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Cost analysis of financial, productivity, and informal care burdens in families raising children with congenital anomalies

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Abstract

Background Congenital anomalies represent a major cause of childhood morbidity and disability, imposing long-term health, social, and financial challenges. While medical expenditures are relatively well-documented, evidence on the broader financial and social impact on families—particularly in Central and Eastern Europe—remains limited. Accordingly, this study aimed to quantify the economic burden associated with congenital anomalies from a caregiver perspective.

Methods A cross-sectional, questionnaire-based study was conducted in Lithuania in 2023 among 160 caregivers of children aged 5–18 years diagnosed with congenital anomalies. Participants were grouped by the number of affected organ systems to reflect the increasing disease complexity. Using standardized cost-of-illness methodology and both the human capital and opportunity cost approaches, we estimated annual direct medical and non-medical, indirect, and informal care costs.

Results Families experienced a substantial and multidimensional financial burden that intensified with disease severity. The annual indirect cost, mainly reflecting productivity losses, represented the largest component of the total economic burden, with a median of €7,566 overall and €12,965 in the most severely affected families. Out-of-pocket payments were also considerable, with a median of €2,040 per year; in the most severe group they exceeded 20% of monthly household income for 42% of families. The economic value of informal care represented a smaller yet meaningful portion of total costs (median €1,967), increasing more than fivefold between the mildest and most severe groups.

Conclusions Congenital anomalies place a substantial economic burden on families, extending far beyond healthcare expenses. These findings highlight the need for policies that improve financial protection, support caregivers' labor market participation, and acknowledge the economic value of informal care within health and welfare systems.

Key points

• Families raising children with congenital anomalies in Lithuania face substantial financial, productivity, and informal caregiving burdens, which increase sharply with condition severity.

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• The largest pressures arise from reduced work participation, high out-of-pocket expenses, and extensive unpaid caregiving time.

• These findings highlight the need for stronger financial protection and caregiver support, particularly for families of children with complex or multisystem anomalies.

Introduction

Congenital anomalies, also referred to as congenital malformations, are structural or functional abnormalities that arise during intrauterine development and are present at birth. These anomalies may affect a single organ system or involve multiple systems simultaneously, ranging from minor morphological differences to severe, life-threatening disorders [1]. Globally, congenital anomalies occur in approximately 3–6% of all live births, depending on surveillance methods and geographic region [2, 3]. They are a leading cause of perinatal mortality, accounting for an estimated 240,000 neonatal deaths annually worldwide, and represent a major contributor to long-term disability, chronic health conditions, and reduced quality of life [1, 4]. Due to advances in prenatal diagnostics, neonatal intensive care, and early therapeutic interventions, long-term survival among children with significant congenital anomalies has improved markedly over recent decades. This trend is expected to continue, particularly in high-income settings, where comprehensive perinatal care and multidisciplinary follow-up are more widely accessible [5, 6]. Nevertheless, surviving children often require ongoing support in the form of medical treatment, special education, assistive devices, and coordinated social services. Consequently, the impact of congenital anomalies extends beyond health outcomes to also place substantial emotional and social strain on families, affecting their daily lives and overall well-being.

Beyond these psychosocial consequences, congenital anomalies also impose a considerable economic burden. Families frequently face substantial direct medical and non-medical costs, including transportation to specialized care centers, expenditures for assistive technologies, payments for therapies not covered by insurance, special education services, and home or vehicle modifications to accommodate the child's needs [7, 8]. These expenses often accumulate over time and are particularly burdensome for families of children with severe or multiple anomalies. In addition, indirect costs—such as reduced work hours, career interruptions, or even permanent job loss among caregivers—represent a significant but often underreported component of the overall burden [9]. These productivity losses may lead to long-term financial insecurity, particularly when public support systems are insufficient to meet the needs of affected families.

Although cost-of-illness (COI) studies are intended to account for all components of the economic burden—namely, direct and indirect costs—data availability and methodological constraints often lead to a predominant focus on direct medical expenditures. This leads to an underestimation of the broader economic and social burden associated with congenital anomalies, particularly in light of the sustained care needs of children with disabilities. The growing recognition of this gap has led to calls for a more comprehensive assessment of illness-related costs, including the adoption of standardized methods to quantify and value both informal care and productivity losses [10, 11].

To better understand the economic impact on families, we conducted a cross-sectional study in Lithuania focusing on the costs incurred by households raising children aged 5–18 diagnosed with congenital anomalies. Specifically, we aimed to quantify both direct medical and non-medical, as well as indirect costs resulting from productivity losses among caregivers, and the economic value of informal caregiving.

