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 hepatosplenomegaly (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?
   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