

Jurnal Info Kesehatan

Vol. 24, No. 3, Sept 2026, pp. 532-539

P-ISSN 0216-504X, E-ISSN 2620-536X

DOI: [10.31965/infokes.Vol24.Iss3.2552](https://doi.org/10.31965/infokes.Vol24.Iss3.2552)

Journal homepage: <https://jurnal.poltekkeskupang.ac.id/index.php/infokes>



RESEARCH

Open Access

Determinant Factors Influencing the Diagnosis of Children with Down Syndrome in Central Java, Indonesia

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Received: 15 May 2026

Revised: 10 June 2026

Accepted: 5 August 2026

Abstract

The prevalence of Down syndrome among children aged 24–59 months was detected at 0.12% in 2010, 0.13% in 2013, and 0.21% in 2018, or approximately 1 per 600 live births. The global incidence is estimated at 3,000–5,000 infants born with Down syndrome each year. Based on CDC data, approximately 6,000 infants are born with Down syndrome (DS) each year, or about 1 in 700 live births. The incidence of DS in the United States (US) increased over the period 2010–2014, rising from 13.7 per 10,000 to 15.7 per 10,000 live births, or 1 in 772 infants born with DS. Early detection of Down syndrome plays a crucial role in improving clinical management and family preparedness. Delayed diagnosis of intellectual disability can cause children to miss the opportunity for early intervention, which can help reduce symptoms as well as behavioral and emotional problems, in addition to resulting in poorer cognitive and adaptive functioning compared with those who receive treatment early. This study aimed to describe the characteristics of children with Down syndrome, maternal characteristics, and the history of birth attendants, birth settings, and modes of delivery in Central Java, Indonesia. Specifically, the study aimed to examine the associations between birth attendant, birth setting, mode of delivery, and the timing of Down syndrome diagnosis in children. A correlational analytical design with a cross-sectional approach was employed to describe the characteristics of children with Down syndrome, maternal characteristics, and birth-related factors, including birth attendant, birth setting, and mode of delivery. The study also examined the associations between these factors and the child's age at Down syndrome diagnosis. Bivariate analysis was performed using the Chi-square test, while multivariable analysis was conducted using binary logistic regression. The findings showed that the majority of children were male and were aged >5–12 years. Most mothers were aged 20–35 years at the time of delivery. Midwives were the most common birth attendants, spontaneous vaginal delivery was the predominant mode of delivery, and independent midwifery practices were the most frequent birth settings. Most children were diagnosed with Down syndrome during the neonatal period. Among the factors examined, birth attendant showed the strongest statistical association with early diagnosis.

Keywords: Down syndrome; Diagnosis; determinant factors.

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1. INTRODUCTION

Down syndrome (DS) is caused by an additional copy of chromosome 21 and is commonly associated with intellectual disability, distinctive physical features, and a range of medical conditions. Because its effects extend across growth, cognition, health, and daily functioning, DS remains an important concern for child health services worldwide (Chisolm et al., 2020). Improvements in medical care have increased survival, but optimal outcomes still depend on continued clinical monitoring, developmental intervention, and sustained family support (Chen et al., 2022).

Down syndrome is one of the most common chromosomal conditions identified in newborns. The Centers for Disease Control and Prevention estimates that approximately 6,000 affected infants are born each year in the United States, equivalent to about 1 in every 700 live births. Reported U.S. incidence rose from 13.7 to 15.7 per 10,000 live births between 2010 and 2014, representing approximately 1 affected birth per 772 live births (Graaf et al., 2021).

Comparable national incidence data are not available for Indonesia. Available estimates are primarily based on prevalence data from the Basic Health Research Survey, which reported DS prevalence among children aged 24–59 months of 0.12% in 2010, 0.13% in 2013, and 0.21% in 2018 (Kementerian Kesehatan Republik Indonesia, 2018).

Maternal age is one of the factors associated with the occurrence of Down syndrome. The risk of a pregnancy affected by trisomy 21 increases with advancing maternal age, particularly after the age of 35 years (Owen et al., 2022). This is clinically important because maternal age at childbirth has tended to increase in many settings. Therefore, appropriate prenatal risk assessment and screening are essential for the early detection of Down syndrome.