Our study contributes to the limited body of evidence from Central and Eastern Europe on the socioeconomic consequences of congenital anomalies. By quantifying out-of-pocket and productivity-related costs from the caregiver perspective, we aim to inform health policy decisions, social support planning, and future cost-effectiveness analyses involving this population.

Methods

Study design

A cross-sectional, questionnaire-based study was conducted in Lithuania in 2023 to assess the burden of care among families raising children with congenital anomalies, focusing on direct medical and non-medical, indirect, and informal care costs. The study focused on caregivers—predominantly parents—of children aged 5 to 18 years with confirmed diagnoses of at least one congenital anomaly, classified according to ICD-10 codes Q00–Q99. The study population was limited to families with children aged five years and older to ensure that participants had passed the initial adaptation period and to allow a more reliable assessment of longer-term economic and social effects, as parental employment is often reduced in the early years regardless of child health. A total of 160 caregivers participated in the study. Participants were recruited during routine hospital visits and through national patient organizations supporting families affected by rare or congenital conditions.

Lithuania operates a tax-funded, universal health coverage system in which most essential healthcare services for children are publicly financed; however, coverage of long-term care and caregiver support services remains limited.

Data were collected using a structured questionnaire adapted from the standardized instrument developed by Landfeldt et al. [11], which integrates the human capital and opportunity cost approaches for evaluating productivity losses and informal care. The Lithuanian version preserved the conceptual structure of the original while incorporating nationally specific terminology and the features of Lithuania's healthcare and social support systems. Adaptations included classifications of disability status, caregiving dependency levels, educational support needs, and regional access to care. Additionally, a range of descriptive data was collected, including, but not limited to, respondents' age, place of residence, educational attainment, employment status, and household composition. The final version of the questionnaire is provided as a supplementary material 1.

To better reflect differences in caregiving burden, participants were categorized into three groups based on the severity of the child's condition, defined by the number of affected organ systems. Diagnostic information used for this classification was obtained from official medical records in the national Electronic Health Services and Cooperation Infrastructure Information System (ESPBI IS) and was not self-reported by caregivers. All diagnoses had been established and recorded by medical specialists and coded according to ICD-10. Medical records were first reviewed to confirm the presence of at least one congenital anomaly diagnosis within ICD-10 chapter Q00–Q99. For each eligible child, all recorded congenital anomaly diagnoses were then extracted from the medical records. The number of affected organ systems was subsequently determined using ICD-10 congenital anomaly subgroups, which are inherently organized by organ system involvement (e.g. nervous, circulatory, musculoskeletal systems, chromosomal abnormalities), as defined within the ICD-10 classification framework. Group I consisted of children with congenital anomalies affecting a single organ system. Group II included those with anomalies involving two organ systems. Group III comprised children with three or more affected organ systems or diagnosed with chromosomal disorders, which typically involve multisystem impairment. This classification served as a proxy for clinical complexity, exploring differences in caregiving time, economic burden, and family impact across severity levels.

Costs were identified according to standard cost-of-illness methodology, encompassing direct, indirect, and informal (caregiver) components [12–16].

Estimation of direct medical and non-medical costs

Direct medical and non-medical costs were estimated as out-of-pocket expenditures, defined as all household payments directly related to the treatment, care, and management of the child's health condition and not reimbursed under national healthcare or social insurance schemes. These included direct medical expenses (e.g., medications, specialist consultations, diagnostic tests) and direct non-medical expenses (e.g., transportation to healthcare facilities, accommodation during treatment, therapy sessions, educational or rehabilitation services, and home care supplies). Caregivers reported these expenditures on a monthly basis, and annual estimates were obtained by multiplying monthly values by twelve.

Estimation of indirect costs

Indirect costs were defined as productivity losses resulting from the impact of caregiving on the caregiver's ability to engage in paid employment. They were estimated using the human capital approach, following the methodology described by Landfeldt et al. [11]. In this approach, productivity loss is quantified as the number of work hours lost, valued at the national average wage, including employer-paid social contributions, which reflects the opportunity cost of labor [12]. Respondents provided data on their typical weekly workload, the number of hours missed due to caregiving, and their perceived reduction in work efficiency.

Productivity losses were calculated in three ways: (1) by estimating the annual number of work hours missed due to caregiving; (2) by adjusting the remaining working hours based on reduced efficiency, using a numerical self-assessment scale; and (3) by calculating lost hours in cases where the caregiver permanently reduced or discontinued employment. In all cases, monthly values were annualized by multiplying by 12, and weekly values were annualized by multiplying by 52, reflecting the standard number of working weeks in a calendar year. Paid vacation weeks were not excluded, as they still represent paid time by the employer. In cases of complete withdrawal from employment, the entitlement to such paid leave—and the associated payment—would also be lost.

