The Role of Orthopaedic Conditions in Lower Extremity Pain and Exercise Intolerance in Children with 22q11.2 Deletion Syndrome

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Introduction

"Just as each knight at King Arthur's Round Table brought a unique skill, my research combines paediatrics, orthopaedics, and psychology — reflecting the teamwork of our multidisciplinary 22q11 clinic at Wilhelmina Children's Hospital in Utrecht, The Netherlands."

Introduction (2)

Our clinical experience

- High frequency of lower extremity pain
- Major impact on daily activities and quality of life
- Sometimes with experience of exercise intolerance
- No obvious cause identified

Research timeline & question

Retrospective data

2023: start

Determine prevalence of reported leg pain

Explore association with heart defects, hypocalcemia and/or orthopaedic problems

2024: findings

Reported leg pain: 50%

No correlation with heart defects or hypocalcemia

Orthopaedic problems tend to be associated with leg pain

2025: next step

Expanded investigation of the correlation between leg pain and orthopaedic conditions

Research question

Are there specific orthopaedic conditions associated with leg pain in children with 22q11.2 deletion syndrome

Aim

Targeted monitoring and treatment during outpatient follow-up

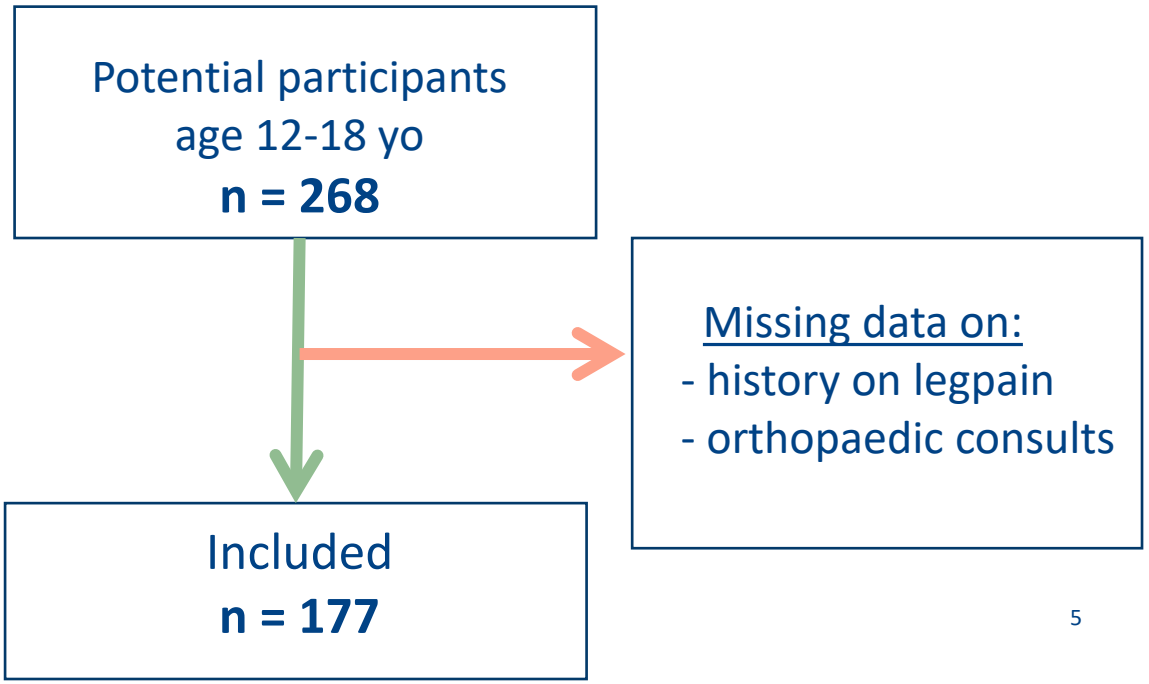
Method

Retrospective data:

- Jan 2015 – July 2025
- Age 12-18 years old
- Latest consultation

The term “leg pain” as reported, includes:

- Analgetics needed
- Nocturnal awakenings
- During excercise
- After excercise



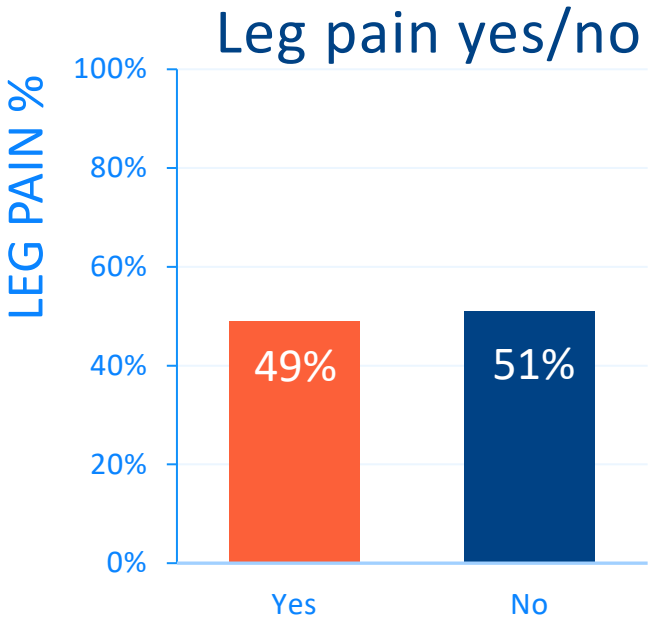
Results: Baseline characteristics

Demographics
n = 177

Characteristics	N (%)	Mean (SD)	Range
Gender			
Girls	91 (51%)		
Boys	86 (49%)		
Age (years)		15 (2,1)	12-18
Intelligence (IQ)		70 (12,3)	41-106
Education level			
Special Education	125 (71%)		
Regular Education	52 (29%)		
Orthopaedic problems*			
Scoliose	97 (55%)		
Pes planus	91 (51%)		
Patellar luxation	11 (6%)		
Clubfeet	6 (3%)		

**Note. As participants could have multiple orthopedic problems, percentages are not mutually exclusive and therefore do not sum to 100%*