Prenatal screening for Down syndrome can be performed using various non-invasive methods at different stages of pregnancy. During the first trimester, screening may include the measurement of *nuchal translucency* and the assessment of maternal serum markers. In the second trimester, screening may involve biochemical tests, such as *triple screening* or *quadruple screening* (Malone et al., 2005). If screening results indicate an increased risk of Down syndrome, further diagnostic testing may be considered, including chorionic villus sampling, amniocentesis, or, in selected cases, percutaneous umbilical blood sampling. Maternal age, pregnancy history, and relevant family history may also be taken into account when determining the most appropriate diagnostic approach (Irwanto I, 2019).

Early detection of Down syndrome is important for supporting appropriate clinical management and helping families prepare for the child's needs. Advances in prenatal screening, including *non-invasive prenatal testing*, together with postnatal clinical assessment, have increased opportunities for earlier diagnosis and timely counselling (Jayashankar et al., 2023). Timely diagnosis may facilitate earlier access to rehabilitation services, including physiotherapy, speech therapy, and occupational therapy, thereby supporting the developmental needs of children with DS (Hu et al., 2020).

The timing of diagnosis is shaped not only by clinical characteristics but also by the health-care setting in which birth occurs. Birth attendants may be the first health professionals to identify phenotypic features associated with DS and initiate further assessment. Delivery setting can also influence access to trained personnel, diagnostic services, and referral pathways. Hospital births may therefore offer more immediate opportunities for recognition and assessment than births in facilities with limited diagnostic resources (Aprigio et al., 2023).

For service planning, it is therefore important to examine child characteristics together with maternal and birth-related factors. In Indonesia, however, evidence integrating these elements remains limited, and studies often address maternal risk factors or clinical characteristics separately (Musdalipa & Maddeppungeng, 2021). An integrated approach that

considers multiple factors may provide a more comprehensive understanding of the relationships among maternal age, birth attendant, and the timing of Down syndrome diagnosis, both in community-based health services and referral facilities (Utari et al., 2024a).

Previous studies have shown that maternal factors and characteristics of the health care system play an important role in the detection and management of children with Down syndrome. Increasing maternal age, particularly beyond 35 years, has consistently been reported to be associated with the occurrence of Down syndrome. In contrast, delays in diagnosis may occur when awareness of Down syndrome remains limited, access to specialist health professionals is inadequate, or referral mechanisms between primary care and higher-level health services are not yet functioning optimally. Differences in health education and family support can further affect how parents understand and respond to the diagnosis (Zevanya et al., 2024). Timely identification is particularly relevant because delayed recognition of intellectual and developmental disabilities can postpone access to interventions that support cognitive, adaptive, behavioral, and emotional functioning (Loubersac et al., 2023; Montiel-Nava et al., 2017).

Evidence from Central Java that links the characteristics of children with DS, maternal characteristics, and birth history is still limited. This study therefore described child and maternal characteristics together with birth attendant, birth setting, and mode of delivery, and examined their associations with the timing of DS diagnosis. The findings were intended to provide locally relevant information for early detection strategies, maternal and child health education, and referral practices for children with DS and their families.

2. RESEARCH METHOD

A correlational analytic study with a cross-sectional design was used to characterize children with DS and their mothers and to examine whether selected birth-related factors were associated with age at diagnosis in Central Java, Indonesia. This study design was used to describe the characteristics of the variables under investigation and to examine the associations between birth setting, birth attendant, mode of delivery, and the child's age at the time of Down syndrome diagnosis within the study population.

The study population comprised mothers of children with Down syndrome who were registered with the Indonesian Association of Parents of Children with Down Syndrome (POTADS) in Central Java. A total of 200 respondents participated in the study. Respondents were eligible if they had a child who had been diagnosed with Down syndrome, provided informed consent to participate, and completed all sections of the questionnaire.

The variables examined included child characteristics, namely sex, age, and age at Down syndrome diagnosis, as well as the maternal characteristic of age at delivery. Birth setting, birth attendant, and mode of delivery were treated as independent variables, whereas the child's age at Down syndrome diagnosis was considered the dependent variable. Data were collected using a questionnaire developed based on the study variables and distributed to respondents via Google Forms.

The collected data were subsequently analysed using IBM SPSS Statistics version 22.0. Participant and study characteristics were summarized using frequencies and percentages. Associations between each birth-related factor and age at diagnosis were examined with chi-square tests. Variables that met the criteria for further analysis were then entered into a binary logistic regression model to identify factors independently associated with the timing of diagnosis.