All time losses from the three categories were converted to annual values and expressed in monetary terms based on average net wages in Lithuania in 2023. That year, the national average net monthly salary was €1,261, corresponding to an hourly rate of approximately €7.90, assuming a standard full-time schedule of 160 h per month (40 h per week) [17] (https://osp.stat.gov.lt/). This provided a standardized estimate of forgone earnings attributable to caregiving-related work limitations.

Estimation of informal care costs

The monetary value of informal care was estimated using the opportunity cost approach, consistent with recommended methodologies for economic evaluations of informal caregiving [11, 18, 19]. Caregivers were asked to report the average daily hours spent on caregiving activities, including supervision, mobility assistance, feeding, hygiene, therapeutic exercises, and administrative tasks related to the child's condition. To avoid overestimation and preserve time for essential personal needs (such as sleep and self-care), reported caregiving time was capped at 18 h per day, equivalent to a maximum of 126 h per week. This upper limit is consistent with prior research applying realistic constraints in valuing time use [10]. In five cases where reported values exceeded this cap, data were recoded to the maximum of 126 h to ensure comparability and prevent distortion of results.

The recall period for all time-related questions (e.g., time spent on caregiving and productivity losses) was one month. Monthly values were annualized to obtain yearly cost estimates. A reduction in employment was considered a long-term adjustment in labour participation, referring to a sustained change in working hours or employment status due to the child's health condition. This distinction allowed for differentiation between short-term productivity losses within the recall period and longer-term changes in employment. Time spent on caregiving was valued using the 2023 net minimum hourly wage in Lithuania (€3.95/hour), calculated from the monthly minimum of €633, assuming a standard full-time workload (https://socmin.lrv.lt). This conservat ive valuation is commonly used in cost-of-illness studies to estimate the economic value of informal care outside paid work hours [18].

Statistical analysis

Data were anonymized and entered into Microsoft Excel for preprocessing and verification. Statistical analyses were performed using RStudio (2024.12.1 + 563). Descriptive statistics summarized socio-demographic characteristics, caregiving time, and cost outcomes, including direct medical and non-medical costs, indirect costs, and informal care costs. Continuous variables were reported as medians with interquartile ranges. We used the Kruskal-Wallis test to evaluate differences in continuous variables between three severity-based groups. The frequency distributions between groups are compared with the Chi-square or Fisher's exact test based on the expected frequencies. All cost estimates are presented in euros (€) and reflect 2023 price levels.

Ethical considerations

The study was approved by the Vilnius Regional Biomedical Research Ethics Committee (approval No.2022/9-1457-926) and conducted in accordance with national ethical standards. Written informed consent was obtained from all participants before their inclusion in the study.

Results

Sociodemographic and clinical characteristics

The study included the parents or guardians of 160 children with congenital anomalies, distributed across three severity-based groups. Children in the mildest category (Group I, single organ system affected) comprised the largest subgroup ($n = 88$), followed by Group II (two organ systems, $n = 34$) and Group III (three or more systems or chromosomal disorders, $n = 38$).

Most respondents were women, accounting for 87% of all participating caregivers. The median age of respondents was 40 years (IQR 36–45), with a statistically significant increase in caregiver age observed across severity groups ($p < 0.001$). In Group I, the median age was 38 years, compared to 41 years in Group II and 45 years in Group III (Table 1). This may reflect prolonged caregiving trajectories or older parental age among families facing more complex medical conditions.

Significant differences were observed in the recognition of official disability status (Table 2). Among children in Group I, two-thirds had not been granted formal disability classification. In contrast, half of the children in Group II and the vast majority in Group III had moderate or severe disability status. This difference was statistically significant ($p < 0.001$) and reflected the increasing clinical burden with greater anomaly complexity.

Marked differences also emerged in the assignment of special care and educational needs. In Group I, only a small proportion of children were recognized as requiring long-term care or supervision. In contrast, nearly two-thirds of children in Group III were officially classified as needing either constant supervision or permanent nursing care ($p < 0.001$). Similarly, special educational support was required for 82% of children in Group III, compared to just 26% in Group I ($p < 0.001$).

