New Patient Checklist:

This check list will help you navigate the manifestations that may be present as well as how to find out if they even exist. Every patient should be screened for dysmorphic features at the time of their diagnosis, whatever age that may occur. These may or may not require care. A complete list is below.

Newborn / Infancy:

Hydrocephalus: This is a buildup of fluid in the parts of the brain called the ventricles. Individuals with hydrocephalus may have a large skull (macrocephaly) which is another manifestation of GS.
How to diagnose: MRI of the brain.

Brain ventricle asymmetry: Brain ventricles are fluid-filled chambers inside the skull that keep the brain cushioned and protected. When there is more fluid in one chamber than another, the brain ventricle is defined as asymmetric.
How to diagnose: MRI of the brain.

Macrocephaly: An individual with macrocephaly has a large skull.
How to diagnose: Health care providers diagnose this by measuring the circumference of the skull, often with a tape measure.

Dysmorphic features: All patients with Gorlin syndrome will have a unique collection of symptoms and manifestations; there is a list below of known dysmorphic features associated with the disease, but no one patient will have all of them.

Frontoparietal bossing: This is a prominent forehead. It may lead to or be part of macrocephaly (see above).
How to diagnose: Health care providers diagnose this by measuring the circumference of the skull, often with a tape measure. It is also noted by observation.

Cleft Lip and or Palate: A cleft lip is an opening or a split in the upper lip. A cleft palate is an opening in the roof of the mouth (palate). Individuals with GS may have one or both.
How to diagnose: If not noted visually, or on exam, they may be diagnosed when an infant has difficulty with bottle or breast feeding. Care and treatment of cleft lip and or palate is accomplished by a team of health care providers that includes an oral surgeon, pediatric plastic surgeon, otolaryngologist (ear, nose & throat doctor), speech therapist, nutritionist, social worker, psychologist.

Syndactyly: People with syndactyly have some or all their fingers or toes fused together by tissue sometimes called "webbing." Syndactyly is not typically painful but can affect the functioning of the hands or feet depending on the level of fusion.
How to diagnose: Syndactyly is noted visually. Evaluation by a pediatric plastic surgeon is ideal.

Epidermal cysts: Also called sebaceous, keratin or epithelial, epidermal cysts are small hard lumps that live just under the skin. Usually slow growing, epidermal cysts can appear anywhere on the body and rarely cause pain or other symptoms unless they become infected or drain.
How to diagnose: These are noted visually. Evaluation by a pediatric plastic or general surgeon is ideal.

Cardiac fibroma: Cardiac fibromas are benign (non-cancerous) tumors of the heart made up of connective tissues.
How to diagnose: Cardiac fibromas may be diagnosed with an MRI or sonogram and/or a tissue biopsy.

Medulloblastoma: Medulloblastoma is a malignant (cancerous) brain tumor that grows quickly. It is seen most often in children younger than 8 years. In those with Gorlin syndrome, medulloblastoma may develop in the first two years. More boys with Gorlin syndrome develop this tumor than girls.
How to diagnose: The diagnosis is made by MRI of the brain. Once diagnosed, the child should be seen by a neurosurgeon and an oncologist as soon as possible.

When discussing treatment options, it is important to recognize that the use of radiation therapy may induce the growth of multiple basal cell carcinomas in patients with Gorlin syndrome. The risks and benefits of each treatment option should be carefully considered by the child's entire care team.

Toddler to Teens

Jaw Cysts: An odontogenic keratocyst (OKC) is a benign (non-cancerous) cyst or tumor that grows in the lower or upper jaw bones. The cysts are formed from the cells and tissue that help develop teeth. OKCs are often found during routine dental x-rays or when pain, swelling and drainage develop in the area. They may be associated with impacted baby teeth, and can cause problems with the new teeth coming in. OKCs can return and must be treated aggressively. If OKCs are left untreated they continue to grow, pushing out permanent teeth and damaging the jaw bones. They can expand into the sinus cavity and cause discomfort in the cheek(s).
How to diagnose: OKCs are usually diagnosed with a panoramic x-ray (panorex) of the jaw or a specialized CT scan (also called a cone beam CT scan or i-cat). They are usually treated by oral and maxillofacial surgeons.

Calcified ovarian fibromas: Ovarian fibromas are non-cancerous tumor-like growths on or near a woman's ovaries. They are made up of an overgrowth of connective tissues. As a girl approaches her teens and starts to menstruate, it is important to start monitoring for these benign calcified tumors on the ovary(s).
How to diagnose: Lower abdominal ultrasound or MRI is used to diagnose these fibromas. Because they are calcified, they appear as white areas, much like bone on an x-ray. These are best treated by gynecologic surgeons.

Calcified Falx Cerebri: The falx cerebri is a tough membrane that separates the right and left sides of the brain. The falx cerebri becomes calcified when

calcium salts build up and cause the membrane to harden. A calcified falx cerebri has no symptoms and does not affect health.
How to diagnose: A health care provider can identify calcified falx cerebri in an x-ray or brain MRI.