Results: Prevalence of leg pain



Leg pain vs orthopaedic problems

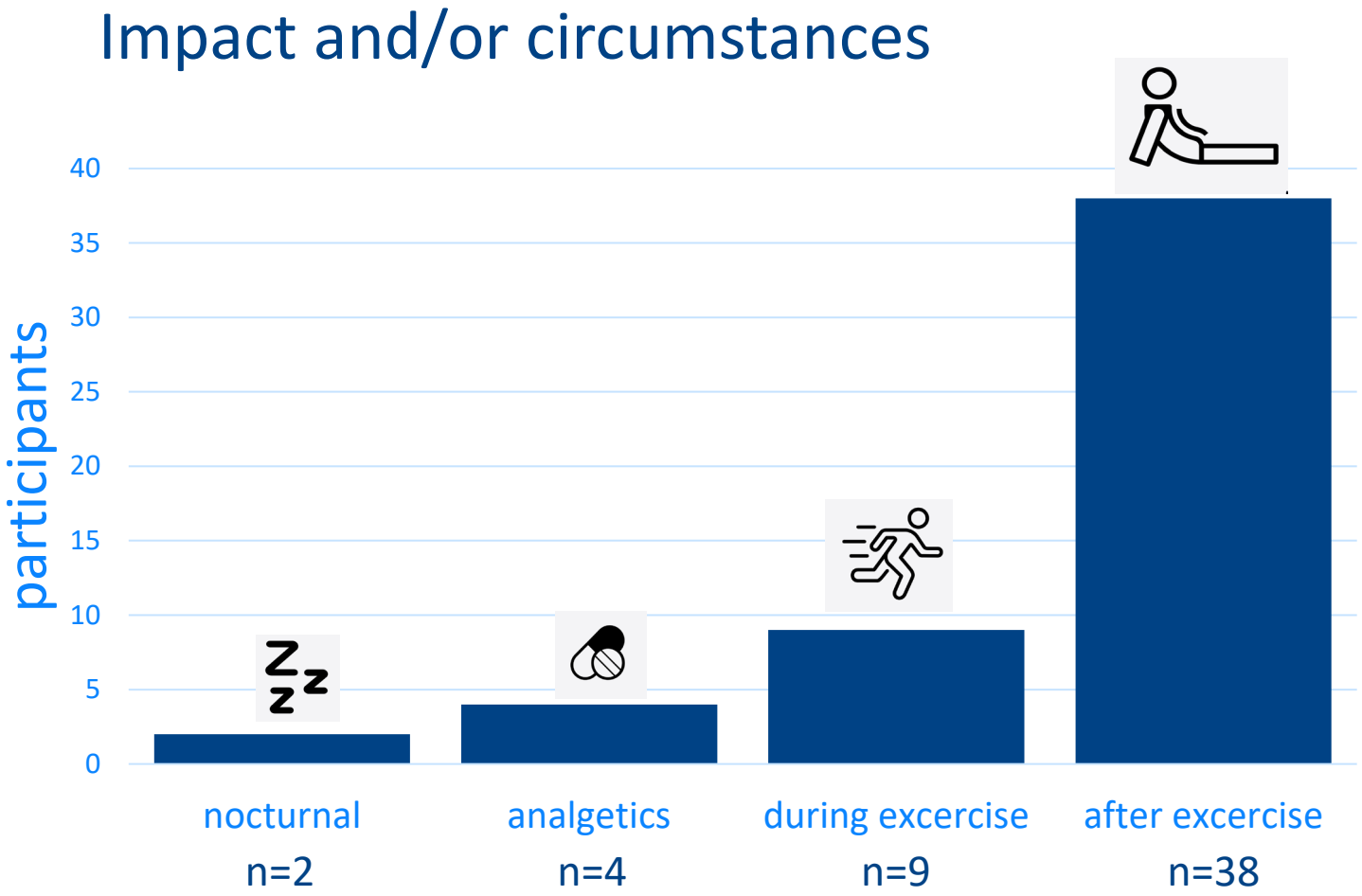
Orthopaedic problems	N (%)	Pain n (%)	No pain n (%)
None	33 (19%)	-	-
One or more	144 (81%)	73 (51%)	71 (49%)
Total	177 (100%)		