Employment status and impact on work

Most caregivers (81%) reported being employed during the survey, with similar proportions across severity groups (84% in Group I, 79% in Group II, and 76% in Group III; $p = 0.563$). Among those employed, the typical workload was 40 h per week, with no difference observed between groups (Table 3).

However, differences became more apparent when examining employment intensity. While 69% of caregivers in Group I reported working full-time, this proportion declined to 50% in Group III, suggesting that the intensity of caregiving responsibilities may limit full-time work engagement. Although this trend did not reach

Table 1 Sociodemographic characteristics of children and caregivers

Variable	Overall $N=160$[1]	Group I $N=88$[1]	Group II $N=34$[1]	Group III $N=38$[1]	*p*-value[2]
Child's age, years	10 (7, 13)	10 (7, 13)	8 (6, 12)	11 (8, 13)	0.047
Residence					0.911
Large city	88 (55%)	49 (56%)	17 (50%)	22 (58%)	
Other city	54 (34%)	30 (34%)	13 (38%)	11 (29%)	
Village	18 (11%)	9 (10%)	4 (12%)	5 (13%)	
Household size	4 (3, 4)	4 (3, 4)	4 (3, 4)	3 (3, 4)	0.017
Caregiver's age, years	40 (36, 45)	38 (34, 44)	41 (36, 44)	45 (39, 49)	< 0.001
Caregiver's gender					0.817
Female (%)	139 (87%)	77 (88%)	30 (88%)	32 (84%)	
Male (%)	21 (13%)	11 (13%)	4 (12%)	6 (16%)	
Marital status					0.606
Married/ cohabiting	120 (75%)	71 (81%)	23 (68%)	26 (68%)	
Single	14 (9%)	7 (8%)	4 (12%)	3 (8%)	
Divorced	24 (15%)	9 (10%)	7 (21%)	8 (21%)	
Widowed	2 (1%)	1 (1%)	0 (0%)	1 (3%)	
Education level					
Lower than secondary	8 (6%)	7 (8%)	0 (0%)	2 (5%)	
Upper secondary or vocational	45 (28%)	22 (25%)	11 (32%)	12 (32%)	
Higher education (Bachelor's/ Master's)	104 (65%)	59 (67%)	23 (68%)	22 (58%)	
Doctorate	2 (1%)	0 (0%)	0 (0%)	2 (5%)	
Household monthly income[3], euros	2,000 (1,370, 2,500)	2,000 (1,300, 2,500)	2,000 (1,500,3,000)	2,000 (1,300,2,400)	0.394

[1]Median (IQR) or Frequency (%)

[2]Fisher's exact test; Kruskal-Wallis rank sum test; Pearson's Chi-squared test

[3]Household income refers to earned income only and does not include disability benefits, which are reported separately

statistical significance ($p=0.173$), it reflects a general pattern of increased caregiver burden with greater care intensity.

The most pronounced difference was the need to adjust work arrangements due to caregiving responsibilities ($p<0.001$). Overall, 18% of caregivers reported having to reduce their working hours, and an additional 10% had to leave the workforce entirely. These effects varied by severity group: in Group I, 8% reduced their hours and 6% left work; in Group II, the corresponding figures were 24% and 15%; while in Group III, 37% reduced their working time and 16% exited the workforce altogether. These findings underscore the growing employment burden faced by families caring for children with more complex health conditions. Even when caregivers remain employed, they often have to adjust or scale back their work, contributing to notable indirect costs at both the household and societal levels.

Direct medical and non-medical costs

Caregivers reported a wide range of monthly out-of-pocket expenditures related to their child's condition, covering various non-reimbursed goods and services. These expenses included medications, transportation to medical facilities, privately paid diagnostic tests or specialist consultations, and fees for therapeutic or educational sessions such as physiotherapy or occupational therapy. The total reported monthly out-of-pocket spending increased markedly with the severity of the child's condition ($p<0.001$), with a median of €87 (IQR €30–€200) in Group I, €190 (IQR €80–€500) in Group II, and €353 (IQR €260–€610) in Group III. The most substantial cost components, where incurred, were specialist developmental or therapeutic sessions and transportation expenditures. A detailed breakdown of out-of-pocket cost components is provided in Supplementary Table 1.

The median annual out-of-pocket costs, estimated from monthly expenditures reported by caregivers, reached €2,040 (IQR €600–€4,470) across the entire sample (Table 4). These costs showed a clear and statistically significant increase with the severity of the child's condition ($p<0.001$). Families caring for children in Group I reported the lowest annual expenditures, with a median of €1,044 (IQR €360–€2,400), while those in Group II spent on average €2,280 (IQR €960–€6,000). The financial burden was highest in Group III, where caregivers reported a median of €4,230 (IQR €3,120–€7,320) annually—over four times higher than in the mildest group.

