

Dear Sir or Madam,

Thank you for reading this information. We hope you want to participate in our study 'Spinal deformities in patients with Koolen-de Vries syndrome'.

We know that spinal deformities exist in a significant number of individuals with KdVS. However, we don't know the exact prevalence and the clinical and radiological features of these spinal deformities.

By doing this study, we are aiming to gain insight in prevalence and clinical and radiological characteristics of these spinal deformities.

- To study clinical characteristics, we would ask you to fill in the additional questionnaire. You may decide to leave some questions unanswered. To study the prevalence of spinal deformities in KdVS, it's necessary that you return this document even if your child doesn't have a spinal deformity. By completing this questionnaire, you give consent to Dr. David A. Koolen and to Arianne Bouman to use the given information in this study.
- To study radiological characteristics, all radiology of the spine and pelvis (X-ray, CT and MRI) and radiology reports are requested. Please fill in the consent form below, to make it possible for us to request this information. There's a possibility you filled in this informed consent form last year, if that's the case, you can continue immediately with the questionnaire. (Some parents have access to their relatives' medical chart and radiology report, if you are willing to send the radiology report to us immediately, we are grateful.)

All questionnaires and radiological materials will be stored in a secured database, pseudonymous, and will only be used for research purpose.

Do not hesitate to contact us by sending an e-mail to Arianne.Bouman@radboudumc.nl. Would you please send this form (digital or scanned) to Arianne.Bouman@radboudumc.nl?

Arianne Bouman
Projectcoordinator study
'Spinal deformities in KdVS'



Dr. David A. Koolen
Clinical geneticist, Radboudumc



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(Attn.) Arianne Bouman, department of pediatrics
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Consent to disclose Personal Health Information

By signing this form, I authorize you to disclose confidential health information about my child by releasing **radiographic images and radiology reports of spine and pelvis** to the persons listed below.

I, *(print your name)*

authorize *(print name of health information custodian / name of your child's treating physician and name of the hospital)*

to disclose the health information of *(name and date of birth of person for whom you are the (substitute) decision maker)*

consisting of diagnostic images and diagnostic imaging reports to:
Dr. D.A. Koolen; clinical geneticist; Radboudumc, Nijmegen, the Netherlands,
and A. Bouman, researcher KdVS, Radboudumc, Nijmegen, The Netherlands

The purpose / reason for this release of the information is as follows:

Research project: Spinal deformities in Koolen- de Vries syndrome

I understand the purpose for disclosing this personal health information to the person noted above. I understand that I can refuse to sign this form.

My name

Address

City: State: Zip Code

Telephone number

E-mail address

Signature

Date

Radboudumc, Amalia Children's Hospital
(Attn.) Arianne Bouman
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6500 HB
Nijmegen, The Netherlands
Fax# (0)24 3616 428



Questionnaire – Study ‘spinal deformities in patients with Koolen-de Vries syndrome’

1. Gender of your relative
 - Male
 - Female
2. Your relative has the diagnosis of KdVS since _____ (year)
3. KdVS is due to:
 - 17q21.31 microdeletion
 - KANSL1 mutation
4. How will you describe the muscle tone of your relative in the first year after birth?
 - Hypotonia (*muscle weakness*)
 - Normal muscle tone
 - Hypertonia (*muscle stiffness or high muscle tension*)
5. How will you describe the current muscle tone of your relative?
 - Hypotonia (*muscle weakness*)
 - Normal muscle tone
 - Hypertonia (*muscle stiffness or high muscle tension*)
6. What is the current height and weight of your relative?
(height) _____ (weight) _____
7. Does your relative have laxity of joints (ligamentous laxity, looseness, hypermobility)
 - Yes
 - No
 - I am not sure
8. Does your relative have walking problems? He or she
 - never learned to walk
 - does walk, but for limited time or distance
 - does not have any walking problems
9. Does your relative experience back pain?
 - No
 - Not anymore
 - Yes, daily
 - Yes, weekly
 - Yes, but not frequently
 - I am not sure
10. Does your relative have any neurological leg symptoms (e.g. leg pain, paresthesia)
 - No
 - Not anymore
 - Yes (namely _____)
 - I am not sure



11. Does your relative have movement restriction of the spine?

- No
- Not anymore
- Yes (namely _____)
- I am not sure

12. Did your relative receive the clinical diagnosis of a spinal deformity (scoliosis, kyphosis or lordosis)?

- Yes (*Continue with question number 13*)
- No (*Continue with question number 16*)

13. What kind of spinal deformity is diagnosed?

- Scoliosis
- Kyphosis
- Lordosis
- I am not sure

14. What is the year of diagnosis of this spinal deformity? _____ (year)

15. What was your relative's BMI at the time the spinal deformity was diagnosed?

- Underweight
- Normal weight
- Overweight
- Obesity

16. Did your relative receive one or more of the following therapies?

- No (*You've finished this questionnaire, please go to the end*)
- Yes, physical therapy - Start date _____ End date _____
 - One or more times a week
 - Less than once a week
- Yes, serial casting - Start date _____ End date _____
- Yes, bracing - Start date _____ End date _____
- Yes, surgical therapy - Date _____
- Yes, other therapy, namely _____ - Start date _____ End date _____ (*If yes, continue with question number 17*)

17. What was the effect of this therapy?

- Spinal deformity became less
- Complains/symptoms became less, namely _____
- No effect
- Complains/symptoms became more severe, namely _____
- Other _____

Thank you for filling in this questionnaire. Please send this form to Arianne.Bouman@radboudumc.nl.

