35	740	28744
Erasmus Medical Center (Netherlands)	33	1082	22604
Chinese University of Hong Kong (China)	30	1538	33767
Great Ormond Street Hospital For Children NHS Foundation Trust (England)	29	1558	37906
University College London (England)	29	974	29392
Vrije Universiteit Amsterdam (Netherlands)	29	875	23633
Mackay Memorial Hospital (Taiwan)	28	129	3165
Maastricht University (Netherlands)	27	1155	27526
Murdoch Childrens Research Institute (Australia)	27	489	21778
National Yang Ming Chiao Tung University (Taiwan)	26	109	3402
Guangzhou Medical University (China)	25	347	16656
Kings College Hospital London (England)	25	2763	24074
Katholieke Universiteit Leuven (Belgium)	24	510	22048
BGI Shenzhen (China)	23	690	15779
Leiden University (Netherlands)	23	1598	20221
Monash University (Australia)	23	325	17306
University College London Institute of Child Health (England)	23	1148	36696
University of Amsterdam (Netherlands)	23	797	15608
University of Copenhagen (Denmark)	22	972	18530
University of Oxford (England)	22	597	14625
Asia University (Taiwan)	21	89	2629
Southern Medical University (China)	21	289	14850
Stanford University (USA)	20	1458	19258