The Health Research Ethics Committee of the Faculty of Medicine, Universitas Islam Sultan Agung approved the study (ethical clearance no. 340/VIII/2024/Komisi Bioetik).

Participants received information about the study purpose and procedures before providing written informed consent. Confidentiality of participant information was maintained throughout data collection and analysis.

3. RESULTS AND DISCUSSION

Table 1. Demographic Characteristics of Children with Down Syndrome and Maternal Age at Delivery

Variable	Frequency	Percent
Gender		
Male	112	56.0
Female	88	44.0
Total	200	100.0
Age Group		
0–2 years	36	18.0
>2–5 years	63	31.5
>5–12 years	80	40.0
>12 years	21	10.5
Total	200	100.0
Maternal Age		
<20 years	5	2.5
20–35 years	104	52.0
>35 years	91	45.5
Total	200	100.0

Table 1 summarizes the demographic profile of the 200 children with DS and maternal age at delivery. Boys comprised 56.0% of the sample and girls 44.0%. The most represented age category was >5-12 years (40.0%), followed by >2-5 years (31.5%), 0-2 years (18.0%), and >12 years (10.5%). At the time of delivery, 52.0% of mothers were aged 20-35 years, 45.5% were older than 35 years, and 2.5% were younger than 20 years.

Table 2. Birth History

Birth Attendant	Frequency	Percent
Midwife	114	57,0
Obstetrician	86	43,0
Total	200	100,0
Place of Delivery		
Midwifery Clinic	98	49,0
Primary Healthcare Center	27	13,5
Hospital	75	37,5
Total	200	100.0
Type of delivery		
Spontaneous	172	86,0
SC	28	14,0
Total	200	100.0
Age of Down syndrome diagnosis		
0-28	126	63,0
>28 day	74	37,0
Total	200	100.0

As shown in Table 2, most deliveries were assisted by a midwife (57.0%), occurred in an independent midwife practice (49.0%), and were spontaneous (86.0%). Overall, 63.0% of children received a DS diagnosis within the first 28 days of life, while 37.0% were diagnosed after the neonatal period.

Table 3. The Association Between Birth Setting, Birth Attendant, and Delivery Method with Age at Diagnosis of Down Syndrome

Variabel	Age of Down syndrome diagnosis				Total	pvalue	Coefisien
	0-28 day		>28 day				
	f	%	f	%			
Birth Attendant							
Midwife	48	42,1	66	57,9	114	,000	,521
Obstetrician	78	90.7	8	9,3	86		
Place of Delivery							
Midwifery	38	38,8	60	61,2	98		
Clinic	18	66,7	9	3,3	27		
Primary	70	93,3	5	6,7	75	,000	,490
Healthcare Center							
Hospital							
Type of delivery							
Spontaneous	106	61,6	66	38,4	172		
SC	20	71,4	8	28,6	28	,000	,367
Total	126	63.0	74	37.0	200		

Among the 200 children, 126 (63.0%) were diagnosed at 0-28 days of age and 74 (37.0%) after 28 days. The chi-square analyses showed statistically significant associations between age at diagnosis and each of the three birth-related variables examined: birth attendant, birth setting, and mode of delivery ($p = 0.000$ for each comparison; Table 3).

The distribution of early diagnosis differed markedly across birth attendants and delivery settings. Among children delivered by obstetricians, 78 of 86 (90.7%) were diagnosed during the neonatal period, compared with 48 of 114 (42.1%) among those delivered by midwives. Early diagnosis was also most frequent among hospital births (70/75; 93.3%), followed by births in community health centers (18/27; 66.7%) and independent midwife practices (38/98; 38.8%). By mode of delivery, 20 of 28 children born by cesarean section (71.4%) and 106 of 172 born vaginally (61.6%) were diagnosed within 28 days.

Table 4. Determinant Factors Associated with Age at Diagnosis of Down Syndrome

Factors	B	S.E	Wald	Sig	Exp (B)
Birth Attendant	5,097	,841	36,750	,000	163,518
Place of Delivery	4,403	,849	26,872	,000	81,694
Type of delivery	3,528	,658	28,770	,000	34,052

In the multivariable analysis, birth attendant produced the largest Wald statistic (36.750) and Exp(B) value (163.518), followed by birth setting (Wald = 26.872; Exp(B) = 81.694) and mode of delivery (Wald = 28.770; Exp(B) = 34.052). Within the fitted model, birth attendant therefore showed the strongest statistical association with early diagnosis, with birth setting ranking next and delivery method showing the smallest Exp(B) among the three variables.

