Stickler Syndrome Clinic
for Children and Adults

Mission statement:
The Stickler Syndrome Clinic will evaluate children and adults for whom the diagnosis of Stickler Syndrome has been raised and will coordinate multidisciplinary care for affected patients.

- MassGeneral Hospital/MassGeneral Hospital for Children
- Yawkey Center for Outpatient Care, Suite 6C, Boston, MA 02114
- Telephone: 617-726-1561
- Director: Ruth Liberfarb, M.D., Ph.D.
- Email: rliberfarb@partners.org

Directions to the Yawkey Center for Outpatient Care

From the North
- Take I-93 south to Exit 26 (Storrow Drive/Cambridge)
- Follow signs to Storrow Drive (stay left)
- Take Storrow Drive west to Government Center exit (left exit)
- At end of ramp, yield to crossing traffic at flashing traffic signal, then proceed straight
- Turn left onto Cambridge Street at traffic signal
- Turn left onto North Grove Street
- Follow signs to park at the Yawkey Center for Outpatient Care

From the South
- Take I-93 north to Exit 26 (Storrow Drive)
- Follow signs to Storrow Drive (stay right)
- Take Storrow Drive west to Government Center exit (left exit)
- At end of ramp, yield to crossing traffic at flashing traffic signal, then proceed straight
- Turn left onto Cambridge Street at traffic signal
- Turn left onto North Grove Street
- Follow signs to park at the Yawkey Center for Outpatient Care

From the West
- Take I-90 east to Exit 18 (Brighton/Cambridge)
- Follow signs to Cambridge/Somerville
- Turn right onto Storrow Drive eastbound to Downtown Boston
- Take Storrow Drive east to Government Center exit
- Follow signs to Downtown/Government Center (Cambridge St.)
- Turn left onto North Grove Street
- Follow signs to park at the Yawkey Center for Outpatient Care

From the East
- Take I-90 west through the Ted Williams Tunnel
- Follow I-90 west to Exit 24 (I-93 North)
- Take I-93 north to Exit 26 (Storrow Drive)
- Follow signs to Storrow Drive (stay right)
- Take Storrow Drive west to Government Center exit (left exit)
- At end of ramp, yield to crossing traffic at flashing traffic signal, then proceed straight
- Turn left onto Cambridge Street at traffic signal
- Turn left onto North Grove Street
- Follow signs to park at the Yawkey Center for Outpatient Care

Please pay for parking at the Cashier Desk in the Lobby.
**Stickler Syndrome Facts**

- First described by Stickler et al, 1965 & 1967
- Originally called Hereditary Progressive Arthro-opthalmopathy
- Inheritance: Autosomal Dominant, Autosomal Recessive
- Prevalence: 1 in 10,000 – probably underestimated
- Penetrance: 100%
- Variable expressivity within a family
- Most common cause of hereditary Retinal Detachment
- Most common cause of juvenile Retinal Detachment
- Genetic heterogeneity
- Support group: Stickler Involved People (SIP) Email: sip@sticklers.org

**Stickler Syndrome: Clinical features**

- **Ocular:** Vitreoretinal degeneration, myopia, cataracts, retinal holes and detachments
- **Auditory features:** high tone sensory neural hearing loss, hypermobile tympanic membranes
- **Craniofacial features:** mid facial hypoplasia, micrognathia, palatal abnormalities (cleft palate, submucous cleft, bifid uvula, Pierre Robin anomaly)
- **Musculoskeletal anomalies:** mild spondylepiphyseal dysplasia, loose joints, early onset degenerative arthritis
- **Cardiovascular abnormality:** mitral valve prolapse

**Stickler Syndrome Clinic Evaluation**

- Comprehensive family and medical history
- Physical examination
- Clinical tests (audiogram, echocardiogram, skeletal X-rays, and DNA analysis)
- Education/ genetic counseling
- Family Screening
- Reproductive options

**Treatment of associated problems:**

corrective lenses; prophylactic laser or cryoretinal treatment; surgery; hearing aids; speech and feeding therapy; physical therapy and appropriate exercise; antiinflammatory and pain medication; referrals to specialists:

- **Ophthalmologist or retina specialist** – Evaluation needed at least annually or more frequently if medically indicated
- **Audiologist** – hearing evaluation is needed every 3-5 years
- **Orthodontist** – for a dental evaluation
- **Rheumatologist or orthopedist** – for musculoskeletal evaluation
- **Physiatrist** – for pain management
- **Specialist in Reproductive Medicine and In Vitro Fertilization**

This Brochure is funded by Stickler Involved People support group Website: [http://www.sticklers.org/sip2/](http://www.sticklers.org/sip2/)