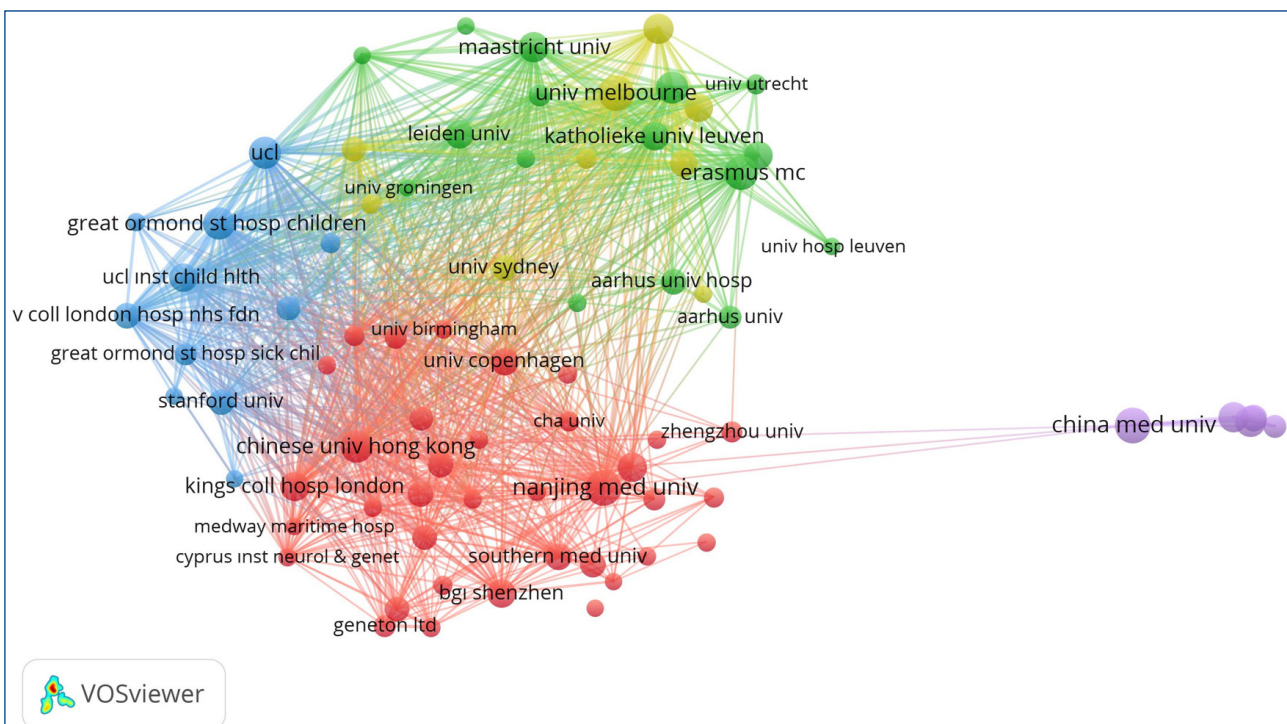


Figure 3. Top published institutions



Top published journals

These articles were published across 444 journals. **Table 3** illustrates the top journals publishing with regard to NIPT. Prenatal Diagnosis was the most productive journal in NIPT publications (highest number of publications and citations; 171; 4243). Second was Ultrasound in Obstetrics & Gynecology, which published 63 documents receiving 3623 citations, followed by PLOS One (58 and 1330). For example, the European Journal of Human Genetics (30 documents, 1082 citations) and Fetal Diagnosis and Therapy (14 documents, 530 citations) exhibited high average citation impact but were reported at relatively lower document counts. On the other hand, some of the titles with higher number of publications (e.g. Taiwanese Journal of Obstetrics & Gynecology: 37 documents, 213 citations) had lower

citation rates. Evaluating the table, it can be concluded that most publications on NIPT were published in specialized journals dealing with obstetrical and genetic topics with several having been submitted also to broad-scope and multidisciplinary high-impact journals such as PLOS One, Scientific Reports.

Keyword analysis

A total of 2,732 keywords were used in publications on NIPT. The most frequently used keywords are summarized in **Table 4**. The keyword analysis in **Table 4** provides important information about the scientific structure and focus of the NIPT research field. The most frequently used keywords, "Non-Invasive Prenatal Testing" (317 times) and "Prenatal Diagnosis" (247 times), clearly define the core topic of the field. These two terms also have the highest total link strength (799 and 605

Table 3. Top published journals on NIPT

Journal	Number of documents	Number of citations
Prenatal Diagnosis	171	4243
Ultrasound in Obstetrics & Gynecology	63	3623
PLOS One	58	1330
BMC Pregnancy and Childbirth	40	624
Taiwanese Journal of Obstetrics & Gynecology	37	213
Journal of Genetic Counseling	36	684
Frontiers in Genetics	35	272
European Journal of Obstetrics & Gynecology and Reproductive Biology	34	453
Molecular Cytogenetics	34	370
European Journal of Human Genetics	30	1082
Scientific Reports	29	366
Genes	25	232
Journal of Clinical Medicine	23	423
Journal of Maternal-Fetal & Neonatal Medicine	21	439
Archives of Gynecology and Obstetrics	20	121
Diagnostics	19	212
Journal of Obstetrics and Gynaecology Canada	17	500
Australian & New Zealand Journal of Obstetrics & Gynaecology	16	241
Journal of Obstetrics and Gynaecology	16	56
Journal of Perinatal Medicine	16	62
Best Practice & Research Clinical Obstetrics & Gynaecology	15	279
Acta Obstetrica et Gynecologica Scandinavica	14	281
BMC Medical Genomics	14	134
Clinica Chimica Acta	14	280
Fetal Diagnosis and Therapy	14	530
BJOG-An International Journal of Obstetrics and Gynaecology	13	466
Frontiers in Medicine	13	88
BMJ Open	11	376
Clinical Chemistry and Laboratory Medicine	11	131
Journal of Obstetrics and Gynaecology Research	11	83
Seminars in Fetal & Neonatal Medicine	10	303
International Journal of Molecular Sciences	10	195

times, respectively), demonstrating their centrality in the research network and their strong relationship to all other concepts. The widespread use of the acronym "NIPT" (163 times) indicates that the term has become a well-established term. Clinical outcomes such as "Down Syndrome" (100 times) and "Trisomy 21" (69 times), the main conditions screened for, along with biological components such as "Cell-Free DNA" (123 times) and "Cell-Free Fetal DNA" (85 times), form the main focus of the research agenda. The fact that invasive procedures such as "Amniocentesis" (71 times) remain a key benchmark reflects the context in which NIPT is integrated into clinical practice. The presence of terms such as "Genetic Counseling" (61 times) and "Ethics" (25 times) demonstrates that the field is not only concerned with technical aspects but also with patient care and ethical implications.