DISCUSSION

Boys represented a slightly larger proportion of the sample than girls (56.0% versus 44.0%). DS results from trisomy 21 rather than an abnormality of the sex chromosomes, so this difference should not be interpreted as evidence of a sex-specific biological cause. Similar modest male predominance has been described in epidemiological observations, although reported differences are generally small and may reflect survival, registration practices, or population structure rather than differential susceptibility by Cassidy et al., (2021).

Children aged >5-12 years formed the largest age group in this study (40.0%). One possible explanation is the substantial improvement in survival and longer-term care of children with DS. Progress in pediatric services, including management of congenital heart disease, infection prevention, nutritional care, and developmental support, has increased the proportion of affected children who survive into school age and adolescence. These findings are consistent with [Ye et al., \(2025\)](#)., similarly reported that improvements in healthcare have contributed to longer life expectancy among people with DS ([Ye et al., 2025](#)).

Maternal age at delivery showed a broad distribution: 52.0% of mothers were aged 20-35 years and 45.5% were older than 35 years. The large absolute number of births among women aged 20-35 years is compatible with the fact that most births in the population occur in this age range, whereas the individual probability of DS rises substantially with advancing maternal age. Age-related meiotic nondisjunction provides the established biological basis for this association. ([Song et al., 2022](#)) identified maternal age as a consistent risk factor for trisomy 21 across populations, while findings from Central Java have also emphasized the relevance of advanced maternal age for prenatal risk assessment and screening ([Utari et al., 2024a](#)). Risk factors reported in relation to DS have also included folic acid deficiency.

The relationship between advanced maternal age and DS has been documented in earlier reviews ([Coppedè, 2016](#)). In a Brazilian hospital-based study, 72.07% of infants with DS were born to mothers older than 35 years and 83.93% were delivered by cesarean section; prenatal diagnosis was reported in 27.68% of cases despite delivery in referral hospitals with NICU facilities ([Alfaro et al., 2024](#)). More recently, anti-zona pellucida antibodies were proposed as an age-independent risk factor, although most DS cases in that study still occurred among mothers older than 35 years (age-adjusted OR = 71.52) ([Noia et al., 2026](#)). In the present sample, the 45.5% proportion of mothers aged >35 years is therefore compatible with the broader literature showing the continuing importance of maternal age, even when additional mechanisms are considered ([Noia et al., 2026](#)).

The birth-history profile showed that deliveries were most often assisted by midwives, occurred in independent midwife practices, and were spontaneous; nevertheless, most children were diagnosed during the neonatal period. Birth attendants are positioned to notice unusual physical findings immediately after birth and can initiate referral or diagnostic evaluation when DS is suspected ([Olorunsaiye et al., 2020](#)). Evidence from newborn care research also indicates that skilled attendance is associated with better clinical assessment and neonatal outcomes, while facility-based care may improve opportunities for prompt identification and management of congenital conditions ([Baruwa et al., 2021](#)). The provider and facility involved in delivery may therefore influence how quickly DS is recognized. Hospitals with specialist personnel and broader diagnostic capacity can support earlier assessment than settings with more limited resources ([Tenorio D et al., 2025](#)).

The pattern observed across delivery settings is consistent with the practical advantages of facilities that provide more comprehensive neonatal services. Hospitals typically offer quicker access to pediatric or neonatal assessment, diagnostic investigations, and structured referral processes, which can facilitate evaluation of newborns with phenotypic features suggestive of DS ([Anane-Fenin et al., 2023](#)). Central Java data have likewise documented delayed identification in settings where access to specialized services is limited, pointing to the importance of strengthening neonatal assessment and referral pathways ([Utari et al., 2024a](#)).

The high proportion of early diagnoses among hospital-born children can plausibly be related to routine newborn examination and the immediate availability of clinicians with experience in recognizing congenital conditions. Characteristic physical findings may raise suspicion during the neonatal period and prompt confirmatory testing or referral. By contrast, lower-level facilities may have fewer specialist staff and more restricted access to diagnostic support, increasing the chance that recognition will be deferred until after discharge or a subsequent healthcare contact ([Monier et al., 2023](#)).