In total: 87/177 =
~ 50% reported leg pain

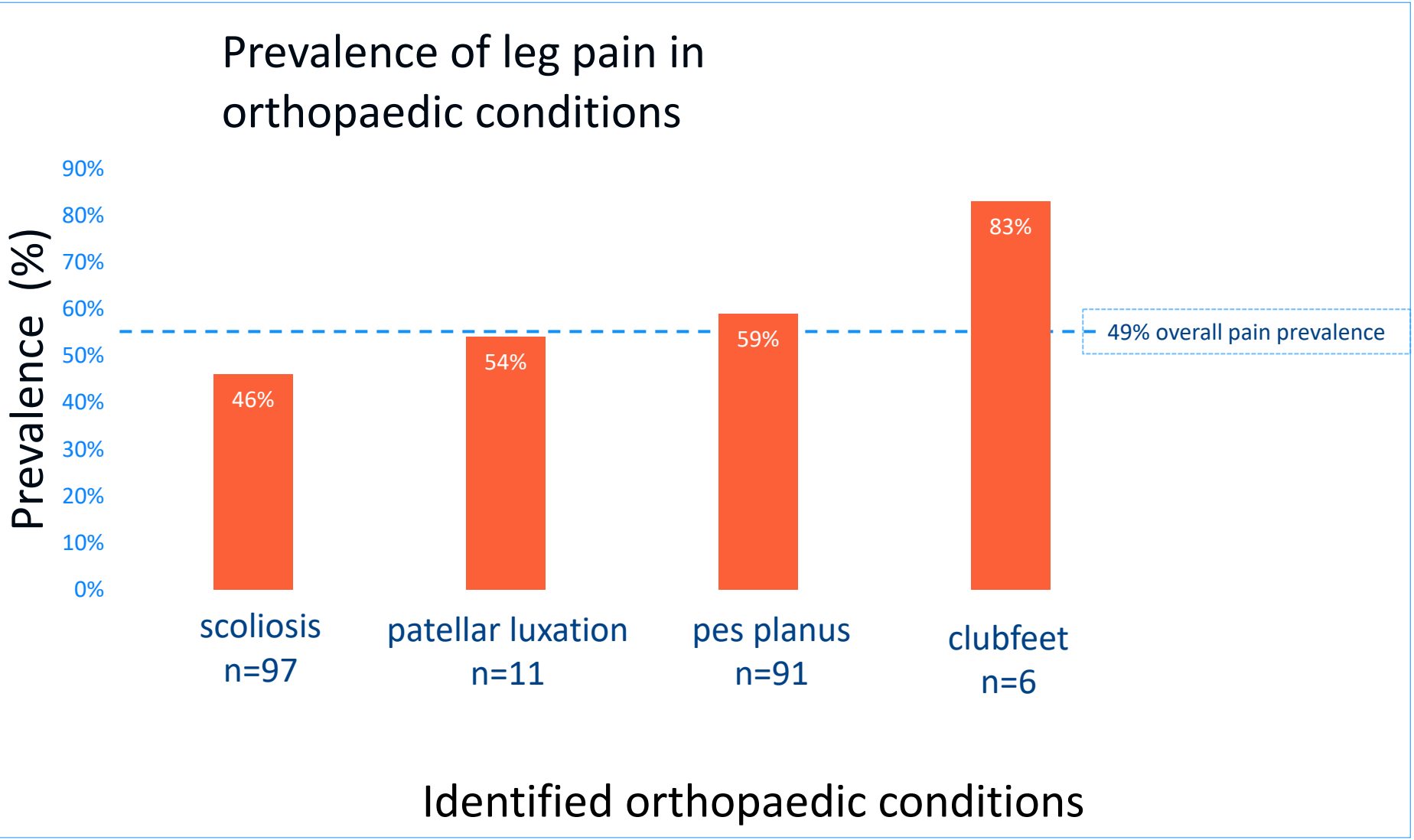
Within group with orthopaedic problems:
~ 50% reported leg pain

Results: Leg pain

n = 87



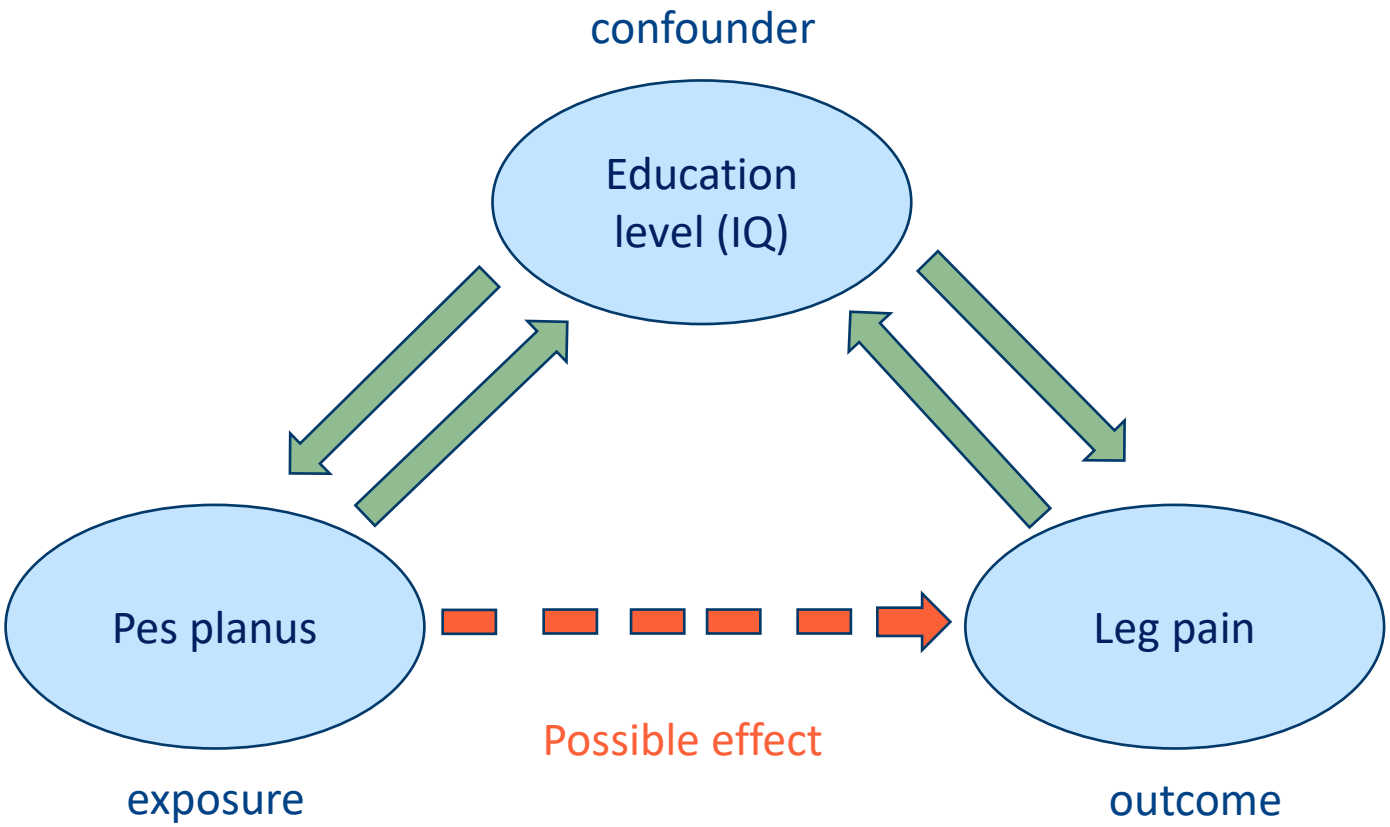
Results: Orthopaedic determinants of pain