Indirect costs

Across the whole sample, caregivers reported a median of 96 missed work hours annually, with reduced work hours being less common. Both indicators showed substantial variation between groups. Reduced work hours reached

Table 2 Clinical and functional characteristics of children

Variable	Overall $N=160$[1]	Group I $N=88$[1]	Group II $N=34$[1]	Group III $N=38$[1]	*p*-value[2]
Disability status					< 0.001
No disability	78 (49%)	58 (66%)	17 (50%)	3 (8%)	
Mild	12 (8%)	7 (8%)	2 (6%)	3 (8%)	
Moderate	33 (21%)	14 (16%)	6 (18%)	13 (34%)	
Severe	37 (23%)	9 (10%)	9 (26%)	19 (50%)	
Disability benefits, €/month (among recipients, $N=82$)	300 (220, 500)	276 (184, 360)	300 (220, 500)	354 (240, 500)	0.127
Special care need					< 0.001
None	109 (68%)	72 (82%)	24 (71%)	13 (34%)	
Constant supervision	8 (5%)	2 (2%)	1 (3%)	5 (13%)	
Permanent nursing	41 (26%)	13 (15%)	9 (26%)	19 (50%)	
Special educational needs					< 0.001
Yes	66 (41%)	23 (26%)	12 (35%)	31 (82%)	

[1]Median (IQR) or Frequency (%)

[2]Fisher's exact test; Kruskal-Wallis rank sum test; Pearson's Chi-squared test

Table 3 Employment status and impact on work

Variable	Overall $N=160$[1]	Group I $N=88$[1]	Group II $N=34$[1]	Group III $N=38$[1]	*p*-value[2]
Employment status					0.563
Employed	130 (81%)	74 (84%)	27 (79%)	29 (76%)	
Not employed	30 (19%)	14 (16%)	7 (21%)	9 (24%)	
Work hours per week (among employed, $N=130$)	40 (40, 40)	40 (40, 40)	40 (40, 40)	40 (30, 40)	0.225
Full-time work (among employed)					0.173
Yes	104 (65%)	61 (69%)	24 (71%)	19 (50%)	
No	26 (16%)	13 (15%)	3 (9%)	10 (26%)	
Work adjustment due to caregiving					< 0.001
None	113 (71%)	74 (84%)	21 (62%)	18 (47%)	
Reduced hours	29 (18%)	7 (8%)	8 (24%)	14 (37%)	
Left workforce	16 (10%)	6 (6%)	5 (15%)	6 (16%)	

[1]Median (IQR) or Frequency (%)

[2]Fisher's exact test; Kruskal-Wallis rank sum test; Pearson's Chi-squared test

the highest median value in Group III (520 h), whereas in Groups I and II the median remained at 0 h ($p<0.001$). Missed work hours tended to be higher in Groups II and III compared with Group I ($p=0.051$). Overall productivity loss in hours followed a similar pattern, peaking in Group III and being lowest in Group I, although the difference was not statistically significant ($p=0.105$).

These time losses translated into notable monetary impacts (Table 4). The cost of reduced work hours was highest in Group III, while it was negligible in Groups I and II ($p<0.001$). Missed work hours were most costly in Group II and least expensive in Group I ($p=0.051$).

Table 4 Direct medical and non-medical, indirect, and informal care costs

Variable	Overall $N=160$[1]	Group I $N=88$[1]	Group II $N=34$[1]	Group III $N=38$[1]	*p*-value[2]
Monthly out-of-pocket costs, €	170 (50, 373)	87 (30, 200)	190 (80, 500)	353 (260, 610)	< 0.001
Annual out-of-pocket costs, €	2,040 (600, 4,470)	1,044 (360, 2,400)	2,280 (960, 6,000)	4,230 (3,120, 7,320)	< 0.001
Annual reduced work, hours	0 (0, 520)	0 (0, 0)	0 (0, 1,040)	520 (0, 1,040)	< 0.001
Annual missed work, hours	96 (0, 288)	96 (0, 192)	186 (78, 408)	156 (0, 480)	0.051
Productivity loss, hours/year	593 (0, 936)	562 (0, 988)	585 (320, 874)	780 (541, 946)	0.105
Cost of reduced work hours, €	0 (0, 4,108)	0 (0, 0)	0 (0, 8,216)	4,108 (0, 8,216)	< 0.001
Cost of missed work hours, €	758 (0, 2,275)	758 (0, 1,517)	1,469 (616, 3,223)	1,232 (0, 3,792)	0.051
Cost of productivity loss, €	4,683 (0, 7,394)	4,437 (0, 7,805)	4,622 (2,526, 6,901)	6,162 (4,272, 7,477)	0.105
Total annual indirect cost, €	7,566 (1,138, 13,196)	5,429 (0, 10,338)	8,695 (5,265, 14,465)	12,965 (9,897, 16,432)	< 0.001
Care time per month, hours	42 (4, 161)	21 (0, 68)	50 (8, 210)	108 (52, 296)	< 0.001
Care time per year, hours	498 (42, 1,932)	246 (0, 810)	600 (96, 2,520)	1,296 (624, 3,552)	< 0.001
Total annual informal care cost, €	1,967 (166, 7,631)	972 (0, 3,200)	2,370 (379, 9,954)	5,119 (2,465, 14,030)	< 0.001

