<table>
<thead>
<tr>
<th>CONSIDERATIONS</th>
<th>RECOMMENDATIONS</th>
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<tr>
<td><strong>1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)</strong></td>
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| Children Vision: strabismus, refractive errors are common | □ Undertake newborn vision and hearing screening and an auditory brainstem response (ABR).  
□ Refer for a comprehensive ophthalmologic examination by 4 years of age.  
□ Visualize tympanic membranes at each visit. |
| Hearing: recurrent otitis media is common | |
| Nose: sinusitis is common | |
| Adults: strabismus and refractive errors are common | □ Undertake hearing and vision screening at each visit with particular attention to myopia and hearing loss. |
| **2. DENTAL** | |
| Children and Adults: High arched palate and dental malocclusion are common | □ Refer to a dentist for an annual exam. |
| **3. CARDIOVASCULAR** | |
| Children: Mitral Valve Prolapse (MVP) is less common in children (~10%), but may develop during adolescence | □ Auscultate for murmurs or clicks at each visit. If present, do an ECG and echocardiogram; refer to cardiologist, if indicated. |
| Adults: MVP is common (~ 80%). Aortic root dilation usually is not progressive Hypertension is common and exacerbated by anxiety | □ Undertake an annual clinical exam. Based on findings, obtain an ECG and echocardiogram. Refer to cardiologist, as appropriate.  
□ Measure BP at each visit and at least annually.  
□ Treat hypertension when present. |
| **4. RESPIRATORY** | |
| Children & Adults: Obstructive sleep apnea (OSA) may be due to enlarged adenoids, hypotonia or connective tissue dysplasia | □ Ascertain a sleep history and assess for evidence of OSA.  
□ Obtain a sleep study as appropriate. |
| **5. GASTROINTESTINAL** | |
| Children: In infants, feeding problems are common with recurrent emesis associated with Gastroesophageal Reflux Disease (GERD) in ~ 30% of infants | □ Refer for assessment of GERD. Thickened liquids and upright positioning may be sufficient to manage GERD. |
| **6. GENITOURINARY** | |
| Children and Adults: Inguinal hernias are relatively common in males  
Macroorchidism generally develops in late childhood and early adolescence and persists  
Ureteral reflux may persist into adulthood | □ Assess for inguinal hernia annually beginning at age 1 year.  
□ Macroorchidism can be measured with an orchidometer; reassure parents and patients that it does not require treatment.  
□ Evaluate recurring urinary tract infections (UTI) with cystourethrogram and renal ultrasound. Refer to a nephrologist or urologist as needed.  
□ Consider and assess for a renal etiology, such as scarring, as the basis for persistent hypertension. |
| **7. SEXUAL FUNCTION** | |
| Adults: Males and females are fertile | □ Consider discussion of recurrence risk and reproductive options as a basis for referral to a geneticist. Make such a referral even if fragile X is only suspected so that molecular testing can be undertaken in the person concerned and relevant family members. |
## CONSIDERATIONS

### 8. MUSCULOSKELETAL (MSK)
- **Children & Adults:** Hyperextensible joints and pes planus are common. Scoliosis, clubfeet, joint dislocations (particularly congenital hip) may also occur

### 9. NEUROLOGY
- **Children & Adults:** ~ 20% have epilepsy (may include generalized tonic-clonic seizures, staring spells, partial motor seizures, and temporal lobe seizures)
- Hypotonia is common, in addition to fine and gross motor delays
- Epilepsy occasionally persists into adulthood

### 10. BEHAVIOURAL/MENTAL HEALTH
- **Children:** 70% - 80% are hyperactive; ~ 30% have autism
- Autistic-like features are common and may indicate concurrent Autism Spectrum Disorder
- Anxiety and mood disorders can also be present
- Some features of autism, tantrums and aggression as well as anxiety and mood disorders may be treated with specific pharmacological agents
- Sensory defensiveness is common

### 11. ENDOCRINE
- **Children:** Precocious puberty may occur
- **Adults:** Premenstrual symptoms (PMS) may be severe

### 12. OTHER
- Occasionally presents as Prader-Willi syndrome-like phenotype

## RECOMMENDATIONS

### 8. MUSCULOSKELETAL (MSK)
- Undertake an MSK exam at birth, then every 4 months to adulthood, then at least annually.
- Elicit a history of possible dislocations.
- Refer to an orthopedic surgeon as dictated by clinical findings.
- Referral to an occupational therapist (OT) in childhood is essential.
- Consider referring to a physiotherapist and podiatrist for orthotics.

### 9. NEUROLOGY
- Ascertain a history of seizures, which usually present in early childhood.
- Assess for atypical seizures in adulthood if suspicious findings occur or if intellectual function decreases.
- Arrange an EEG if epilepsy is suspected from the history.
- Refer to a neurologist as dictated by clinical findings.

### 10. BEHAVIOURAL/MENTAL HEALTH
- Make an early referral to a clinical psychologist for essential parental teaching of appropriate behaviour modification techniques following diagnosis.
- Encourage use of antioxidants including vitamin E, vitamin C, folate and fruit juices.
- Hyperactivity may be managed using stimulant medications after age 5 years.
- Refer to an Intensive Behavioural Intervention (IBI) Autism treatment program if Autism Spectrum Disorder is present.
- Consider a referral to a psychiatrist for possible mental health disorders.
- Refer to a speech and language therapist following diagnosis.
- Refer to an occupational therapist (OT) for a sensory diet and sensory integration program.
- Consider referral to a psychiatrist or psychologist to assess and manage possible mental health disorders.
- Violent outbursts are frequent, especially in males, and may respond to behavioural and/or pharmacological measures (as for children).

### 11. ENDOCRINE
- Include attention in clinical examination to signs of precocious puberty in females. Refer to an endocrinologist for consideration of use of a gonadotropin agonist to manage precocious puberty.
- Ascertain history of PMS with attention to menstruation, anxiety, depression, and mood lability. Consider an SSRI to stabilize mood if PMS symptoms are severe enough.

### 12. OTHER
- For management of obesity and hyperphagia, consider approaches recommended for persons with Prader-Willi syndrome.
- Refer to appropriate specialists (e.g., neurologist, endocrinologist, psychiatrist) as indicated to assist in managing Prader-Willi syndrome-
### Health Watch Table – Fragile X Syndrome

#### CONSIDERATIONS

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<th>PREMUTATION CARRIERS:</th>
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<td>A late onset tremor/ataxia syndrome has been reported in ~ 40 – 50% of male and ~ 8% of female fragile X permutation carriers</td>
<td>like symptoms.</td>
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<td>Premature ovarian failure by age 45 has been reported in ~ 20 – 40% of female fragile X premutation carriers</td>
<td>□ If premutation is suspected but not yet identified, order fragile X DNA testing or refer to a genetics clinic.</td>
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<td>Psychiatric problems (e.g., mood and anxiety disorders) seem likely to occur in both male and female fragile X premutation carriers</td>
<td>□ To manage depression or anxiety in premutation carriers, SSRIs, regular exercise and counseling have been helpful.</td>
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#### WEB SITES THAT MAY BE HELPFUL FOR FAMILIES AND CAREGIVERS

- FRAXA Research Foundation  □ [www.fraxa.org](http://www.fraxa.org)
- Fragile X Research Foundation of Canada  □ [www.fragile-x.ca/default2.htm](http://www.fragile-x.ca/default2.htm)

#### REFERENCES CITED


#### PUBLISHED HEALTH CARE GUIDELINES REVIEWED AND COMPARED


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