Table 4. Keyword analysis

Keyword	Number of occurrences	Total link strength
Non-Invasive Prenatal Testing	317	799
Prenatal Diagnosis	247	605
NIPT	163	469
Prenatal Screening	135	427
Cell-Free DNA	123	353
Down Syndrome	100	328
Cell-Free Fetal DNA	85	233
Aneuploidy	83	295
Non-Invasive Prenatal Testing (NIPT)	75	123
Amniocentesis	71	187
Trisomy 21	69	271
Genetic Counseling	61	165
Non-Invasive Prenatal Diagnosis	58	118
Pregnancy	49	132
Fetal Fraction	48	141
Non-Invasive Prenatal Screening	46	108
Trisomy	44	168
Noninvasive Prenatal Testing	36	89
Screening	35	124
Prenatal Testing	31	71
CfDNA	30	113
Non-Invasive Prenatal Test	29	81
Ultrasound	26	62
Ethics	25	76
CffDNA	24	64
Genetic Testing	24	65
Mosaicism	24	59
Next-Generation Sequencing	24	82
Trisomy 13	24	134
Trisomy 18	24	127
Chromosomal Microarray Analysis	22	59
Nuchal Translucency	22	65
Fetal DNA	21	53
Prenatal	21	53
Chromosomal Abnormalities	20	55
Non-Invasive	20	48

The VOSviewer keyword network map, presented in **Figure 4**, visualizes the rich conceptual structure and key focus areas of the NIPT research field. The map clearly shows that core terms such as "Non-Invasive Prenatal Testing," "Prenatal Diagnosis," and "NIPT" are located at the center of the network as the largest nodes and are connected to many other concepts by thick lines with high connection strength. Around these central terms, a strong core cluster is observed consisting of terms such as "Cell-Free DNA" and "Cell-Free Fetal DNA," which form the biological basis of the technology; "Down Syndrome," "Trisomy 21," "Trisomy 18," and "Aneuploidy," which are the main conditions screened; and "Amniocentesis," which facilitates comparison with traditional methods. The presence of nodes such as "Genetic Counseling" and "Ethics" within the network suggests that the field encompasses not only technical but also counseling and ethical dimensions. On the periphery of the map are more specific, specialized subtopics such as "Next-Generation Sequencing," "Fetal Fraction," and "Mosaicism."

The VOSviewer overlay visualization presented in **Figure 5** clearly illustrates the evolution of research focuses in the field of Non-Invasive Prenatal Testing (NIPT) between 2011 and 2025. The visualization exhibits a distinct color shift from early clusters in blue and green to more recent research trends in yellow and orange. Founding concepts such as "Non-Invasive Prenatal Diagnosis," "Cell-Free Fetal DNA," "Down Syndrome Screening," and comparisons to "Amniocentesis"—the foundations of the field—are concentrated in the cool (blue/green) regions of the map, reflecting the nascent research agenda. Over time, the research focus shifts to the central yellow/orange regions, focusing on standardized "Non-Invasive Prenatal Testing (NIPT)" terminology, routine prenatal screening practices, and mature topics such as "Genetic Counseling," and "Ethics." In particular, topics focusing on more specific, advanced technical and clinical challenges, such as "Fetal Fraction," "Mosaicism," and "Next-Generation Sequencing," appear in warmer colors (yellow), suggesting that they represent current and prominent research frontiers in the field. This temporal map summarizes the dynamic and maturing evolution of the NIPT field.

DISCUSSION

This bibliometric analysis, which aims to map scientific publication trends and the structure of knowledge in the NIPT field, when considered alongside similar previous studies, clearly reveals the dynamic and maturing nature of the field. While the results of our current study largely align with the findings of Koçbulut et al. (5) and Fan and Zhang (6), it offers a complementary and updated perspective with its focus on data source (WoS), coverage period (2011–2025), and specific focus on thematic evolution.