[1]Median (IQR)

[2]Fisher's exact test; Kruskal-Wallis rank sum test; Pearson's Chi-squared test

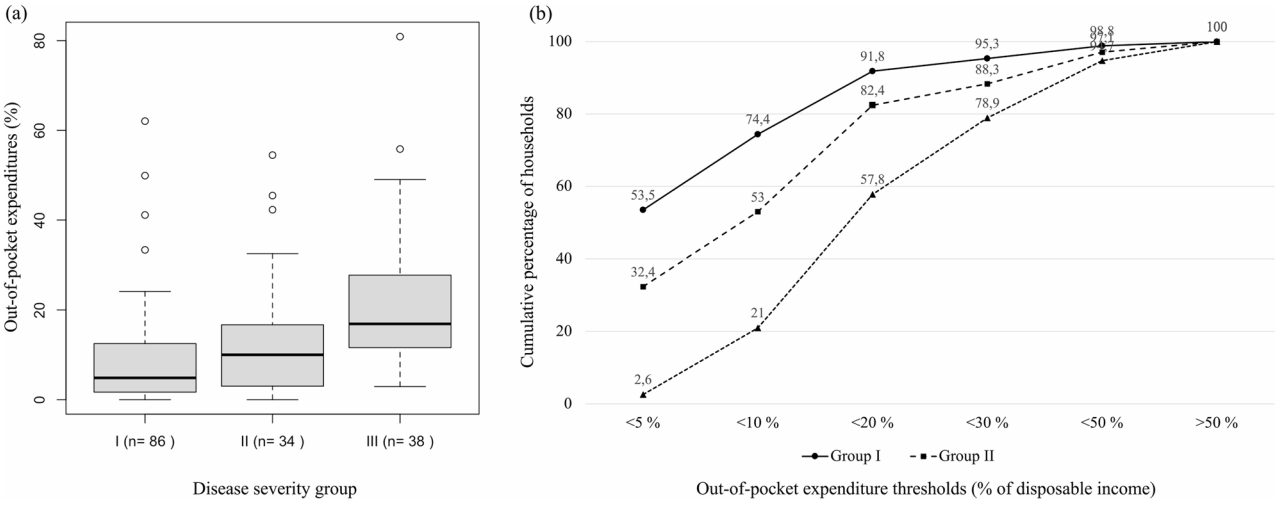

Fig. 1 Out-of-pocket expenditure burden and distribution across income thresholds: (a) Share of disposable household income allocated to out-of-pocket expenditures; (b) Cumulative distribution of households by out-of-pocket expenditure thresholds. Lines show the cumulative percentage of households exceeding successive thresholds of disposable income spent out-of-pocket (< 5%, < 10%, < 20%, < 30%, < 50%)

Productivity loss costs were greater in Groups II and III compared with Group I, but the difference did not reach statistical significance ($p = 0.105$). Combining all components, the total annual indirect cost per caregiver amounted to a median of €7,566, ranging from €5,429 in the mildest group to €12,965 in the most severely affected group ($p < 0.001$).

Informal care costs

The amount of time spent on informal caregiving varied widely among respondents, ranging from only a few hours to several hundred hours per month. Across the whole sample, caregivers provided a median of 42 h of informal care per month (IQR 4–161), with substantial variation by disease severity ($p < 0.001$). Monthly care time increased progressively with severity, from 21 h in Group I to 50 h in Group II and 108 h in Group III. Considerable variability was present within each group, with some caregivers in Group III reporting more than 250 h per month.

