

Name of study subject: _____

ID #: _____

Examiner: _____

Date: _____

Respondent (if applicable): _____

DOB: _____ Sex: _____

Data Derived From (Check): Physical Examination Medical Record (Date of exam): _____

ACE Subject Physical Exam

NOTE: This form should be completed by a physician or nurse, or extracted from the medical record.

Instructions: Fill in the information requested or circle the relevant response (Yes, No, NK=Not Available or Not Examined or Not Known).

Measurements

1. Height _____(cm)
2. Weight _____(kg)
3. Head circumference: _____(cm) (measure maximal occipital-frontal circumference)

HEENT (Head, Eyes, Ears, Nose, Throat/mouth)

4. Is there any facial asymmetry? Yes No NK
5. Are there dysmorphic facial features (e.g. eyes too closely or widely spaced, ears lowset)? Yes No NK
6. Is vision abnormal? Yes No NK
If yes, is there any correction/what kind? (check): Glasses or Corrective lens Surgery
7. Is hearing abnormal? Yes No NK
If yes, is there any correction/what kind? (check): Hearing aids Cochlear implants
8. Are the eye movements full and symmetric? Yes No NK
9. Is there strabismus (eye malalignment)? Yes No NK
10. Is there nystagmus (fine back and forth movements of the eyes)? Yes No NK
11. Is there ptosis (drooping of the upper eyelids)? Yes No NK
12. Do ears appear abnormal? Yes No NK
If yes, how do they appear? (circle): Cupped / Prominent / Lowset / Tags
13. Does the jaw appear abnormal? Yes No NK
14. Is there a cleft lip and/or cleft palate? Yes No NK

Neck, Chest, Back

15. Is the neck short and/or webbed? Yes No NK
16. Is there a pectus deformity (breastbone scooped in or prominent)? Yes No NK
17. Is there scoliosis (curvature of the spine)? Yes No NK

18. Is there a heart murmur or abnormal heart examination? Yes No NK

Abdomen

19. Is there evidence of hepato(spleno)megaly (large liver or spleen)? Yes No NK

Genitourinary (optional)

20. Is the genital exam abnormal? Yes No NK

If yes, what findings? (circle): Hypospadias / Cryptorchidism / Enlarged testicles / Micropenis / Hypoplastic labia

Limbs

21. Are there any digital abnormalities (extra or missing fingers or toes)? Yes No NK

If yes, what findings? (circle): Extra fingers / Missing fingers / Extra toes / Missing toes / Fused digits

22. Are joints excessively loose? Yes No NK

23. Are there joint contractures? Yes No NK

Skin

24. Are there areas of **hypopigmentation** (ash-leaf spots)? Yes No NK

25. Are there areas of **hyperpigmentation** (café-au-lait spots)? Yes No NK

26. Are there freckles under the arms and/or groin area? Yes No NK

27. Are there neurofibromas (bumps under the skin, sometimes with a bluish tinge)? Yes No NK

28. Any other skin abnormalities? Yes No NK

Neurologic

29. Is there any evidence of **hypotonia** (low muscle tone)? Yes No NK

30. Is there any evidence of **hypertonia** (increased muscle tone)? Yes No NK

31. Is muscle strength abnormal? Yes No NK

If yes, what findings (check): Generalized weakness Focal weakness

32. Is there evidence of poor coordination or gait ataxia (difficulty walking straight)? Yes No NK

33. Are there any involuntary movements? Yes No NK

34. Are the cranial nerves normal (do face, tongue and palate move normally)? Yes No NK

35. Is sensory exam normal? Yes No NK