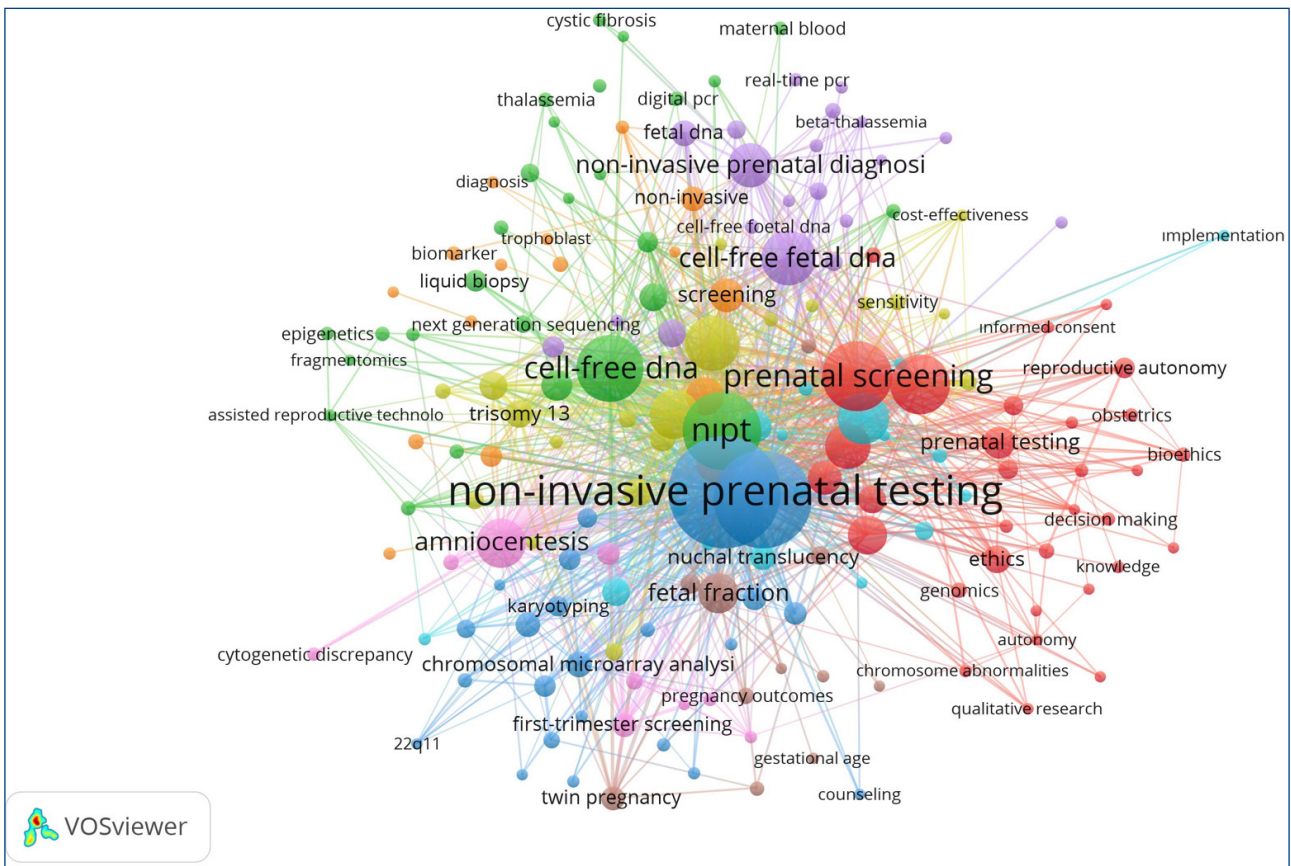


Figure 4. The keyword analysis

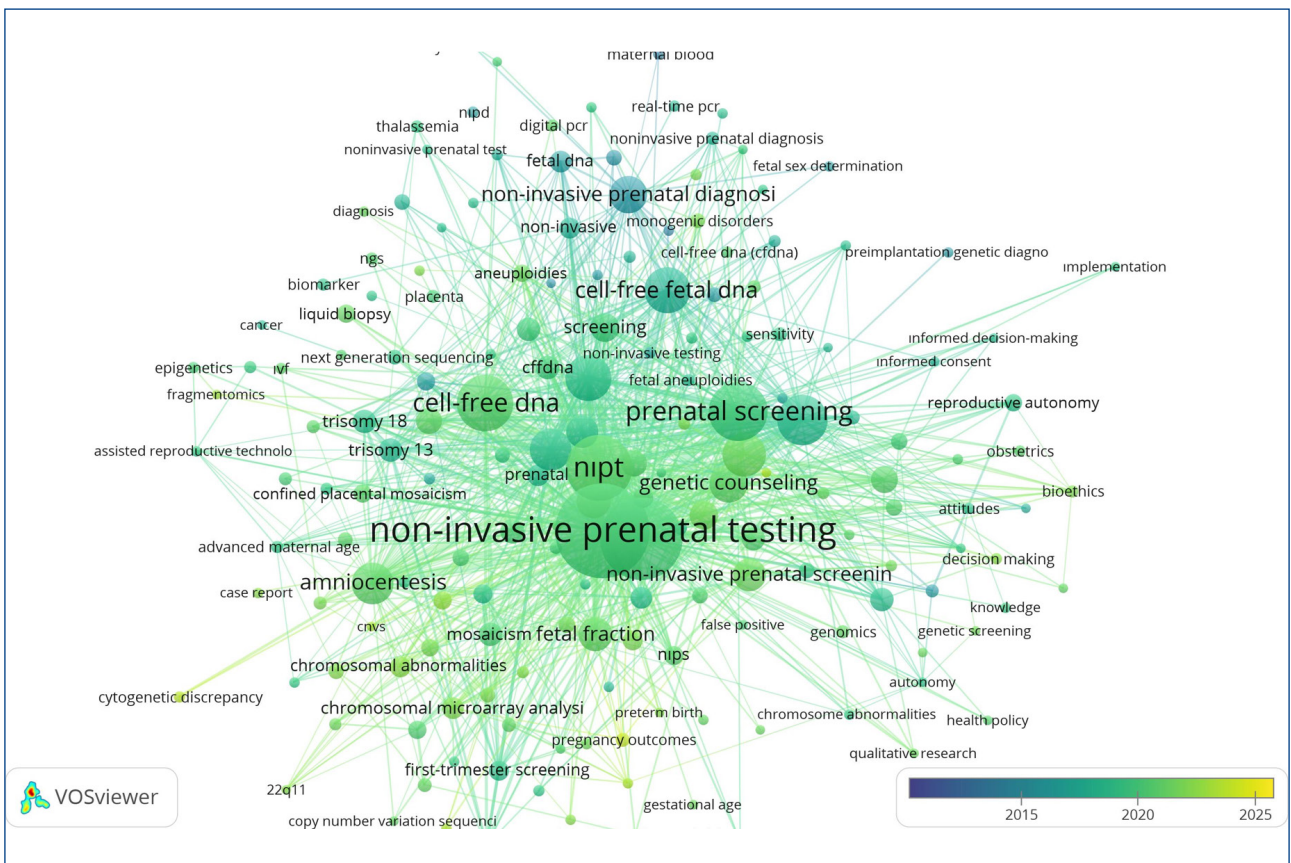


Figure 5. Overlay visualisation of keywords

This study (2011–2025) shows that the number of NIPT publications has increased steadily over the years, with the highest number recorded in 2022 (173 publications). This finding confirms the continuous increase in scientific interest since NIPT's introduction into clinical use in 2011 and is consistent with Koçbulut et al.'s (5) observation of an increase in overall cfDNA studies between 1991 and 2024. A PubMed-based study by Fan and Zhang (6) also indicated the increasing academic focus of NIPT over the past 10 years (2011–2020). These studies (5,6) and our study suggest that scientific production has increased along with the clinical acceptance of NIPT.

When examining country contributions to NIPT studies, similar to previous studies (5,6), our study also identified a global hierarchy. While Koçbulut et al. (5) listed the USA, China, and the UK among the top three contributing countries, our current study identified China (n=384), the USA (n=262), and the UK (n=196) as the top three contributing countries. This suggests that China has solidified its leadership in the number of publications, particularly with the inclusion of data from 2022 onward.