Results: Univariate en multivariate analysis

VARIABELE	UNIVARIATE	MULTIVARIATE: ORTHOPAEDIC VARIABLES		
	P-value	OR	(95% CI)	*P*-value
Scoliosis	0,42	0,7	(0,4-1,3)	0,31
Pes Planus	**0,03**	1,9	(1,0-3,4)	**0,04**
Patellar luxation	0,71	1,5	(0,4-5,1)	0,54
Clubfeet	0,09	6,1	(0,7-54,9)	0,11
Gender (female vs male)	0,70	-	-	-
Age	0,23	-	-	-
Education level (standard versus special)	**0,005**	-	-	-

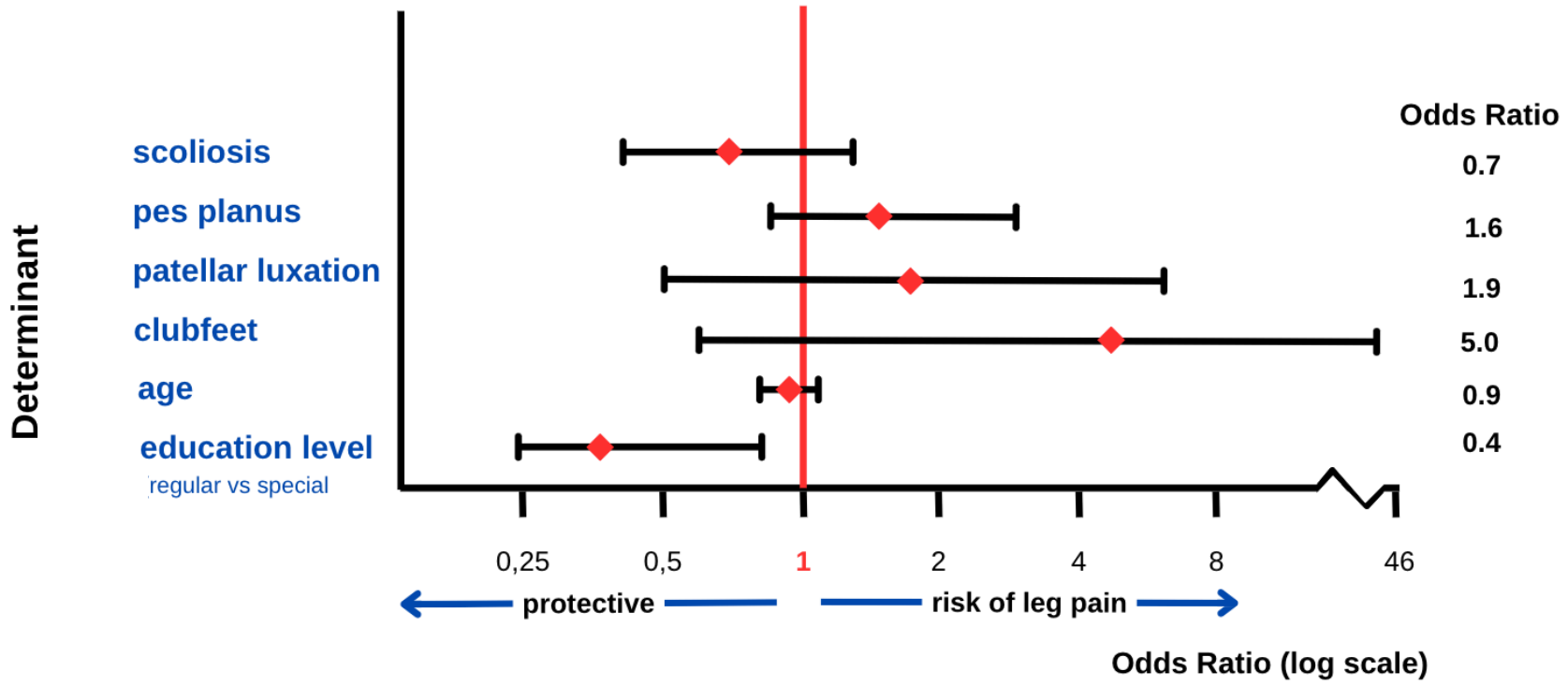
Confounding



Results: Multivariate analysis (1)

VARIABELE	UNIVARIATE	MULTIVARIATE: ORTHOPAEDIC VARIABLES			MULTIVARIATE: ORTHOPAEDIC VARIABLES+ CONFOUNDERS		
	P-value	OR	(95% CI)	*P*-value	OR	(95% CI)	*P*-value
Scoliosis	0,42	0,7	(0,4-1,3)	0,31	0,7	(0,4-1,4)	0,29
Pes Planus	**0,03**	1,9	(1,0-3,4)	**0,04**	1,6	(0,9-3,1)	**0,11**
Patellar luxation	0,71	1,5	(0,4-5,1)	0,54	1,9	(0,5-6,8)	0,33
Clubfeet	0,09	6,1	(0,7-54,9)	0,11	5,0	(0,6-45,0)	0,15
Gender (female vs male)	0,70	-	-	-	-	-	-
Age	0,23	-	-	-	0,9	(0,8-1,1)	0,29
Education level (standard versus special)	**0,005**	-	-	-	0,4	(0,2-0,8)	**0,02**

Results: Multivariate analysis (2)



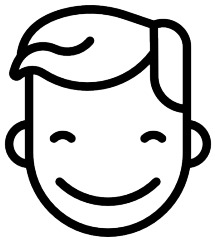
Summary & discussion

- 50% reported leg pain with orthopaedic problems
- Correlation between only the orthopaedic condition of flat feet for getting leg pain

BUT

- Education level did affect the outcomes for the factor flat feet

Limitations:



Conclusion

- High prevalence of leg pain present in children aged 12-18 years with 22q11DS
- 50% reported leg pain with orthopaedic problems
- Flat feet is initially significantly associated with leg pain, but this association disappears after adjusting for potential confounders
- Education level appears to be a stronger predictor, suggesting cognitive factors might play a larger role than physical factors alone
- Further research needed

Take home message

1.  Careful follow-up

2.  Insole study

3.  Context matters

4.  Comprehensive assessment

THANK YOU

Acknowledgement

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