The study by (Alnoman et al., 2024) using a population-based database, examined pregnancy and neonatal outcomes among mothers with DS and emphasized the need for enhanced antenatal surveillance in the context of limited information on the timing of chromosomal diagnoses. In the present study, 63.0% of children were diagnosed during the first 28 days, and early diagnosis was substantially more frequent when delivery was attended by an obstetrician or occurred in a hospital. Although the populations and research questions differ, both sets of findings draw attention to the importance of access to services with greater capacity for surveillance and recognition. The present data further show that birth attendant, birth setting, and delivery method were each statistically associated with age at diagnosis ($p = 0.000$). Infants born in hospitals and attended by obstetricians were more often identified during the neonatal period, whereas delayed diagnosis was more common after births in independent midwife practices and births attended by midwives (Utari et al., 2024b).

All three birth-related variables remained associated with the timing of diagnosis at the bivariate level ($p < 0.05$). Early diagnosis was especially frequent among hospital births and births attended by obstetricians. Highlighted the value of integrated healthcare and collaboration across providers and medical specialties in the management of children with DS. The present findings add a service-delivery perspective by suggesting that the setting and professional context at birth may shape how quickly a child enters the diagnostic and referral pathway (Krell et al., 2026).

Clinical follow-up after birth is particularly important because newborns with DS may require assessment for associated conditions such as congenital heart disease, pulmonary hypertension, airway problems, and feeding intolerance. Earlier recognition creates an opportunity for these concerns to be investigated and managed without unnecessary delay (Muhammad Zain et al., 2025). Delayed diagnosis is also encountered in other developmental conditions; an exploratory retrospective study reported a mean diagnostic age of 29.9 months for autism spectrum disorder and 6.8 years for intellectual disability (Noia et al., 2026).

Other research has reported that many DS diagnoses are made postnatally, with a median diagnostic age of 0 days, reinforcing the neonatal period as a key window for clinical recognition. When characteristic features are identified soon after birth, cytogenetic confirmation can be arranged where indicated (Ammar et al., 2024) did not specifically evaluate birth attendant, birth setting, or delivery method as predictors of diagnostic timing; however, the current results show that hospital delivery and obstetric attendance were associated with a higher proportion of early diagnoses. Greater access to specialist assessment, more comprehensive newborn examination, and faster availability of supporting investigations may contribute to this difference.

From a healthcare service perspective, the findings of this study highlight the importance of strengthening the capacity of primary healthcare facilities and independent midwifery practices to recognise clinical features suggestive of Down syndrome and to initiate timely referrals. Enhancing healthcare professionals' competencies through training in neonatal assessment, establishing clear referral pathways, and improving coordination between primary care and referral facilities may help accelerate the diagnostic process after birth. Earlier diagnosis may, in turn, facilitate more timely interventions and services tailored to the needs of affected children.

Nevertheless, the statistical findings of this study should be interpreted with caution because several cells had expected counts of fewer than five. This limitation may affect the reliability of the statistical tests and should therefore be taken into consideration when interpreting the associations between the study variables.

4. CONCLUSION

Most children in this study were male and the largest age group was >5-12 years. More than half of the mothers were 20-35 years old at delivery, although a substantial proportion were older than 35 years. Midwives were the most common birth attendants, independent midwife practices were the most frequent delivery settings, and spontaneous delivery predominated. The majority of children in this study were diagnosed with Down syndrome during the neonatal period, accounting for nearly two-thirds of all participants. Age at diagnosis was associated with birth attendant, place of delivery, and mode of delivery. Earlier diagnosis was more frequently observed among children who were born in hospitals and whose deliveries were attended by obstetricians.

Multivariable analysis showed that birth attendant had the strongest statistical association with early diagnosis, followed by place of delivery and mode of delivery. These findings highlight the importance of healthcare professionals' ability to recognise clinical features suggestive of Down syndrome from birth, as well as the availability of healthcare facilities with adequate diagnostic resources and effective referral systems.

Based on these findings, strengthening competencies in newborn assessment and improving the effectiveness of referral pathways should be prioritised across maternity and primary healthcare services to support earlier diagnosis of Down syndrome.

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