When expressed annually, this corresponds to 246 h in Group I, 600 h in Group II, and 1,296 h in Group III ($p < 0.001$); these differences in time commitment translate into marked economic disparities (Table 4). The median annual monetary value of informal care was €972 in Group I, €2,370 in Group II, and €5,119 in Group III, compared with €1,967 in the total sample ($p < 0.001$).

Distribution of out-of-pocket expenditure burden

Out-of-pocket expenditure burden, defined as the share of monthly disposable household income (earnings, pensions, disability benefits, and other social transfers) allocated to disease-related expenses, increased markedly with disease severity ($p < 0.001$). The median share was 4.9% (IQR 1.9–11.9) in the first group, 10% (IQR 3.1–16.3) in the second group, and 16.9% (IQR 11.7–27.8) in the third group (Fig. 1a). A few families reported notably high values, which may reflect either very high expenditures or possible over-reporting. These observations were retained in the analysis to preserve data integrity and reflect the full range of variability observed in real-world settings.

To further illustrate the pattern, we analyzed the distribution of households across predefined expenditure thresholds. In the mildest group, 53.5% of families spent less than 5% of their income, and only 8.2% spent more than 20% of their income. In the moderate group, 17.6% exceeded the 20% threshold. In the most severe group, 42.2% of families spent more than 20%, 21.1% more than 30%, and 5.3% more than 50% of household income. As shown in Fig. 1b, increasing disease severity was associated with a rightward shift in the cumulative distribution, indicating that fewer households incurred only minimal expenditures. At the same time, a larger proportion faced substantial out-of-pocket burdens.

Furthermore, only 19 families (11.9%) reported having no out-of-pocket expenditures, whereas nearly half (48.8%) incurred such costs without receiving any financial benefits. This imbalance highlights a considerable gap between the financial burden experienced by families and the compensatory support available to them.

Discussion

This study demonstrates that congenital anomalies impose a substantial and multifactorial economic burden on affected families in Lithuania. Importantly, this burden intensifies with disease severity and spans across multiple cost domains. In our cohort, indirect costs

(productivity losses) represented the largest component, with a median annual value of nearly €13,000 in the most severe group. These losses are mainly driven by reduced working hours and labour-force exits among caregivers. Similar employment disruptions have been reported in U.S. cohorts of children with spina bifida, where parents often face marked challenges in maintaining stable work participation [20]. Consistent with these findings, evidence from Sweden indicates that income losses among caregivers constitute a significant component of the overall economic burden in rare and disabling conditions [10]. Notably, the magnitude of productivity loss observed here underlines that labour market consequences are not incidental but structural outcomes of sustained caregiving demands.

Out-of-pocket expenditures represented the second largest component of the overall burden. These costs rose sharply with severity and were primarily driven by transportation, therapy, and educational services. A key aspect of our findings is the substantial relative burden of out-of-pocket costs compared to disposable household income. A considerable share of families crossed international thresholds for catastrophic health expenditures, defined by WHO and the World Bank as health spending that exceeds 10% of total household consumption [21, 22]. In our most severely affected group, more than 40% of families devoted at least 20% of their income to condition-related costs, with some exceeding 30–50%. These figures indicate that families of children with complex congenital anomalies face a significant risk of financial catastrophe. This pattern aligns with international evidence on the substantial economic impact of congenital anomalies. In Germany, families of children with congenital cardiac defects reported that transportation and accommodation alone accounted for more than one-fifth of all non-medical expenditures, second only to wage losses, illustrating how substantial out-of-pocket spending often co-occurs with pronounced productivity losses [23]. In the United States, nearly half of families of children with congenital heart disease reported financial hardship due to medical bills, and about one in six were unable to pay them, highlighting the substantial out-of-pocket exposure that families may face in systems with high cost-sharing. Such hardship was also associated with food insecurity and delayed medical care, indicating that the financial impact extends beyond healthcare expenses [24]. Similarly, analyses of privately insured U.S. children revealed that families of those with Down syndrome faced persistently higher out-of-pocket medical costs than families of unaffected children, with expenditures remaining elevated throughout childhood and adolescence [25]. According to U.S. national cost-of-illness data, congenital malformations and chromosomal abnormalities rank among the costliest rare disease categories in childhood, with mean per-person annual costs of approximately $80,000–$91,000. Notably, non-medical and out-of-pocket components— including expenditures not covered by insurance—account for nearly 20% of total costs, reflecting the substantial financial burden borne directly by families [26]. Taken together, these findings underscore that even within well-resourced health systems, families raising children with congenital anomalies remain vulnerable to financial hardship driven by both medical and non-medical spending.

