Gardner syndrome

Eponyms:
- APC
- FPC
- GS
- Oldfield syndrome
- polyposis adenomatous intestinal
- polyposis intestinal III

Inheritance: autosomal dominant

Semiological Synthesis: Cutaneous-gastrointestinal-ocular disorder. Multiple colorectal polyposis associated with various soft- and hard- tissue tumours including, osteomas, dermoids, fibromas; dental growth defects, retinal changes, liver cysts; occasionally Horner syndrome. High likelihood of colorectal cancer.

Group Sub group Signs:

DERMATOLOGICAL DISORDERS
cutis, nodules
- cutis, cysts
- cutis, nodules; skin polyps; warty, verrucous lesions
- lipoma

GASTROINTESTINAL DISORDERS
ileum and/or colon, anomalies
- intestinal polyposis
intestinal dysfunctions
- abdominal pain, abdominal colic
- intestinal perforation, intestinal hemorrhagy
liver dysfunctions, liver anomalies
- liver, cysts

LABORATORY DATA
chromosomal assignment
- chromosome 5q localization
chromosomal disorders
- chromosomal numerical and/or structural anomalies
gene, structural-functional anomalies
- APC (GS) (FPC) protein adenomatous polyposis coli, gene chr.5q21-q22
- gene analysis-DNA analysis
tissue, biochemical changes
- postglandin E2 high levels, PGE2, high levels

MUSCULAR DISEASES
systemic muscular defects
- desmoids, fibroblastic nodules
- leiomyoma

NEOPLASTIC DISEASES
cancer, genetic features
- tumour susceptibility

OCULAR DISORDERS
choroidoretinal defects
- choroidoretinal dystrophy
- retinal aplasia
- retinal pigment, congenital hypertrophy
- retinal pigmentation changes, retinal flecks

OROCRANIOFACIAL ANOMALIES
oral mucous membranes, changes
- phakomatosis, mesodermal
teeth, modified structures
- teeth, carious
- teeth, cysts
- teeth, late eruption
- teeth, supernumerary
### OTHERS

**inheritance**
- inheritance, autosomal dominant

**supergroups**
- gastrointestinal-oculo-skeletal disorders

### PRENATAL-NEONATAL MODIFIED DATA

**foetal changes**
- congenital tumour
- dermoid cysts, teratomas
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

**prenatal diagnosis**
- prenatal diagnosis, echographic
- prenatal diagnosis, molecular

### SKELETAL DISORDERS

**bones, lesions, structural changes**
- bone cysts, osteoma, osteochondroma, bones tumefactions

**ossification, changes**
- calcification ectopic not including: intracranial calcifications (calcinosis)

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**Super group:** gastrointestinal-oculo-skeletal disorders

**Super aggreg.**

**Aggregations:**
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

### NEOPLASTIC DISORDERS

**tumour, congenital**
**tumour, susceptibility**

### OTHER

- dermoid cysts, teratomas

### PHAKOMATOSIS, HAMARTOMATA

**phakomatosis, mesodermal**

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**Differential diagnosis:**

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<th>Condition</th>
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<td>Turcot syndrome</td>
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**Bibliography**

- OMIM ID: 175100
- Annuario Orphanet-Italia delle Malattie Rare, 2005 pag.671