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Kyphoscoliosis: Kyphoscoliosis is a combination of kyphosis (when the spine turns outward) and scoliosis (when the spine curves into a "c" or "s" shape). In addition to disfigurement, kyphoscoliosis can cause pain, trouble breathing, stiffness fatigue and nerve issues.
How to diagnose: This condition is generally diagnosed with x-rays, MRI, or CT scan and treated by an orthopedic surgeon, neurosurgeon, physical therapist, occupational therapist.

Spina bifida occulta: In spina bifida occulta, or closed spina bifida, one or more bones of the spine do not form correctly. People with Gorlin syndrome typically have spina bifida occulta in the bones of the neck (cervical vertebrae) or the upper/back (thoracic vertebra).
Unlike in spina bifida, the spinal column in spina bifida occulta is not significantly moved out of place. For this reason, spina bifida occulta usually does not cause any symptoms. Sometimes there are outward signs of spina bifida occulta such as a tuft of hair or a small dimple or birthmark on a child's lower back. In rare

instances, the spinal cord may be compressed by abnormalities in the skeleton as a result of Gorlin syndrome. This may result in abnormal feet, numb or weak hands or feet, or problems with bladder and bowels.
How to diagnose: People with spina bifida occulta of the cervical or thoracic vertebrae should be evaluated by a neurologist and or neurosurgeon who has experience with these spine abnormalities.

Pectus Excavatum or Carinatum: Pectus excavatum happens when the sternum (breastbone) and rib cage grow abnormally, causing sunken or caved in appearance of the chest. Pectus carinatum happens when the breastbone and rib cage develop in a manner that causes the chest to jut out like a bird. Many people with pectus excavatum or carinatum experience no other symptoms related to these conditions. Sometimes these conditions can cause breathing problems, heart issues and pain in the chest and back.
How to diagnose: These conditions are diagnosed by observation. If the noted symptoms develop, particularly in pectus excavatum, it is important to see medical advice.

Prognathism: Prognathism is the position of the upper jaw in relation to the lower jaw. In mandibular (lower jaw) prognathism, the lower jaw protrudes or sticks out further from the upper jaw (maxilla). Prognathism of the maxilla means the upper jaw protrudes out further than the lower. This problem can lead to misalignment of the upper and lower teeth.
How to diagnose: People concerned about this issue should be evaluated by their dentist, and possibly an orthodontist and oral surgeon.

Rib Abnormalities: People with Gorlin syndrome may have a variety of changes in the shape of their ribs. These include ribs that are splayed (turned outward), fused (stuck together), bifid (split in two at the end), and partially missing.
How to diagnose: Though possibly noted by observation or feel, rib abnormalities generally do not cause any symptoms and therefore do not require any treatment.

Short Fourth Metacarpal: The fourth metacarpal bone is the bone in the hand leading to the ring finger (the fourth finger counting from the thumb). Some people with Gorlin syndrome have a shorter metacarpal bone than average.

How to diagnose: This change in the length of the bone is noted with observation and does not cause any problems for the individual.

Synophrys: Often called a unibrow or monobrow, synophrys is a fusion of the two eyebrows above the bridge of the nose, creating a single eyebrow. Some people with a unibrow choose to wax, shave, or pluck the hairs in the middle to create the look of two eyebrows. Sometimes, the hair does not grow back after repeated removal.
How to diagnose: This change is noted with observation.

Strabismus: Crossed eyes or strabismus is when of one or both eyes turn inward. Strabismus may happen to a person sometimes (intermittent) or be present all the time (constant).
How to diagnose: This problem is noted when looking at the affected person. Individuals with strabismus should see an ophthalmologist for treatment.

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When discussing treatment options, it is important to recognize that the use of radiation therapy may induce the growth of multiple basal cell carcinomas in patients with Gorlin syndrome. The risks and benefits of each treatment option should be carefully considered by the child's entire care team.

Meningioma: Meningiomas are generally slow growing, benign (non-cancerous) brain tumors that come from the covering of the brain and spinal cord. They do not grow from the brain tissue itself.
How to diagnose: Meningioma is diagnosed by an MRI of the brain and typically managed by a neurosurgeon. Symptoms of meningiomas vary depending on the location of the tumor. Symptoms may include headache, weakness of an

arm or leg. Less common signs include personality changes, seizures, and vision problems.

Ocular Hypertelorism: Ocular hypertelorism is a wide distance between the eye sockets, resulting in "wide-set" appearing eyes. Occasionally, hypertelorism can cause visual problems, but usually this is not the case.
How to diagnose: Hypertelorism is diagnosed by observation and individuals with this condition may be evaluated by a pediatric ophthalmologist.

Lymphomesenteric Cysts: Lymphomesenteric cysts are benign (non-cancerous) tumors of the abdomen that can cause belly pain, and bowel blockage when large or widespread. These cysts are made up mostly of the fluid from the lymph system.
How to diagnose: These are generally diagnosed with an abdominal ultrasound or MRI.

Twenties through Adulthood

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