The economic value of informal care in our study was also substantial, particularly for families of children with multisystem or chromosomal anomalies. This finding is consistent with a broad body of international evidence showing that informal caregiving constitutes a major share of the economic burden associated with rare and disabling childhood conditions [10, 27, 28]. In some European studies, informal care has even been identified as the single largest cost component. For example, in an Italian cost-of-illness study of patients with Cri du Chat syndrome, a rare chromosomal anomaly caused by a deletion on the short arm of chromosome 5, informal care accounted for approximately 87% of the total cost of illness [29]. Similarly, in a European study of individuals with Fragile X syndrome, an X-linked chromosomal disorder, informal care represented the largest share of total societal costs, exceeding the direct healthcare component [30]. By contrast, in our Lithuanian cohort, informal care was outweighed by productivity losses and out-of-pocket spending. This discrepancy likely reflects both contextual and methodological factors. First, we valued caregiving time conservatively, using the minimum wage and imposing daily hour caps. Second, the pronounced labour-market consequences observed in Lithuania may have shifted the balance towards indirect costs. Overall, the results reinforce that informal caregiving represents a significant and often under-recognized component of the total societal cost of congenital anomalies.

The multifactorial nature of the burden observed here indicates that isolated measures are insufficient. Families face concurrent pressures from out-of-pocket spending, productivity losses, and unpaid caregiving, underscoring the need for integrated policies. Given that productivity losses represent the largest cost component, labour market–oriented measures for caregivers are particularly relevant. Evidence from comparative European policy research highlights the role of paid care leave and workplace flexibility as key policy instruments aimed at enabling the integration of paid employment with caregiving responsibilities [31, 32]. While short-term sickness benefits for parents exist in many countries, including Lithuania, these schemes are primarily designed to address acute and time-limited care episodes. In contrast, several European countries have established

caregiver-related leave arrangements that allow longer-term and more flexible responses to care needs [33]. For example, in Sweden, parents are entitled to a temporary parental benefit allowing up to 120 days per child per year to care for a sick child, in addition to a flexible parental leave scheme of 480 days that can be taken intermittently over several years, providing formal mechanisms to reconcile prolonged caregiving with paid employment [34].

In parallel, the substantial out-of-pocket burden observed in this study—particularly for transportation and therapeutic services—highlights gaps in financial protection extending beyond the healthcare sector. Recent European policy frameworks point to the importance of addressing non-medical care-related costs and formally recognising informal caregiving in order to limit cumulative economic vulnerability among families with long-term caregiving responsibilities [35]. Strengthening such mechanisms may reduce both short-term financial pressure and longer-term economic risks among caregivers. Priority should be given to high-severity cases, where the economic burden is highest.

Limitations

Several limitations should be considered when interpreting these findings. First, the recall method was used for data collection, which relies on participants' ability to accurately remember and report their expenses, working hours lost, and time spent on caregiving. This may have introduced recall bias. Additionally, some respondents may have rounded figures to the nearest convenient number, which could potentially reduce precision. Second, all cost estimates were self-reported and not verified against administrative or employment records, which may have led to under- or overestimation of specific cost categories. Third, the study did not capture the costs incurred by other household members or potential productivity losses for secondary caregivers. Fourth, intangible costs, such as the psychological burden, reduced quality of life, and health effects on caregivers, were not quantified. These factors may be substantial, and their exclusion likely results in an underestimation of the actual economic impact.

Conclusion

This study demonstrates that congenital anomalies impose a substantial and multidimensional economic burden on families, encompassing considerable productivity losses, out-of-pocket expenditures, and the high economic value of informal care. The burden intensifies with disease severity, reflecting the cumulative pressures of complex caregiving demands. Addressing these challenges requires a comprehensive policy framework that recognizes the full economic impact of congenital anomalies and strengthens financial and social protection for affected households.

Supplementary Information

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Supplementary Material 1

Supplementary Material 2

Authors contributions

E.M.V. contributed to the conception and design of the study, coordinated data collection, performed data entry, participated in data interpretation, and wrote the main manuscript.A.J. contributed to the study design, performed the statistical analysis, prepared figures, and participated in the interpretation of the findings.L.M. contributed to the conception and design of the study, participated in data interpretation, and substantially revised the manuscript.A.U. contributed to the conception of the study and the interpretation of the findings.All authors reviewed and approved the final manuscript.

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Data availability

The datasets generated and analysed during the current study are not publicly available due to privacy and confidentiality restrictions but are available from the corresponding author on reasonable request.

Declarations

Competing interests

The authors declare no competing interests.

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