Limitations

Although this bibliometric analysis presents a very complete view of the research field on NIPT, some limitations have to be acknowledged when interpreting the results. First is the data source that was only limited to the Web of Science (WoS) Core Collection, which although a prestigious database could have excluded some important relevant articles indexed in other databases such as Scopus, PubMed, and regional databases and might have introduced a selection bias. Second, the fact that a comprehensive search strategy was adopted using pre-defined keywords means it may not have captured all nuances and emerging terminologies in NIPT which is a rapidly evolving field condition even that this might potentially affect some relevant publications to be missed. Third, only English articles and reviews were included in this analysis; research published in other languages and by non-English-speaking countries were not taken into account, so the findings may not entirely represent the worldwide endeavor on NIPT testing but more likely part of it. In addition, the bibliometric method relies mainly on publication metrics (number of citations and co-author patterns) to assess research quality but not on the qualitative nature or clinical impact of the work; and citation analysis may underestimate newer high quality studies by inherently favoring meanest older articles that have had a longer time period in which they could rack up more citations.

CONCLUSION

Scientific landscape The present bibliometric analysis gives a global overview of the scientific production on NIPT research from 2011 to 2025. The study shows how the NIPT field has developed into a mature and active research area, with robust growth in publications and strong international collaborations. China is the main leader in terms of publication counts for the NIPT literature, although influence and research network are mostly driven by the United States and UK. The majority of the field's literature is already in specialist obstetrical, gynecological and genetic journals with Prenatal Diagnosis now as the chief method of communication. Thematic evolution analysis obviously shows that the research context has progressed from the foundational investigation focusing on basic technology and common aneuploidy screening to a maturing and complex period dealing with technological issues, including FF (fetal fraction), mosaicism, genetic counselling, ethical problems etc.

In contrast, the TLS studies of Koçbulut et al. (5) as well as the present study clearly demonstrated, that USA and UK possess the densest international collaboration networks and act as global research hubs. This indicates that well-counting researcher quantity in China is published while research influence of the USA and UK fails to form, international cooperation has evolved but its concentration level is high. Zhang et al. citation-affiliated report (7) corroborated this historic trend as authors from the USA, UK, Hong Kong and China collaborated most frequently within their working group with respective coauthors in the highest cited papers.

According to current study the institutions that contribute most to and have the greatest impact on NIPT research: While institutions in the People's Republic of China (Chinese University of Hong Kong, China Medical University) lead with their high publication volume, institutions in the USA, UK and the Netherlands (Great Ormond Street Hospital, UCL, Erasmus MC) are at the centre in terms of the number of citations and international collaboration networks (Total Link Strength - TLS), representing the quality and global integration of research. In the study of Koçbulut et al. (5), the Chinese University of Hong Kong came to the fore in terms of the number of publications and citations.

In terms of publication platforms, the journal "Prenatal Diagnosis" was identified as the journal with the highest number of publications in both our current study (171 publications) and Koçbulut et al.'s study (5) (269 publications). This demonstrates that the journal is unquestionably the primary specialized platform for NIPT/cfDNA research. However, when we look at the list of most cited articles by Zhang et al. (7), the leadership of general, multidisciplinary, high-impact journals such



as the New England Journal of Medicine and Lancet is striking. This suggests that while basic NIPT research tends to be published in specialized journals, the field's most groundbreaking and influential (highly cited) studies are published in broad-based, high-impact journals.

Keyword analysis is the most striking indicator of scientific progress in a research field (8,9). Fan and Zhang (6) identified seven major research hotspots, while Koçbulut et al. (5) found that cfDNA-based screening primarily focuses on chromosome aberrations. The VOSviewer layout visualization in our current study (**Figure 5**) clearly illustrates how these themes have evolved. The most striking finding in our study is thematic evolution. The focus of the initial publications (Down Syndrome, basic cfDNA technology) has shifted to more complex technical, clinical, and social dimensions such as "Fetal Fraction," "Mosaicism," "Genetic Counseling," and "Ethics," indicating that the field of NIPT has moved from fundamental principles to implementation and standardization.

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